



# GLB1 Variant Catalog

Comprehensive Database of *GLB1* Gene Variants · Cure GM1 Foundation

<b>1295</b> Total Variants	<b>329</b> Pathogenic / LP	<b>248</b> VUS	<b>54</b> Conflicting	<b>586</b> Benign / LB	<b>78</b> Not Classified	<b>24</b> PIN Observed
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■ 24 variants observed in PIN cohort are highlighted in amber — Cure GM1 AllStripes Patient Insights Network (2025 WORLD Symposium)

## Row Colour Legend

Pathogenic	Likely Pathogenic	P/LP	Conflicting	VUS	Likely Benign	Benign	Not Classified	Amber = PIN Obs.
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PMID (PubMed Identifier): a unique numeric ID assigned by the National Library of Medicine to each article indexed in PubMed (pubmed.ncbi.nlm.nih.gov). Click any PMID to view the source publication.

† PMIDs marked † were submitted to ClinVar by independent laboratories and may not have been published specifically in relation to *GLB1*. Retained as submitted; independent verification is advisable.

## About This Catalog

This programmatically curated catalog aggregates all publicly available *GLB1* variants from five major databases and six landmark peer-reviewed studies. It covers the *GLB1* gene (3p22.3; 16 exons; 62.5 kb), associated with GM1 gangliosidosis (Types I, II, and III) and MPS IVB (Morquio B disease), including the intermediate MBD Plus phenotype. Each variant is annotated with cDNA and protein changes, classification, exon/intron location, pathogenicity, gnomAD frequency, associated phenotypes, PubMed references, and source attribution.

## Classification Source Hierarchy

Pathogenicity labels are drawn from ClinVar's aggregate germline classification. When multiple ClinVar submitters report different interpretations for the same variant, ClinVar flags the entry as "Conflicting interpretations of pathogenicity"; this catalog inherits that label unchanged. For variants present in both ClinVar and non-ClinVar sources (LOVD, OMIM, NTSAD, or published literature), ClinVar's classification takes precedence. For variants absent from ClinVar entirely, classification is taken as reported by the source, or "Not Classified" where no interpretation was provided.

## Limitations:

- Sources are listed in the appendix. Variants from other databases and peer-reviewed publications where variant data are not submitted into the listed databases are not otherwise included.
- Phenotypes and PubMed IDs (PMIDs) are extracted from ClinVar submissions. Some entries may be incomplete or misdirected. Consult ClinVar directly (via ClinVar ID link) for the full current phenotype and PMID listing.
- Exon assignments are approximate. Nomenclature follows NM\_000404.4 (HGVS). This catalog reflects databases as of April 21, 2026.
- Variants are listed as reported. Sequencing methodologies were not included, therefore, the presence of additional, undetected variants cannot be excluded.

Have a correction? Email us at [info@curegm1.org](mailto:info@curegm1.org)

**Not a diagnostic tool.** The information provided is for informational purposes only and should not be used to diagnose or treat any health condition. While we strive to accurately provide data from the sources cited, we do not guarantee the completeness, accuracy, or reliability of the information and assume no responsibility for its use.

**Sources:** ClinVar · LOVD · gnomAD · NTSAD · OMIM · Santamaria 2006 · Bidchol 2015 · Hofer 2009 · Caciotti 2005 · Caciotti 2003 · Cure GM1 PIN · World Symposium Poster

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cDNA Change	Protein Change	Location	Pathogenicity	Classified	Phenotype	gnomAD AF	Sources	PMID(s)	PIN	ClinVar ID
c.-48G>C		5'UTR	Conflicting	Jan 13, 2018	GM1 gangliosidosis; Mucopolysaccharidosis; MPS-IV-B	<a href="#">0.00019</a>	ClinVar; gnomAD			<a href="#">902457</a>
c.-37C>G		5'UTR	Benign	Jan 13, 2018	GM1 gangliosidosis; Mucopolysaccharidosis; MPS-IV-B	<a href="#">0.00078</a>	ClinVar; gnomAD			<a href="#">344799</a>
c.-25C>G		5'UTR	Conflicting	Sep 22, 2025	GM1 gangliosidosis; Mucopolysaccharidosis; MPS-IV-B	<a href="#">0.00030</a>	ClinVar; gnomAD			<a href="#">344797</a>
c.-17C>T		5'UTR	VUS	Apr 27, 2017	Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis	<a href="#">2.06e-06</a>	ClinVar; gnomAD			<a href="#">900794</a>
c.-11G>A		5'UTR	Likely Benign	Oct 03, 2022		<a href="#">1.16e-05</a>	ClinVar; gnomAD			<a href="#">4684685</a>
c.1A>C	p.Met1Leu	Exon 1	Likely Pathogenic	Apr 30, 2015			ClinVar			<a href="#">193093</a>
c.2T>C		Exon 1	Not Classified				LOVD			
c.3G>A	p.Met1Ile	Exon 1	Likely Pathogenic	Mar 04, 2025	Infantile GM1 gangliosidosis; GLB1-related disorder	<a href="#">6.85e-07</a>	ClinVar; gnomAD; Bidchol 2015 (PMID:25936995)	<a href="#">25936995</a>		<a href="#">1895462</a>
c.4C>A	p.Pro2Thr	Exon 1	VUS	Sep 01, 2022	GM1 gangliosidosis; Mucopolysaccharidosis; MPS-IV-B; Inborn genetic diseases; MPS-IV-B;GM1 gangliosidosis	<a href="#">2.74e-05</a>	ClinVar; gnomAD			<a href="#">344796</a>
c.4C>G	p.Pro2Ala	Exon 1	VUS	Aug 10, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">1.31e-05</a>	ClinVar; gnomAD			<a href="#">1500439</a>
c.5C>G	p.Pro2Arg	Exon 1	Likely Benign	Oct 22, 2025	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">2.12e-05</a>	ClinVar; gnomAD			<a href="#">972610</a>
c.5C>T	p.Pro2Leu	Exon 1	VUS	Aug 22, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">8.22e-06</a>	ClinVar; gnomAD			<a href="#">2194800</a>
c.6G>A	p.Pro2=	Exon 1	Likely Benign	Dec 08, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">2932642</a>
c.8G>T		Exon 1	Not Classified			1.92e-05	LOVD; gnomAD			
c.9G>A	p.Gly3=	Exon 1	Likely Benign	Apr 25, 2022	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">1558724</a>
c.9G>C	p.Gly3=	Exon 1	Likely Benign	Oct 23, 2024	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">1.37e-06</a>	ClinVar; gnomAD			<a href="#">3751693</a>
c.12C>T	p.Phe4=	Exon 1	Likely Benign	Sep 27, 2023	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">2.74e-06</a>	ClinVar; gnomAD			<a href="#">2940136</a>
c.15G>T	p.Leu5=	Exon 1	Likely Benign	Aug 08, 2021	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">1552945</a>
c.20G>C	p.Arg7Pro	Exon 1	VUS	Oct 13, 2022	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">1.37e-06</a>	ClinVar; gnomAD			<a href="#">1363728</a>
c.27C>G	p.Leu9=	Exon 1	Likely Benign	Jun 27, 2024	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">1984037</a>
c.27C>T	p.Leu9=	Exon 1	Likely Benign	Feb 25, 2025	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">6.85e-07</a>	ClinVar; gnomAD			<a href="#">1142155</a>
c.29C>T	p.Pro10Leu	Exon 1	Benign	Feb 04, 2026	GM1 gangliosidosis; Mucopolysaccharidosis; MPS-IV-B; MPS-IV-B;GM1 gangliosidosis; Infantile GM1 gangliosidosis; GM1 gangliosidosis type 2; GM1 gangliosidosis type 3	<a href="#">0.59800</a>	ClinVar; LOVD; gnomAD; Santamaria 2006 (PMID:16941474)	<a href="#">16941474</a>		<a href="#">92904</a>

cDNA Change	Protein Change	Location	Pathogenicity	Classified	Phenotype	gnomAD AF	Sources	PMID(s)	PIN	ClinVar ID
c.29dup	p.Leu11fs	Exon 1	Likely Pathogenic	Jan 24, 2024	GM1 gangliosidosis type 3;Infantile GM1 gangliosidosis;GM1 gangliosidosis type 2;Mucopolysaccharidosis; MPS-IV-B	<a href="#">6.57e-06</a>	ClinVar; gnomAD			<a href="#">3589144</a>
c.31C>G	p.Leu11Val	Exon 1	Conflicting	Jan 27, 2026	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis; Infantile GM1 gangliosidosis; GLB1-related disorder	<a href="#">0.00013</a>	ClinVar; LOVD; gnomAD			<a href="#">425289</a>
c.31C>T	p.Leu11=	Exon 1	Likely Benign	Apr 17, 2025	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">6.57e-06</a>	ClinVar; gnomAD			<a href="#">1119074</a>
c.33G>C	p.Leu11=	Exon 1	Likely Benign	Jun 15, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">2.74e-06</a>	ClinVar; gnomAD			<a href="#">2938204</a>
c.33_34delinsCTGC	p.Leu12fs	Exon 1	Likely Pathogenic	Oct 16, 2024	Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">4818319</a>
c.34T>C	p.Leu12=	Exon 1	Benign	Feb 04, 2026	GM1 gangliosidosis; Mucopolysaccharidosis; MPS-IV-B; MPS-IV-B;GM1 gangliosidosis; Infantile GM1 gangliosidosis; GM1 gangliosidosis type 2; GM1 gangliosidosis type 3	<a href="#">0.88900</a>	ClinVar; LOVD; gnomAD			<a href="#">92905</a>
c.37C>T	p.Leu13=	Exon 1	Likely Benign	Jul 15, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">2017467</a>
c.39G>A	p.Leu13=	Exon 1	Likely Benign	Mar 12, 2024	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">3755227</a>
c.40G>C	p.Val14Leu	Exon 1	VUS	Aug 03, 2025	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis; Inborn genetic diseases	<a href="#">1.10e-05</a>	ClinVar; gnomAD			<a href="#">1496444</a>
c.42T>C	p.Val14=	Exon 1	Likely Benign	Mar 14, 2019	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">6.85e-07</a>	ClinVar; gnomAD			<a href="#">1105356</a>
c.45G>C	p.Leu15=	Exon 1	Likely Benign	Feb 08, 2023	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">2944052</a>
c.47del	p.Leu16fs	Exon 1	Likely Pathogenic	May 07, 2024	GM1 gangliosidosis type 3;Infantile GM1 gangliosidosis;GM1 gangliosidosis type 2;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">3589143</a>
c.51dup	p.Leu18fs	Exon 1	Pathogenic	Oct 03, 2021	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">6.85e-07</a>	ClinVar; gnomAD	<a href="#">18524657</a> ; <a href="#">23757202</a> ; <a href="#">26646981</a>		<a href="#">92911</a>
c.61A>G	p.Thr21Ala	Exon 1	Conflicting	Jan 19, 2026	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis; Inborn genetic diseases	<a href="#">6.03e-05</a>	ClinVar; gnomAD			<a href="#">1109494</a>
c.62C>T	p.Thr21Met	Exon 1	VUS	Oct 07, 2024	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis; Inborn genetic diseases	<a href="#">7.53e-06</a>	ClinVar; gnomAD			<a href="#">2037639</a>
c.63G>A	p.Thr21=	Exon 1	Likely Benign	Jul 19, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">1637447</a>
c.65_75+1del		Exon 1	P/LP	Dec 07, 2023	Infantile GM1 gangliosidosis;GM1 gangliosidosis type 2;Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis type 3; GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B; Infantile GM1 gangliosidosis	<a href="#">2.06e-06</a>	ClinVar; gnomAD	<a href="#">16199547</a> ; <a href="#">18524657</a> ; <a href="#">36265282</a>		<a href="#">555329</a>

cDNA Change	Protein Change	Location	Pathogenicity	Classified	Phenotype	gnomAD AF	Sources	PMID(s)	PIN	ClinVar ID
c.66C>T	p.Arg22=	Exon 1	Likely Benign	Apr 14, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">3.42e-06</a>	ClinVar; gnomAD			<a href="#">1124539</a>
c.67G>A	p.Gly23Ser	Exon 1	VUS	Jul 11, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">6.85e-07</a>	ClinVar; gnomAD			<a href="#">2185797</a>
c.68G>A	p.Gly23Asp	Exon 1	VUS	Jun 01, 2025	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">1.78e-05</a>	ClinVar; gnomAD			<a href="#">2084657</a>
c.69C>A	p.Gly23=	Exon 1	Likely Benign	May 16, 2025	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">4.11e-06</a>	ClinVar; gnomAD			<a href="#">1414552</a>
c.69C>G	p.Gly23=	Exon 1	Likely Benign	Feb 03, 2024	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">1.92e-05</a>	ClinVar; gnomAD			<a href="#">795413</a>
c.69_75+1del		Exon 1	Pathogenic	May 28, 2022	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar	<a href="#">18524657</a>		<a href="#">2000083</a>
c.70T>C	p.Leu24=	Exon 1	Likely Benign	Mar 18, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">6.85e-07</a>	ClinVar; gnomAD			<a href="#">2934776</a>
c.70_73del	p.Leu24fs	Exon 1	Pathogenic	Sep 23, 2025	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar	<a href="#">18524657</a>		<a href="#">4792851</a>
c.75+1G>C		Intron 1	P/LP	Oct 24, 2025	Mucopolysaccharidosis; MPS-IV-B; Inborn genetic diseases; MPS-IV-B;GM1 gangliosidosis	<a href="#">6.17e-06</a>	ClinVar; gnomAD	<a href="#">16199547</a> ; <a href="#">18524657</a> ; <a href="#">38313286</a>		<a href="#">92913</a>
c.75+2T>G		Intron 1	Pathogenic	May 12, 2021	Mucopolysaccharidosis; MPS-IV-B		ClinVar	<a href="#">33558080</a>		<a href="#">1077168</a>
c.75+2dup		Intron 1	Pathogenic	Dec 29, 2025	Infantile GM1 gangliosidosis; Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis; MPS-IV-B; GM1 gangliosidosis type 3; GM1 gangliosidosis type 2; Inborn genetic diseases	<a href="#">0.00015</a>	ClinVar; LOVD; gnomAD	<a href="#">17576681</a> ; <a href="#">21497194</a> ; <a href="#">29160035</a> ; <a href="#">33038107</a> ; <a href="#">33737400</a> ; <a href="#">8198123</a> ; <a href="#">8199591</a> ; <a href="#">9536098</a>		<a href="#">936</a>
c.75+3A>G		Intron 1	VUS	Apr 26, 2024	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">3.56e-05</a>	ClinVar; gnomAD	<a href="#">17576681</a> ; <a href="#">9536098</a>		<a href="#">235596</a>
c.75+3_75+4del		Intron 1	VUS	Dec 24, 2022	Infantile GM1 gangliosidosis		ClinVar			<a href="#">1878428</a>
c.75+4dup		Intron 1	Pathogenic	-	Infantile GM1 gangliosidosis		ClinVar			<a href="#">1332869</a>
c.75+5G>C		Intron 1	Not Classified	-	-		ClinVar			<a href="#">264674</a>
c.75+7C>G		Intron 1	Likely Benign	Apr 07, 2025	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">4.11e-06</a>	ClinVar; gnomAD			<a href="#">1555641</a>
c.75+8del		Intron 1	Likely Benign	Jun 24, 2024	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">6.85e-07</a>	ClinVar; gnomAD			<a href="#">2021523</a>
c.75+9G>A		Intron 1	Likely Benign	May 08, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">6.85e-07</a>	ClinVar; gnomAD			<a href="#">1568849</a>
c.75+10C>T		Intron 1	Likely Benign	May 15, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">1.37e-06</a>	ClinVar; gnomAD			<a href="#">1110965</a>
c.75+16C>A		Intron 1	Likely Benign	Mar 02, 2024	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">6.86e-07</a>	ClinVar; gnomAD			<a href="#">3752548</a>
c.75+16C>T		Intron 1	Likely Benign	Apr 13, 2023	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">2946678</a>
c.75+17C>G		Intron 1	Likely Benign	Jan 16, 2026	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">4794458</a>
c.75+19G>A		Intron 1	Likely Benign	May 12, 2023	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">6.86e-07</a>	ClinVar; gnomAD			<a href="#">2940661</a>

cDNA Change	Protein Change	Location	Pathogenicity	Classified	Phenotype	gnomAD AF	Sources	PMID(s)	PIN	ClinVar ID
c.75+20G>A		Intron 1	Likely Benign	May 22, 2023	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">2948082</a>
c.75+90G>T		Intron 1	Likely Benign	Nov 13, 2019			ClinVar			<a href="#">1191383</a>
c.76-5388A>G		Intron 1	Not Classified				LOVD			
c.76-5137dup		Intron 1	Not Classified				LOVD			
c.76-4971dup		Intron 1	Not Classified				LOVD			
c.76-4841T>A		Intron 1	Not Classified				LOVD			
c.76-4746T>C		Intron 1	Benign	May 13, 2021			ClinVar			<a href="#">1221622</a>
c.76-4537T>C		Intron 1	Not Classified				LOVD			
c.76-4532G>T		Intron 1	Not Classified			0.00020	LOVD; gnomAD			
c.76-4520G>A		Intron 1	Benign	Nov 06, 2019	GLB1-related disorder		ClinVar			<a href="#">3050297</a>
c.76-4483G>A		Intron 1	Benign	Jun 01, 2025	GLB1-related disorder		ClinVar			<a href="#">2653649</a>
c.76-4450T>C		Intron 1	Likely Benign	Oct 01, 2025			ClinVar			<a href="#">4534950</a>
c.76-4438_76-4437insAAAGAGGTA		Intron 1	Not Classified				LOVD			
c.76-4436_76-4435insAA		Intron 1	Not Classified				LOVD			
c.76-4429G>A		Intron 1	Likely Benign	Sep 01, 2022	GLB1-related disorder		ClinVar			<a href="#">2653648</a>
c.76-4426C>T		Intron 1	Benign	Jun 15, 2021	Mucopolysaccharidosis; MPS-IV-B; Infantile GM1 gangliosidosis		ClinVar; LOVD			<a href="#">1174058</a>
c.76-4414C>T		Intron 1	Likely Benign	Apr 08, 2019	GLB1-related disorder		ClinVar			<a href="#">3036381</a>
c.76-4399_76-4397del		Intron 1	VUS	Apr 04, 2024	Infantile GM1 gangliosidosis		ClinVar			<a href="#">3068200</a>
c.76-4391G>A		Intron 1	Not Classified				LOVD			
c.76-4376G>A		Intron 1	Likely Benign	Jul 01, 2025	GLB1-related disorder	<a href="#">0.00028</a>	ClinVar; gnomAD			<a href="#">3039574</a>
c.76-288A>G		Intron 1	Likely Benign	Sep 06, 2018			ClinVar			<a href="#">1200095</a>
c.76-17C>T		Intron 1	Likely Benign	Apr 08, 2024	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">2109636</a>
c.76-16del		Intron 1	Benign	Dec 19, 2025	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">2.74e-06</a>	ClinVar; gnomAD			<a href="#">2948603</a>
c.76-12C>A		Intron 1	Likely Benign	Jan 01, 2024	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">2938172</a>
c.76-9G>A		Intron 1	Likely Benign	Oct 03, 2020	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">1103897</a>
c.76-9G>T		Intron 1	Likely Benign	May 11, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">6.84e-07</a>	ClinVar; gnomAD			<a href="#">2061639</a>
c.76-9_76-8delinsAA		Intron 1	Likely Benign	Nov 08, 2021	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">1581024</a>
c.76-5T>C		Intron 1	Likely Benign	Aug 19, 2022	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">2.05e-06</a>	ClinVar; gnomAD			<a href="#">2165427</a>
c.76-4G>A		Intron 1	Likely Benign	Jul 01, 2020	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">1091619</a>
c.76-1G>T		Intron 1	Likely Pathogenic	Apr 17, 2023	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">7.12e-07</a>	ClinVar; gnomAD	<a href="#">16199547</a> ; <a href="#">18524657</a>		<a href="#">2946862</a>
c.77A>G	p.Asn26Ser	Exon 2	VUS	Mar 13, 2025	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">2.05e-06</a>	ClinVar; gnomAD			<a href="#">3761955</a>

cDNA Change	Protein Change	Location	Pathogenicity	Classified	Phenotype	gnomAD AF	Sources	PMID(s)	PIN	ClinVar ID
c.78T>C	p.Asn26=	Exon 2	Likely Benign	Dec 04, 2020	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">6.84e-07</a>	ClinVar; gnomAD			<a href="#">1658795</a>
c.81C>T	p.Ala27=	Exon 2	Likely Benign	Aug 21, 2022	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">6.57e-06</a>	ClinVar; gnomAD			<a href="#">2174765</a>
c.84C>T	p.Thr28=	Exon 2	Likely Benign	Oct 05, 2021	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">1607646</a>
c.87G>T	p.Gln29His	Exon 2	VUS	Sep 13, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">2067152</a>
c.93G>A	p.Met31Ile	Exon 2	VUS	Mar 28, 2017	GM1 gangliosidosis type 3;Mucopolysaccharidosis; MPS-IV-B;Infantile GM1 gangliosidosis;GM1 gangliosidosis type 2		ClinVar			<a href="#">551262</a>
c.101T>C	p.Ile34Thr	Exon 2	VUS	May 23, 2024		<a href="#">6.84e-07</a>	ClinVar; gnomAD	<a href="#">30675867</a>		<a href="#">3336417</a>
c.102T>C	p.Ile34=	Exon 2	Likely Benign	Jan 18, 2026	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">4783819</a>
c.105C>T	p.Asp35=	Exon 2	Likely Benign	Jun 20, 2019	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">1080660</a>
c.107A>G	p.Tyr36Cys	Exon 2	P/LP	Jan 28, 2026	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B; Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis type 3;Infantile GM1 gangliosidosis;GM1 gangliosidosis type 2	<a href="#">1.09e-05</a>	ClinVar; gnomAD	<a href="#">23046582</a> ; <a href="#">26646981</a> ; <a href="#">36233161</a> ; <a href="#">38313286</a>		<a href="#">1063917</a>
c.112C>A	p.Arg38=	Exon 2	Likely Benign	Feb 27, 2024	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">7.38e-07</a>	ClinVar; gnomAD			<a href="#">3754984</a>
c.113G>A	p.Arg38Gln	Exon 2	VUS	Aug 06, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">3.01e-05</a>	ClinVar; gnomAD			<a href="#">2072119</a>
c.114G>A	p.Arg38=	Exon 2	Likely Benign	Oct 11, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">1.37e-06</a>	ClinVar; gnomAD			<a href="#">2937455</a>
c.119C>T	p.Ser40Phe	Exon 2	VUS	Feb 20, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">2.23e-06</a>	ClinVar; gnomAD			<a href="#">2421838</a>
c.123C>T	p.Phe41=	Exon 2	Likely Benign	Nov 22, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">4.10e-06</a>	ClinVar; gnomAD			<a href="#">1160930</a>
c.126C>A	p.Leu42=	Exon 2	Likely Benign	Jul 26, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">4.79e-06</a>	ClinVar; gnomAD			<a href="#">2201713</a>
c.126C>G	p.Leu42=	Exon 2	Likely Benign	Jul 22, 2021	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">1529799</a>
c.126C>T	p.Leu42=	Exon 2	Likely Benign	Mar 28, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">2416103</a>
c.128A>G	p.Lys43Arg	Exon 2	VUS	Feb 11, 2025	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">7.52e-07</a>	ClinVar; LOVD; gnomAD			<a href="#">4788536</a>
c.130G>T	p.Asp44Tyr	Exon 2	Likely Pathogenic	May 06, 2021	Infantile GM1 gangliosidosis		ClinVar			<a href="#">2500787</a>
c.132T>C	p.Asp44=	Exon 2	Likely Benign	Aug 11, 2025	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">1.37e-06</a>	ClinVar; gnomAD			<a href="#">1120335</a>
c.136C>T	p.Gln46Ter	Exon 2	Likely Pathogenic	Apr 19, 2024	Mucopolysaccharidosis; MPS-IV-B	<a href="#">7.60e-07</a>	ClinVar; gnomAD			<a href="#">4818312</a>
c.137_138delinsCC	p.Gln46Pro	Exon 2	VUS	Feb 02, 2023	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">2943875</a>
c.138G>A	p.Gln46=	Exon 2	Likely Benign	Jun 13, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">1568237</a>

cDNA Change	Protein Change	Location	Pathogenicity	Classified	Phenotype	gnomAD AF	Sources	PMID(s)	PIN	ClinVar ID
c.141A>G	p.Pro47=	Exon 2	Likely Benign	Aug 28, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">2.74e-06</a>	ClinVar; gnomAD			<a href="#">1628978</a>
c.145C>T	p.Arg49Cys	Exon 2	Conflicting	Aug 13, 2024	Infantile GM1 gangliosidosis; GM1 gangliosidosis type 3;Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis type 2;Infantile GM1 gangliosidosis; Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis; MPS-IV-B	<a href="#">9.58e-06</a>	ClinVar; LOVD; gnomAD; Bidchol 2015 (PMID:25936995)	<a href="#">15365997</a> ; <a href="#">15986423</a> ; <a href="#">1909089</a> ; <a href="#">21520340</a> ; <a href="#">22128166</a> ; <a href="#">23337983</a> ; <a href="#">25936995</a> ; <a href="#">26646981</a> ; <a href="#">30138938</a> ; <a href="#">30267299</a> ; <a href="#">30408610</a>		<a href="#">923</a>
c.146G>A	p.Arg49His	Exon 2	P/LP	Aug 27, 2025	Inborn genetic diseases; GM1 gangliosidosis type 2;GM1 gangliosidosis type 3;Mucopolysaccharidosis; MPS-IV-B;Infantile GM1 gangliosidosis; Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis; MPS-IV-B	<a href="#">4.79e-06</a>	ClinVar; LOVD; gnomAD	<a href="#">15986423</a> ; <a href="#">20175788</a> ; <a href="#">30267299</a>		<a href="#">521991</a>
c.146G>C	p.Arg49Pro	Exon 2	Likely Pathogenic	May 13, 2024	GM1 gangliosidosis; Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis type 3;GM1 gangliosidosis type 2;Infantile GM1 gangliosidosis	<a href="#">3.82e-06</a>	ClinVar; gnomAD			<a href="#">3251806</a>
c.148T>C	p.Tyr50His	Exon 2	Likely Pathogenic	Aug 24, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar	<a href="#">29439846</a> ; <a href="#">30267299</a>		<a href="#">2937299</a>
c.149A>G	p.Tyr50Cys	Exon 2	P/LP	Nov 10, 2025	Infantile GM1 gangliosidosis; GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis; Mucopolysaccharidosis	<a href="#">6.84e-07</a>	ClinVar; LOVD; gnomAD	<a href="#">29439846</a> ; <a href="#">30267299</a> ; <a href="#">37381921</a> ; <a href="#">37597066</a>		<a href="#">917831</a>
c.150C>T	p.Tyr50=	Exon 2	Likely Benign	Feb 19, 2025	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">1141627</a>
c.151A>C	p.Ile51Leu	Exon 2	Likely Pathogenic	Mar 01, 2023	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar	<a href="#">16617000</a> ; <a href="#">1907800</a>		<a href="#">2942831</a>
c.152T>A	p.Ile51Asn	Exon 2	P/LP	Jul 09, 2025	Infantile GM1 gangliosidosis;GM1 gangliosidosis type 2;Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis type 3; GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis type 2		ClinVar	<a href="#">16617000</a> ; <a href="#">1907800</a> ; <a href="#">21497194</a> ; <a href="#">28577204</a>		<a href="#">554550</a>
c.152T>C	p.Ile51Thr	Exon 2	Pathogenic	Jan 22, 2026	GM1 gangliosidosis type 3; GM1 gangliosidosis type 3;Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis type 2;Infantile GM1 gangliosidosis; Infantile GM1 gangliosidosis; Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis; MPS-IV-B	<a href="#">3.42e-06</a>	ClinVar; LOVD; gnomAD	<a href="#">1353343</a> ; <a href="#">16617000</a> ; <a href="#">1907800</a> ; <a href="#">1909089</a> ; <a href="#">19644515</a> ; <a href="#">21520340</a> ; <a href="#">30267299</a> ; <a href="#">8112731</a>		<a href="#">926</a>
c.159A>C	p.Gly53=	Exon 2	Likely Benign	Dec 22, 2025	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">4791182</a>
c.160A>G	p.Ser54Gly	Exon 2	Likely Pathogenic	Mar 17, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar	<a href="#">16314480</a>		<a href="#">2930972</a>
c.161G>A	p.Ser54Asn	Exon 2	Likely Pathogenic	Dec 18, 2024	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis	<a href="#">1.37e-06</a>	ClinVar; gnomAD	<a href="#">16314480</a> ; <a href="#">21520340</a>		<a href="#">1514446</a>
c.161G>T		Exon 2	Not Classified			<a href="#">6.84e-07</a>	LOVD; gnomAD			

cDNA Change	Protein Change	Location	Pathogenicity	Classified	Phenotype	gnomAD AF	Sources	PMID(s)	PIN	ClinVar ID
c.163del	p.Ile55fs	Exon 2	Pathogenic	Aug 15, 2018	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">2.05e-06</a>	ClinVar; gnomAD	<a href="#">18524657</a> ; <a href="#">20175788</a>		<a href="#">657106</a>
c.165T>C	p.Ile55=	Exon 2	Likely Benign	Aug 13, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">6.84e-07</a>	ClinVar; gnomAD			<a href="#">2023994</a>
c.168C>G	p.His56Gln	Exon 2	VUS	Aug 16, 2022	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">1387949</a>
c.169del	p.Tyr57fs	Exon 2	Pathogenic	Jan 28, 2015			ClinVar			<a href="#">195073</a>
c.171C>G	p.Tyr57Ter	Exon 2	Pathogenic	Sep 10, 2025	GM1 gangliosidosis type 2;Infantile GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis type 3; Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis; GM1 gangliosidosis; Infantile GM1 gangliosidosis		ClinVar	<a href="#">15365997</a> ; <a href="#">16617000</a> ; <a href="#">18524657</a> ; <a href="#">25525159</a>		<a href="#">92900</a>
c.173C>T	p.Ser58Phe	Exon 2	VUS	Jul 25, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">1429163</a>
c.174C>G	p.Ser58=	Exon 2	Likely Benign	Jan 09, 2024	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">6.84e-07</a>	ClinVar; gnomAD			<a href="#">2929054</a>
c.175C>T	p.Arg59Cys	Exon 2	Pathogenic	Feb 13, 2025	GM1 gangliosidosis type 3;Mucopolysaccharidosis; MPS-IV-B;Infantile GM1 gangliosidosis;GM1 gangliosidosis type 2; GM1 gangliosidosis type 2; Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis; Infantile GM1 gangliosidosis; GM1 gangliosidosis	<a href="#">2.26e-05</a>	ClinVar; LOVD; gnomAD; Caciotti 2005 (PMID:15714521); Santamaria 2006 (PMID:16941474)	<a href="#">10338095</a> ; <a href="#">15714521</a> ; <a href="#">16314480</a> ; <a href="#">16941474</a> ; <a href="#">17309651</a> ; <a href="#">17664528</a> ; <a href="#">21520340</a>		<a href="#">496895</a>
c.176G>A	p.Arg59His	Exon 2	P/LP	Nov 16, 2025	Infantile GM1 gangliosidosis; GM1-gangliosidosis; type I; with cardiac involvement; GM1 gangliosidosis type 2; Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis; MPS-IV-B;GM1 gangliosidosis; GM1 gangliosidosis type 3;Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis type 2;Infantile GM1 gangliosidosis; GM1 gangliosidosis type 3; Inborn genetic diseases; GLB1-related disorder	<a href="#">2.80e-05</a>	ClinVar; LOVD; gnomAD; OMIM; NTSAD; Caciotti 2005 (PMID:15714521); Santamaria 2006 (PMID:16941474)	<a href="#">10338095</a> ; <a href="#">10737981</a> ; <a href="#">15714521</a> ; <a href="#">15906092</a> ; <a href="#">16941474</a> ; <a href="#">17309651</a> ; <a href="#">17664528</a> ; <a href="#">21214877</a> ; <a href="#">21520340</a> ; <a href="#">21637542</a> ; <a href="#">23046582</a> ; <a href="#">26337817</a> ; <a href="#">28939701</a> ; <a href="#">31216405</a> ; <a href="#">31497487</a> ; <a href="#">31761138</a> ; <a href="#">31776384</a> ; <a href="#">32036093</a> ; <a href="#">9781688</a>		<a href="#">945</a>
c.176G>T	p.Arg59Leu	Exon 2	Conflicting	Sep 04, 2025	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar; Caciotti 2005 (PMID:15714521); Santamaria 2006 (PMID:16941474)	<a href="#">15714521</a> ; <a href="#">16941474</a> ; <a href="#">17309651</a>		<a href="#">1497708</a>
c.184C>T	p.Arg62Cys	Exon 2	VUS	Jul 15, 2025	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis; Infantile GM1 gangliosidosis;GM1 gangliosidosis type 2;Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis type 3	<a href="#">2.87e-05</a>	ClinVar; gnomAD			<a href="#">2040930</a>
c.188T>A	p.Phe63Tyr	Exon 2	VUS	Nov 07, 2025	Infantile GM1 gangliosidosis;GM1 gangliosidosis type 2;GM1 gangliosidosis type 3;Mucopolysaccharidosis; MPS-IV-B	<a href="#">6.57e-06</a>	ClinVar; gnomAD	<a href="#">23046582</a>		<a href="#">558551</a>

cDNA Change	Protein Change	Location	Pathogenicity	Classified	Phenotype	gnomAD AF	Sources	PMID(s)	PIN	ClinVar ID
c.189C>G	p.Phe63Leu	Exon 2	VUS	Oct 25, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">6.84e-07</a>	ClinVar; gnomAD			<a href="#">1926010</a>
c.191A>G	p.Tyr64Cys	Exon 2	Conflicting	Dec 08, 2025	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis; MPS-IV-B; Inborn genetic diseases; GM1 gangliosidosis type 3;Mucopolysaccharidosis; MPS-IV-B;Infantile GM1 gangliosidosis;GM1 gangliosidosis type 2	<a href="#">8.89e-06</a>	ClinVar; gnomAD; Cure GM1 AllStripes PIN	<a href="#">29396176</a> ; <a href="#">31497487</a> ; <a href="#">34258138</a> ; <a href="#">40069543</a>	1	<a href="#">958153</a>
c.192C>T	p.Tyr64=	Exon 2	Likely Benign	Oct 23, 2021	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">1612752</a>
c.202C>G	p.Arg68Gly	Exon 2	Likely Pathogenic	Oct 20, 2020	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">1.57e-05</a>	ClinVar; gnomAD; Caciotti 2003 (PMID:12644936); Bidchol 2015 (PMID:25936995)	<a href="#">12644936</a> ; <a href="#">25936995</a>		<a href="#">1068202</a>
c.202C>T	p.Arg68Trp	Exon 2	Pathogenic	Mar 10, 2025	GM1 gangliosidosis type 2; Infantile GM1 gangliosidosis; Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis; Infantile GM1 gangliosidosis;GM1 gangliosidosis type 3;Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis type 2; GM1 gangliosidosis type 3; MPS-IV-B	<a href="#">2.46e-05</a>	ClinVar; gnomAD; Caciotti 2003 (PMID:12644936); Hofer 2009 (PMID:19472408); Bidchol 2015 (PMID:25936995)	<a href="#">12644936</a> ; <a href="#">19472408</a> ; <a href="#">25936995</a> ; <a href="#">29439846</a> ; <a href="#">31761138</a> ; <a href="#">33737400</a>		<a href="#">944</a>
c.203G>A	p.Arg68Gln	Exon 2	P/LP	Oct 17, 2025	GM1 gangliosidosis type 2;Mucopolysaccharidosis; MPS-IV-B;Infantile GM1 gangliosidosis;GM1 gangliosidosis type 3; GM1 gangliosidosis; Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis; MPS-IV-B	<a href="#">5.47e-06</a>	ClinVar; LOVD; gnomAD; Caciotti 2003 (PMID:12644936); Hofer 2009 (PMID:19472408); Bidchol 2015 (PMID:25936995)	<a href="#">12644936</a> ; <a href="#">19472408</a> ; <a href="#">22128166</a> ; <a href="#">25936995</a> ; <a href="#">29439846</a>		<a href="#">195074</a>
c.203G>C	p.Arg68Pro	Exon 2	Likely Pathogenic	Feb 20, 2025			ClinVar			<a href="#">4081424</a>
c.203G>T	p.Arg68Leu	Exon 2	Conflicting	Dec 22, 2025	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar; Caciotti 2003 (PMID:12644936); Bidchol 2015 (PMID:25936995)	<a href="#">12644936</a> ; <a href="#">25936995</a>		<a href="#">1482642</a>
c.204G>T	p.Arg68=	Exon 2	Likely Benign	Jul 19, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">6.84e-07</a>	ClinVar; gnomAD			<a href="#">755403</a>
c.206T>C	p.Leu69Pro	Exon 2	Conflicting	Apr 18, 2025	Mucopolysaccharidosis; MPS-IV-B		ClinVar; LOVD	<a href="#">21497194</a>		<a href="#">2432139</a>
c.209T>C	p.Leu70Pro	Exon 2	VUS	Aug 21, 2022	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">2131662</a>
c.210G>A	p.Leu70=	Exon 2	Likely Benign	Jan 11, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">2.74e-06</a>	ClinVar; gnomAD			<a href="#">2925615</a>
c.216G>A		Exon 2	Not Classified			8.01e-07	LOVD; gnomAD			
c.218A>G	p.Lys73Arg	Exon 2	VUS	Jun 03, 2024	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis type 3	<a href="#">5.47e-06</a>	ClinVar; LOVD; gnomAD	<a href="#">15986423</a>		<a href="#">1005888</a>
c.228G>A	p.Gly76=	Exon 2	Likely Benign	Jul 16, 2023	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">2107729</a>

cDNA Change	Protein Change	Location	Pathogenicity	Classified	Phenotype	gnomAD AF	Sources	PMID(s)	PIN	ClinVar ID
c.228G>C	p.Gly76=	Exon 2	Likely Benign	May 22, 2023	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">1592850</a>
c.229C>T	p.Leu77=	Exon 2	Likely Benign	Mar 03, 2024	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">1.37e-06</a>	ClinVar; gnomAD			<a href="#">1091207</a>
c.230_231del	p.Leu77fs	Exon 2	Likely Pathogenic	Mar 31, 2022	Infantile GM1 gangliosidosis;GM1 gangliosidosis type 2;GM1 gangliosidosis type 3		ClinVar			<a href="#">1725712</a>
c.234C>T	p.Asn78=	Exon 2	Conflicting	Jan 11, 2026	GM1 gangliosidosis; Mucopolysaccharidosis; MPS-IV-B; MPS-IV-B;GM1 gangliosidosis	<a href="#">8.62e-05</a>	ClinVar; gnomAD			<a href="#">344795</a>
c.235G>A	p.Ala79Thr	Exon 2	VUS	Jul 19, 2025	GM1 gangliosidosis type 3; GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">4.31e-05</a>	ClinVar; LOVD; gnomAD			<a href="#">1029824</a>
c.244_245+2delCGGT	p.Arg82fs	Exon 2	Pathogenic		GM1 gangliosidosis type 2		Caciotti 2003 (PMID:12644936)	<a href="#">12644936</a>		
c.245C>T	p.Thr82Met	Exon 2	P/LP	Jan 31, 2026	GM1 gangliosidosis type 3; Inborn genetic diseases; Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis; GM1 gangliosidosis type 3;Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis type 2;Infantile GM1 gangliosidosis; Infantile GM1 gangliosidosis; MPS-IV-B	<a href="#">1.51e-05</a>	ClinVar; LOVD; gnomAD; Hofer 2009 (PMID:19472408)	<a href="#">11511921</a> ; <a href="#">19472408</a> ; <a href="#">20175788</a> ; <a href="#">21520340</a> ; <a href="#">25326637</a> ; <a href="#">6791574</a> ; <a href="#">8198123</a>		<a href="#">935</a>
c.245+1G>A		Intron 2	Pathogenic	Sep 15, 2025	Infantile GM1 gangliosidosis;GM1 gangliosidosis type 2;Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis type 3; GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis; Infantile GM1 gangliosidosis; Colon adenocarcinoma; Mucopolysaccharidosis	<a href="#">7.53e-06</a>	ClinVar; LOVD; gnomAD; Santamaria 2006 (PMID:16941474)	<a href="#">15365997</a> ; <a href="#">16941474</a> ; <a href="#">17221873</a> ; <a href="#">20175788</a>		<a href="#">417873</a>
c.245+1G>C		Intron 2	Pathogenic	Dec 25, 2021	Infantile GM1 gangliosidosis;GM1 gangliosidosis type 2;Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis type 3; GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">2.05e-06</a>	ClinVar; gnomAD; Santamaria 2006 (PMID:16941474)	<a href="#">15365997</a> ; <a href="#">16199547</a> ; <a href="#">16941474</a> ; <a href="#">17221873</a> ; <a href="#">18524657</a>		<a href="#">554728</a>
c.245+3A>G		Intron 2	VUS	Jun 02, 2022	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">6.57e-06</a>	ClinVar; gnomAD	<a href="#">17576681</a> ; <a href="#">9536098</a>		<a href="#">2117302</a>
c.245+6T>C		Intron 2	VUS	Jan 19, 2026	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">1.37e-06</a>	ClinVar; gnomAD	<a href="#">17576681</a> ; <a href="#">9536098</a>		<a href="#">2127795</a>
c.245+10A>T		Intron 2	Likely Benign	Nov 17, 2023	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">2953954</a>
c.245+13G>A		Intron 2	Likely Benign	Sep 10, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">6.57e-06</a>	ClinVar; gnomAD			<a href="#">2937720</a>
c.245+16T>C		Intron 2	Likely Benign	Jan 27, 2024	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">1.97e-05</a>	ClinVar; gnomAD			<a href="#">2927153</a>
c.245+19G>A		Intron 2	Likely Benign	Sep 10, 2025	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">4.11e-06</a>	ClinVar; gnomAD			<a href="#">2943642</a>
c.245+19G>T		Intron 2	Likely Benign	Apr 22, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">2938317</a>
c.245+44A>C		Intron 2	Likely Benign	Jan 29, 2019		<a href="#">0.00340</a>	ClinVar; gnomAD			<a href="#">1193352</a>

cDNA Change	Protein Change	Location	Pathogenicity	Classified	Phenotype	gnomAD AF	Sources	PMID(s)	PIN	ClinVar ID
c.245+141A>T		Intron 2	Likely Benign	Apr 21, 2019			ClinVar			<a href="#">1204232</a>
c.245+244G>T		Intron 2	Not Classified	-	Familial cancer of breast		ClinVar			<a href="#">4295493</a>
c.246-252A>C		Intron 2	Benign	Sep 26, 2018			ClinVar			<a href="#">1221335</a>
c.246-194A>G		Intron 2	Benign	Jun 19, 2018			ClinVar			<a href="#">678065</a>
c.246-140G>A		Intron 2	Likely Benign	Sep 06, 2018			ClinVar			<a href="#">1203630</a>
c.246-20G>A		Intron 2	Likely Benign	Jul 14, 2023	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">2.05e-06</a>	ClinVar; gnomAD			<a href="#">2938561</a>
c.246-20G>C		Intron 2	Likely Benign	Dec 24, 2024	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">2950945</a>
c.246-19G>C		Intron 2	Likely Benign	Jun 17, 2025	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">4793019</a>
c.246-18C>T		Intron 2	Likely Benign	Jul 11, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">6.84e-07</a>	ClinVar; gnomAD			<a href="#">2937534</a>
c.246-17_246-3del		Intron 2	Likely Pathogenic	-	Infantile GM1 gangliosidosis	<a href="#">6.84e-07</a>	ClinVar; gnomAD			<a href="#">1332779</a>
c.246-16G>C		Intron 2	Likely Benign	Nov 24, 2025	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">1.37e-06</a>	ClinVar; gnomAD			<a href="#">2944086</a>
c.246-16GT[2]		Intron 2	VUS	Aug 21, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis; GLB1-related disorder		ClinVar			<a href="#">1975416</a>
c.246-13T>C		Intron 2	Likely Benign	Feb 19, 2024	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">1.09e-05</a>	ClinVar; gnomAD			<a href="#">2933472</a>
c.246-11T>G		Intron 2	Likely Benign	Jan 27, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">6.84e-07</a>	ClinVar; gnomAD			<a href="#">2195217</a>
c.246-10G>A		Intron 2	Likely Benign	Jun 20, 2023	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">1.37e-06</a>	ClinVar; gnomAD			<a href="#">2949039</a>
c.246-10G>T		Intron 2	Likely Benign	Jan 13, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">1937658</a>
c.246-2A>G		Intron 2	Pathogenic	Mar 10, 2023	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">1.37e-06</a>	ClinVar; gnomAD; Bidchol 2015 (PMID:25936995)	<a href="#">16199547</a> ; <a href="#">18524657</a> ; <a href="#">25936995</a>		<a href="#">1419594</a>
c.247T>C	p.Tyr83His	Exon 3	Pathogenic	Aug 01, 1995	Mucopolysaccharidosis; MPS-IV-B		ClinVar	<a href="#">7586649</a>		<a href="#">937</a>
c.248A>G	p.Tyr83Cys	Exon 3	P/LP	Dec 13, 2024	Infantile GM1 gangliosidosis;GM1 gangliosidosis type 2;Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis type 3; GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B; Mucopolysaccharidosis		ClinVar; Santamaria 2006 (PMID:16941474)	<a href="#">16941474</a> ; <a href="#">17664528</a> ; <a href="#">21520340</a> ; <a href="#">23337983</a> ; <a href="#">30138938</a> ; <a href="#">7586649</a>		<a href="#">555286</a>
c.253C>A	p.Pro85Thr	Exon 3	VUS	May 07, 2019	GM1 gangliosidosis type 2		ClinVar			<a href="#">684412</a>
c.253C>T	p.Pro85Ser	Exon 3	Likely Pathogenic	-	Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">2627665</a>
c.256_278dup	p.Gln95fs	Exon 3	Pathogenic	May 01, 1992	Infantile GM1 gangliosidosis		ClinVar	<a href="#">1606711</a> ; <a href="#">1907800</a>		<a href="#">934</a>
c.257G>A	p.Trp86Ter	Exon 3	P/LP	Feb 16, 2025	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B; Mucopolysaccharidosis	<a href="#">6.84e-07</a>	ClinVar; gnomAD	<a href="#">18524657</a>		<a href="#">1355177</a>
c.261C>T	p.Asn87=	Exon 3	Likely Benign	May 24, 2022	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">6.57e-06</a>	ClinVar; gnomAD			<a href="#">2123699</a>
c.262T>C	p.Phe88Leu	Exon 3	VUS	Sep 26, 2022	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">1716905</a>
c.266A>T	p.His89Leu	Exon 3	Pathogenic	-	Infantile GM1 gangliosidosis		ClinVar			<a href="#">1332778</a>

cDNA Change	Protein Change	Location	Pathogenicity	Classified	Phenotype	gnomAD AF	Sources	PMID(s)	PIN	ClinVar ID
c.268G>C	p.Glu90Gln	Exon 3	VUS	Mar 09, 2022	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">2107803</a>
c.270G>A	p.Glu90=	Exon 3	Likely Benign	Nov 23, 2025	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">1.37e-06</a>	ClinVar; gnomAD			<a href="#">1398390</a>
c.271C>A	p.Pro91Thr	Exon 3	VUS	Apr 20, 2021		<a href="#">1.37e-06</a>	ClinVar; gnomAD			<a href="#">1314581</a>
c.275G>A	p.Trp92Ter	Exon 3	Pathogenic	May 04, 2024	GM1 gangliosidosis type 3;Infantile GM1 gangliosidosis;GM1 gangliosidosis type 2;Mucopolysaccharidosis; MPS-IV-B	<a href="#">6.84e-07</a>	ClinVar; gnomAD			<a href="#">3589141</a>
c.276G>A	p.Trp92Ter	Exon 3	Pathogenic	Feb 06, 2025	GM1 gangliosidosis type 3;Infantile GM1 gangliosidosis;GM1 gangliosidosis type 2;Mucopolysaccharidosis; MPS-IV-B; Infantile GM1 gangliosidosis; GM1 gangliosidosis;Mucopolysaccharidosis	<a href="#">9.58e-06</a>	ClinVar; LOVD; gnomAD; Bidchol 2015 (PMID:25936995)	<a href="#">18524657</a> ; <a href="#">21520340</a> ; <a href="#">25936995</a>		<a href="#">264673</a>
c.276dup		Exon 3	Not Classified			6.84e-07	LOVD; gnomAD			
c.278C>A	p.Pro93Gln	Exon 3	VUS	Aug 15, 2022	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">2119492</a>
c.279A>G	p.Pro93=	Exon 3	Likely Benign	Dec 19, 2023	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">6.16e-06</a>	ClinVar; gnomAD			<a href="#">2952355</a>
c.281G>A	p.Gly94Glu	Exon 3	VUS	May 28, 2024	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">6.84e-07</a>	ClinVar; gnomAD			<a href="#">591171</a>
c.288C>G	p.Tyr96Ter	Exon 3	Likely Pathogenic	May 20, 2024	GM1 gangliosidosis type 3;Infantile GM1 gangliosidosis;GM1 gangliosidosis type 2;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">3589140</a>
c.289C>T	p.Gln97Ter	Exon 3	Likely Pathogenic	Feb 13, 2024	GM1 gangliosidosis type 3;Infantile GM1 gangliosidosis;GM1 gangliosidosis type 2;Mucopolysaccharidosis; MPS-IV-B	<a href="#">6.84e-07</a>	ClinVar; gnomAD			<a href="#">3589139</a>
c.295T>A	p.Ser99Thr	Exon 3	VUS	Dec 22, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">4.79e-06</a>	ClinVar; gnomAD			<a href="#">1368937</a>
c.302del	p.Asp101fs	Exon 3	Likely Pathogenic	Mar 29, 2018	Infantile GM1 gangliosidosis;GM1 gangliosidosis type 2;Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis type 3		ClinVar			<a href="#">557540</a>
c.304C>G	p.His102Asp	Exon 3	Pathogenic	Aug 08, 2024	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">1.37e-06</a>	ClinVar; gnomAD	<a href="#">20920281</a>		<a href="#">2203327</a>
c.312G>A	p.Val104=	Exon 3	Likely Benign	Nov 03, 2025	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">1.57e-05</a>	ClinVar; gnomAD			<a href="#">1659283</a>
c.315A>G	p.Glu105=	Exon 3	Likely Benign	Oct 10, 2021	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">1576082</a>
c.318T>C	p.Tyr106=	Exon 3	Likely Benign	Apr 16, 2023	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">1.37e-06</a>	ClinVar; gnomAD			<a href="#">2946577</a>

cDNA Change	Protein Change	Location	Pathogenicity	Classified	Phenotype	gnomAD AF	Sources	PMID(s)	PIN	ClinVar ID
c.319T>C	p.Phe107Leu	Exon 3	Likely Pathogenic	Oct 08, 2025	GM1 gangliosidosis type 2; Inborn genetic diseases; GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis; GM1 gangliosidosis type 3;Infantile GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis type 2; Mucopolysaccharidosis	<a href="#">6.84e-07</a>	ClinVar; gnomAD; Cure GM1 AllStripes PIN	<a href="#">20175788</a> ; <a href="#">21520340</a> ; <a href="#">22128166</a> ; <a href="#">23337983</a> ; <a href="#">26108645</a> ; <a href="#">34514040</a>	1	<a href="#">68478</a>
c.321T>C	p.Phe107=	Exon 3	Likely Benign	Feb 17, 2023	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">2944092</a>
c.323T>C		Exon 3	Not Classified				LOVD			
c.325C>T	p.Arg109Trp	Exon 3	Likely Benign	Feb 04, 2026	Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">0.01900</a>	ClinVar; LOVD; gnomAD			<a href="#">129154</a>
c.326G>A	p.Arg109Gln	Exon 3	VUS	Jul 22, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis; Gastric cancer	<a href="#">8.89e-06</a>	ClinVar; gnomAD			<a href="#">2201707</a>
c.326G>C	p.Arg109Pro	Exon 3	VUS	Feb 18, 2025	Infantile GM1 gangliosidosis	<a href="#">1.37e-06</a>	ClinVar; gnomAD			<a href="#">3773739</a>
c.327G>A	p.Arg109=	Exon 3	Likely Benign	Aug 14, 2025	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">1.85e-05</a>	ClinVar; gnomAD			<a href="#">704462</a>
c.327G>C	p.Arg109=	Exon 3	Likely Benign	Sep 06, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">2934568</a>
c.327G>T	p.Arg109=	Exon 3	Likely Benign	Feb 13, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">1.97e-05</a>	ClinVar; gnomAD			<a href="#">1081707</a>
c.331G>A	p.Ala111Thr	Exon 3	VUS	May 16, 2022	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">2.05e-06</a>	ClinVar; gnomAD			<a href="#">2125855</a>
c.334C>T	p.His112Tyr	Exon 3	Likely Benign	Jan 01, 2019	Intellectual disability		ClinVar	<a href="#">21956720</a> ; <a href="#">25157020</a> ; <a href="#">34131312</a> ; <a href="#">34211152</a>		<a href="#">975402</a>
c.335A>C	p.His112Pro	Exon 3	P/LP	Nov 08, 2024	Infantile GM1 gangliosidosis;GM1 gangliosidosis type 2;Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis type 3; GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis	<a href="#">2.05e-05</a>	ClinVar; gnomAD	<a href="#">20175788</a> ; <a href="#">31720227</a> ; <a href="#">35937492</a>		<a href="#">555030</a>
c.335A>G		Exon 3	Not Classified			8.89e-06	LOVD; gnomAD			
c.336T>C	p.His112=	Exon 3	Likely Benign	Dec 30, 2025	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">1.23e-05</a>	ClinVar; gnomAD			<a href="#">1104940</a>
c.342G>A	p.Leu114=	Exon 3	Likely Benign	Nov 18, 2021	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">1139388</a>
c.344del	p.Gly115fs	Exon 3	Pathogenic	Sep 02, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar	<a href="#">18524657</a>		<a href="#">2028587</a>
c.349C>T	p.Leu117=	Exon 3	Likely Benign	Sep 29, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">2931878</a>
c.357C>G	p.Ile119Met	Exon 3	VUS	May 05, 2025	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">6.16e-06</a>	ClinVar; gnomAD			<a href="#">1947618</a>
c.360G>A	p.Leu120=	Exon 3	Likely Benign	Sep 25, 2021	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">1584786</a>
c.363G>A	p.Arg121=	Exon 3	Likely Benign	Jan 07, 2024	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">6.84e-07</a>	ClinVar; gnomAD			<a href="#">2093091</a>
c.363G>T	p.Arg121Ser	Exon 3	VUS	Dec 20, 2024		<a href="#">6.84e-07</a>	ClinVar; gnomAD	<a href="#">10338095</a>		<a href="#">3768497</a>

cDNA Change	Protein Change	Location	Pathogenicity	Classified	Phenotype	gnomAD AF	Sources	PMID(s)	PIN	ClinVar ID
c.366C>G	p.Pro122=	Exon 3	Likely Benign	Nov 01, 2022	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">1.37e-06</a>	ClinVar; gnomAD			<a href="#">1559726</a>
c.366C>T	p.Pro122=	Exon 3	Likely Benign	Jan 28, 2024	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">2.53e-05</a>	ClinVar; gnomAD			<a href="#">1089433</a>
c.367G>A	p.Gly123Arg	Exon 3	Pathogenic	Dec 26, 2025	Infantile GM1 gangliosidosis; Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis; MPS-IV-B	<a href="#">7.52e-06</a>	ClinVar; gnomAD; Bidchol 2015 (PMID:25936995)	<a href="#">17221873</a> ; <a href="#">1817034</a> ; <a href="#">1907800</a> ; <a href="#">22371915</a> ; <a href="#">25326637</a> ; <a href="#">25936995</a> ; <a href="#">33240792</a>		<a href="#">928</a>
c.370C>T	p.Pro124Ser	Exon 3	Likely Pathogenic	Nov 21, 2024	GM1 gangliosidosis		ClinVar	<a href="#">34813777</a>		<a href="#">4531892</a>
c.371C>G		Exon 3	Not Classified				LOVD			
c.374A>G	p.Tyr125Cys	Exon 3	VUS	Apr 28, 2017	Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis	<a href="#">3.42e-06</a>	ClinVar; gnomAD			<a href="#">903250</a>
c.375C>T	p.Tyr125=	Exon 3	Likely Benign	Aug 24, 2024	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">3757528</a>
c.376A>C	p.Ile126Leu	Exon 3	VUS	Jul 08, 2024			ClinVar			<a href="#">3572598</a>
c.376A>G	p.Ile126Val	Exon 3	VUS	Aug 24, 2021	Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">6.84e-07</a>	ClinVar; gnomAD			<a href="#">344794</a>
c.377T>C	p.Ile126Thr	Exon 3	VUS	Jan 21, 2022		<a href="#">1.97e-05</a>	ClinVar; gnomAD			<a href="#">1693866</a>
c.378C>T	p.Ile126=	Exon 3	Likely Benign	May 09, 2023	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">2947580</a>
c.380G>A	p.Cys127Tyr	Exon 3	Likely Pathogenic	Dec 04, 2024	GM1 gangliosidosis; Mucopolysaccharidosis; MPS-IV-B		ClinVar; Cure GM1 AllStripes PIN	<a href="#">20175788</a> ; <a href="#">25600812</a> ; <a href="#">39303319</a>	1	<a href="#">918041</a>
c.380G>T	p.Cys127Phe	Exon 3	Conflicting	Jul 25, 2025	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis; GM1 gangliosidosis type 3;Mucopolysaccharidosis; MPS-IV-B;Infantile GM1 gangliosidosis;GM1 gangliosidosis type 2	<a href="#">6.57e-06</a>	ClinVar; gnomAD	<a href="#">20175788</a> ; <a href="#">30548430</a>		<a href="#">1485842</a>
c.384A>G	p.Ala128=	Exon 3	Likely Benign	Sep 15, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">2930246</a>
c.385G>A	p.Glu129Lys	Exon 3	VUS	Oct 21, 2015		<a href="#">1.37e-06</a>	ClinVar; gnomAD			<a href="#">281025</a>
c.385G>C	p.Glu129Gln	Exon 3	Likely Pathogenic	Oct 21, 2019	Infantile GM1 gangliosidosis	<a href="#">1.37e-06</a>	ClinVar; gnomAD			<a href="#">800730</a>
c.390G>A	p.Trp130Ter	Exon 3	Pathogenic	Jul 20, 2025	GM1 gangliosidosis; GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar	<a href="#">18524657</a> ; <a href="#">31761138</a>		<a href="#">1699092</a>
c.391G>A	p.Glu131Lys	Exon 3	VUS	Dec 23, 2024			ClinVar; LOVD	<a href="#">21520340</a>		<a href="#">3768483</a>
c.395T>C	p.Met132Thr	Exon 3	P/LP	May 04, 2022	Infantile GM1 gangliosidosis;GM1 gangliosidosis type 2;Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis type 3; Mucopolysaccharidosis; MPS-IV-B; Infantile GM1 gangliosidosis		ClinVar; Hofer 2009 (PMID:19472408)	<a href="#">19472408</a>		<a href="#">556862</a>
c.396+2T>C		Intron 3	P/LP	Jan 20, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis; Thyroid cancer; nonmedullary; 1		ClinVar	<a href="#">16199547</a> ; <a href="#">18524657</a>		<a href="#">372370</a>
c.396+2T>G		Intron 3	Not Classified	-	Thyroid cancer; nonmedullary; 1		ClinVar			<a href="#">4295492</a>
c.396+7G>T		Intron 3	Likely Benign	Jul 11, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">2016069</a>

cDNA Change	Protein Change	Location	Pathogenicity	Classified	Phenotype	gnomAD AF	Sources	PMID(s)	PIN	ClinVar ID
c.396+9G>A		Intron 3	Likely Benign	Mar 28, 2023	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">2945914</a>
c.396+10G>T		Intron 3	Likely Benign	Apr 16, 2024	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">3755894</a>
c.396+11C>G		Intron 3	Likely Benign	Oct 27, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">6.84e-07</a>	ClinVar; gnomAD			<a href="#">1935015</a>
c.396+14G>T		Intron 3	Likely Benign	Nov 20, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">2.05e-06</a>	ClinVar; gnomAD			<a href="#">2929017</a>
c.396+15A>G		Intron 3	Likely Benign	Feb 12, 2025	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">6.84e-07</a>	ClinVar; gnomAD			<a href="#">2934647</a>
c.396+18G>T		Intron 3	Likely Benign	Feb 16, 2024	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">6.84e-07</a>	ClinVar; gnomAD			<a href="#">3754703</a>
c.396+20G>A		Intron 3	Likely Benign	Mar 27, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">4.11e-06</a>	ClinVar; gnomAD			<a href="#">2931457</a>
c.396+61G>A		Intron 3	Likely Benign	Sep 06, 2018		<a href="#">0.00954</a>	ClinVar; gnomAD			<a href="#">1212122</a>
c.396+122C>A		Intron 3	Benign	Jun 19, 2018			ClinVar			<a href="#">678066</a>
c.397-200G>T		Intron 3	Not Classified	-	Lung cancer		ClinVar	<a href="#">29398453</a>		<a href="#">4295491</a>
c.397-19C>T		Intron 3	Likely Benign	Jul 30, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">2020412</a>
c.397-17T>C		Intron 3	Likely Benign	Jun 05, 2023	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">2948279</a>
c.397-16G>C		Intron 3	Likely Benign	Oct 26, 2025	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">1.37e-06</a>	ClinVar; gnomAD			<a href="#">4784114</a>
c.397-11C>T		Intron 3	Likely Benign	Nov 08, 2024	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">2.53e-05</a>	ClinVar; gnomAD			<a href="#">2150056</a>
c.397-10C>T		Intron 3	Likely Benign	Mar 01, 2024	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">3754671</a>
c.397-8T>C		Intron 3	Likely Benign	Apr 05, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">6.84e-07</a>	ClinVar; gnomAD			<a href="#">1971537</a>
c.397-7G>C		Intron 3	Likely Benign	Sep 28, 2025	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">4786259</a>
c.397-5A>G		Intron 3	Likely Benign	Jan 28, 2026	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">2.60e-05</a>	ClinVar; gnomAD			<a href="#">705730</a>
c.397-2A>G		Intron 3	Likely Pathogenic		GM1 gangliosidosis		Bidchol 2015 (PMID:25936995)	<a href="#">25936995</a>		
c.397-1G>A		Intron 3	P/LP	Oct 16, 2024	GM1 gangliosidosis type 2;Infantile GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis type 3; Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis; MPS-IV-B	<a href="#">2.74e-06</a>	ClinVar; gnomAD; Bidchol 2015 (PMID:25936995)	<a href="#">16199547</a> ; <a href="#">18524657</a> ; <a href="#">25936995</a>		<a href="#">92906</a>
c.397G>T	p.Gly133Ter	Exon 4	Pathogenic	May 13, 2021	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">6.84e-07</a>	ClinVar; gnomAD	<a href="#">18524657</a>		<a href="#">1452262</a>
c.399A>G	p.Gly133=	Exon 4	Likely Benign	Mar 17, 2025	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">8.89e-06</a>	ClinVar; gnomAD			<a href="#">2039525</a>
c.400G>A	p.Gly134Arg	Exon 4	Pathogenic	Jun 26, 2024	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar; Bidchol 2015 (PMID:25936995)	<a href="#">17309651</a> ; <a href="#">25936995</a> ; <a href="#">33558080</a>		<a href="#">3759085</a>

cDNA Change	Protein Change	Location	Pathogenicity	Classified	Phenotype	gnomAD AF	Sources	PMID(s)	PIN	ClinVar ID
c.401G>T	p.Gly134Val	Exon 4	Pathogenic	Jul 23, 2024	Infantile GM1 gangliosidosis;GM1 gangliosidosis type 2;Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis type 3; GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">6.84e-07</a>	ClinVar; gnomAD; Bidchol 2015 (PMID:25936995)	<a href="#">17309651</a> ; <a href="#">21520340</a> ; <a href="#">22128166</a> ; <a href="#">23337983</a> ; <a href="#">25936995</a> ; <a href="#">33558080</a>		<a href="#">556153</a>
c.402A>T	p.Gly134=	Exon 4	Likely Benign	May 24, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">7.53e-06</a>	ClinVar; gnomAD			<a href="#">1133197</a>
c.407C>T	p.Pro136Leu	Exon 4	Likely Pathogenic	Jan 13, 2020	Infantile GM1 gangliosidosis	<a href="#">6.84e-07</a>	ClinVar; gnomAD			<a href="#">818226</a>
c.411T>C	p.Ala137=	Exon 4	Likely Benign	Aug 05, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">6.84e-07</a>	ClinVar; gnomAD			<a href="#">1528681</a>
c.414G>A	p.Trp138Ter	Exon 4	Likely Pathogenic	Nov 06, 2025	Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">4818320</a>
c.416T>A	p.Leu139Gln	Exon 4	Likely Pathogenic	Jul 05, 2019	Infantile GM1 gangliosidosis		ClinVar			<a href="#">872974</a>
c.424A>G		Exon 4	Not Classified			1.37e-06	LOVD; gnomAD			
c.424A>T	p.Lys142Ter	Exon 4	Pathogenic	Feb 19, 2024	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">1.37e-06</a>	ClinVar; gnomAD	<a href="#">18524657</a> ; <a href="#">29439846</a> ; <a href="#">29451896</a>		<a href="#">1395005</a>
c.425_426del	p.Lys142fs	Exon 4	Likely Pathogenic	Jun 06, 2018	Infantile GM1 gangliosidosis;GM1 gangliosidosis type 2;GM1 gangliosidosis type 3;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">558711</a>
c.425_428del	p.Lys142fs	Exon 4	Pathogenic	Feb 22, 2023	Infantile GM1 gangliosidosis; GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">6.57e-06</a>	ClinVar; gnomAD	<a href="#">18524657</a> ; <a href="#">21497194</a>		<a href="#">1332777</a>
c.426A>G	p.Lys142=	Exon 4	Likely Benign	Jun 19, 2025	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">7.53e-06</a>	ClinVar; gnomAD			<a href="#">1645136</a>
c.426dup	p.Glu143fs	Exon 4	Pathogenic	May 27, 2023	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar	<a href="#">18524657</a>		<a href="#">2948270</a>
c.427G>A		Exon 4	Not Classified				LOVD			
c.428dup		Exon 4	Not Classified				LOVD			
c.429G>A	p.Glu143=	Exon 4	Likely Benign	Apr 18, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">1919979</a>
c.433_437del	p.Ile145fs	Exon 4	P/LP	Mar 27, 2024	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis type 2;GM1 gangliosidosis type 3;Infantile GM1 gangliosidosis; MPS-IV-B;GM1 gangliosidosis; MPS-IV-B		ClinVar	<a href="#">18524657</a>		<a href="#">550513</a>
c.434T>C	p.Ile145Thr	Exon 4	VUS	Aug 28, 2021	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">6.84e-07</a>	ClinVar; gnomAD			<a href="#">659663</a>
c.435T>C	p.Ile145=	Exon 4	Likely Benign	Jan 11, 2024	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">6.84e-07</a>	ClinVar; gnomAD			<a href="#">2923012</a>
c.435TCT[1]	p.Leu147del	Exon 4	P/LP	Jul 21, 2025	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis; Infantile GM1 gangliosidosis;GM1 gangliosidosis type 2;Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis type 3; GLB1-related disorder; MPS-IV-B		ClinVar	<a href="#">17309651</a> ; <a href="#">21520340</a> ; <a href="#">29800929</a>		<a href="#">1473187</a>
c.435_436delinsA	p.Leu146fs	Exon 4	Likely Pathogenic	May 19, 2024	GM1 gangliosidosis type 3;Infantile GM1 gangliosidosis;GM1 gangliosidosis type 2;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">3589138</a>

cDNA Change	Protein Change	Location	Pathogenicity	Classified	Phenotype	gnomAD AF	Sources	PMID(s)	PIN	ClinVar ID
c.438_440del		Exon 4	Not Classified			1.30e-05	LOVD; gnomAD			
c.439C>T	p.Leu147Phe	Exon 4	Likely Pathogenic	May 22, 2023	Infantile GM1 gangliosidosis; Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">1.37e-06</a>	ClinVar; LOVD; gnomAD	<a href="#">33737400</a>		<a href="#">522669</a>
c.441C>T	p.Leu147=	Exon 4	Likely Benign	Jul 01, 2025		<a href="#">6.84e-07</a>	ClinVar; gnomAD			<a href="#">4084442</a>
c.442C>A	p.Arg148Ser	Exon 4	P/LP	Jan 16, 2026	GM1 gangliosidosis type 3;Mucopolysaccharidosis; MPS-IV-B;Infantile GM1 gangliosidosis;GM1 gangliosidosis type 2; Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis; GM1 gangliosidosis; GLB1-related disorder; MPS-IV-B; Lung cancer	<a href="#">8.28e-05</a>	ClinVar; LOVD; gnomAD; Bidchol 2015 (PMID:25936995); Cure GM1 AllStripes PIN	<a href="#">10839995</a> ; <a href="#">10841810</a> ; <a href="#">15365997</a> ; <a href="#">15986423</a> ; <a href="#">17221873</a> ; <a href="#">20175788</a> ; <a href="#">23151865</a> ; <a href="#">25600812</a> ; <a href="#">25936995</a> ; <a href="#">2715932</a> ; <a href="#">29352662</a>	2	<a href="#">551757</a>
c.442C>T	p.Arg148Cys	Exon 4	P/LP	Aug 20, 2025	GM1 gangliosidosis type 2;Infantile GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis type 3; GM1 gangliosidosis; Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis; Infantile GM1 gangliosidosis; GLB1-related disorder; MPS-IV-B	<a href="#">7.53e-05</a>	ClinVar; LOVD; gnomAD; NTSAD; Bidchol 2015 (PMID:25936995); Cure GM1 AllStripes PIN	<a href="#">10839995</a> ; <a href="#">15986423</a> ; <a href="#">16674934</a> ; <a href="#">17221873</a> ; <a href="#">20175788</a> ; <a href="#">21497194</a> ; <a href="#">21520340</a> ; <a href="#">22128166</a> ; <a href="#">23151865</a> ; <a href="#">25936995</a> ; <a href="#">30712135</a> ; <a href="#">33240792</a> ; <a href="#">33737400</a> ; <a href="#">37381921</a>	2	<a href="#">92907</a>
c.443G>A	p.Arg148His	Exon 4	P/LP	Jan 20, 2026	Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis type 2;GM1 gangliosidosis type 3;Mucopolysaccharidosis; MPS-IV-B;Infantile GM1 gangliosidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">5.47e-06</a>	ClinVar; gnomAD; Bidchol 2015 (PMID:25936995)	<a href="#">15986423</a> ; <a href="#">17221873</a> ; <a href="#">21497194</a> ; <a href="#">23151865</a> ; <a href="#">25936995</a> ; <a href="#">30267299</a> ; <a href="#">33737400</a>		<a href="#">522884</a>
c.443G>T	p.Arg148Leu	Exon 4	Likely Pathogenic	Jul 25, 2025	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis; MPS-IV-B		ClinVar	<a href="#">10839995</a> ; <a href="#">15986423</a> ; <a href="#">20175788</a> ; <a href="#">21497194</a> ; <a href="#">23151865</a> ; <a href="#">25600812</a> ; <a href="#">33240792</a>		<a href="#">2935679</a>
c.444C>T	p.Arg148=	Exon 4	Likely Benign	Sep 11, 2025	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">4789948</a>
c.446C>T	p.Ser149Phe	Exon 4	P/LP	May 23, 2025	GM1 gangliosidosis type 3;Mucopolysaccharidosis; MPS-IV-B;Infantile GM1 gangliosidosis;GM1 gangliosidosis type 2; GM1 gangliosidosis type 2; Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis; MPS-IV-B; GLB1-related disorder	<a href="#">2.74e-06</a>	ClinVar; gnomAD; Hofer 2009 (PMID:19472408)	<a href="#">19472408</a> ; <a href="#">25326635</a> ; <a href="#">29451896</a> ; <a href="#">30267299</a> ; <a href="#">30675867</a>		<a href="#">553205</a>
c.447C>G	p.Ser149=	Exon 4	Likely Benign	Mar 05, 2025	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">3749817</a>
c.447C>T	p.Ser149=	Exon 4	Likely Benign	Dec 10, 2023	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">2944112</a>
c.450C>T	p.Ser150=	Exon 4	Likely Benign	Jan 26, 2026	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">3.35e-05</a>	ClinVar; gnomAD			<a href="#">1136346</a>
c.451G>A	p.Asp151Asn	Exon 4	Likely Pathogenic	Apr 09, 2024	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">1.30e-05</a>	ClinVar; gnomAD	<a href="#">15365997</a> ; <a href="#">15791924</a> ; <a href="#">32219518</a>		<a href="#">1474580</a>
c.451G>T	p.Asp151Tyr	Exon 4	Likely Pathogenic	Oct 04, 2024	Infantile GM1 gangliosidosis; GM1 gangliosidosis		ClinVar	<a href="#">32219518</a>		<a href="#">1300015</a>

cDNA Change	Protein Change	Location	Pathogenicity	Classified	Phenotype	gnomAD AF	Sources	PMID(s)	PIN	ClinVar ID
c.452A>T	p.Asp151Val	Exon 4	P/LP	Dec 04, 2025	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B; Mucopolysaccharidosis		ClinVar; Santamaria 2006 (PMID:16941474)	<a href="#">15365997</a> ; <a href="#">15791924</a> ; <a href="#">16941474</a> ; <a href="#">23337983</a> ; <a href="#">32219518</a>		<a href="#">3759084</a>
c.454C>A	p.Pro152Thr	Exon 4	VUS	Feb 14, 2022	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">2097572</a>
c.456A>C		Exon 4	Not Classified				LOVD			
c.456A>T	p.Pro152=	Exon 4	Not Classified	-	GM1 gangliosidosis type 2		ClinVar			<a href="#">68480</a>
c.457+2T>C		Intron 4	P/LP	Sep 06, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar	<a href="#">16199547</a> ; <a href="#">18524657</a>		<a href="#">92908</a>
c.457+3A>C		Intron 4	VUS	Dec 01, 2020			ClinVar			<a href="#">1012494</a>
c.457+3A>G		Intron 4	VUS	Mar 14, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">2.05e-06</a>	ClinVar; gnomAD	<a href="#">17576681</a> ; <a href="#">9536098</a>		<a href="#">2083621</a>
c.457+11dup		Intron 4	Likely Benign	Aug 18, 2025	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">4.10e-06</a>	ClinVar; gnomAD			<a href="#">2947505</a>
c.457+12C>A		Intron 4	Likely Benign	Dec 01, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">6.57e-06</a>	ClinVar; gnomAD			<a href="#">2930143</a>
c.457+14G>A		Intron 4	Likely Benign	Aug 02, 2023	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">2944594</a>
c.457+15A>G		Intron 4	Likely Benign	Jun 23, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">2.05e-06</a>	ClinVar; gnomAD			<a href="#">2922894</a>
c.457+19C>T		Intron 4	Likely Benign	Feb 06, 2024	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">1.37e-06</a>	ClinVar; gnomAD			<a href="#">2189626</a>
c.457+20T>C		Intron 4	Likely Benign	Jan 15, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">6.57e-06</a>	ClinVar; gnomAD			<a href="#">2933086</a>
c.457+53C>A		Intron 4	Likely Benign	Dec 31, 2018	Lung cancer	<a href="#">0.00096</a>	ClinVar; gnomAD			<a href="#">1204645</a>
c.457+154C>T		Intron 4	Benign	Sep 26, 2018			ClinVar			<a href="#">1286465</a>
c.457+235A>G		Intron 4	Benign	Jun 19, 2018			ClinVar			<a href="#">678493</a>
c.457+268A>G		Intron 4	Not Classified	-	Ovarian serous cystadenocarcinoma		ClinVar			<a href="#">4295490</a>
c.457+283T>C		Intron 4	Benign	Sep 26, 2018			ClinVar			<a href="#">1279985</a>
c.458-401_552+1033del		Intron 4	Not Classified				LOVD			
c.458-14T>A		Intron 4	Likely Benign	Dec 01, 2025	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">2930433</a>
c.458-13T>C		Intron 4	Likely Benign	Mar 17, 2023	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">7.09e-07</a>	ClinVar; gnomAD			<a href="#">2946129</a>
c.458-13del		Intron 4	Benign	Nov 28, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">2.84e-06</a>	ClinVar; gnomAD			<a href="#">2925762</a>
c.458-11T>C		Intron 4	Benign	Feb 04, 2026	Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis; MPS-IV-B;GM1 gangliosidosis; Infantile GM1 gangliosidosis; Thymoma; Nonpapillary renal cell carcinoma; Familial cancer of breast; Ovarian cancer; Familial pancreatic carcinoma; Chronic lymphocytic leukemia/small lymphocytic lymphoma; Gastric cancer	<a href="#">0.11000</a>	ClinVar; LOVD; gnomAD			<a href="#">92909</a>
c.458-5T>C		Intron 4	Likely Benign	Mar 23, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">1946827</a>

cDNA Change	Protein Change	Location	Pathogenicity	Classified	Phenotype	gnomAD AF	Sources	PMID(s)	PIN	ClinVar ID
c.462C>G	p.Tyr154Ter	Exon 5	Likely Pathogenic	Apr 17, 2024	GM1 gangliosidosis type 3;Infantile GM1 gangliosidosis;GM1 gangliosidosis type 2;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">3589137</a>
c.462C>T	p.Tyr154=	Exon 5	Likely Benign	Jan 21, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">1965746</a>
c.464T>G	p.Leu155Arg	Exon 5	Pathogenic	Jan 21, 2026	GM1 gangliosidosis type 3;Infantile GM1 gangliosidosis;GM1 gangliosidosis type 2;Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis;Mucopolysaccharidosis; Mucopolysaccharidosis; Infantile GM1 gangliosidosis	<a href="#">9.85e-06</a>	ClinVar; gnomAD; Hofer 2009 (PMID:19472408); Cure GM1 AllStripes PIN	<a href="#">17309651</a> ; <a href="#">19472408</a> ; <a href="#">20175788</a> ; <a href="#">21520340</a> ; <a href="#">25557439</a>	3	<a href="#">265179</a>
c.467C>T	p.Ala156Val	Exon 5	VUS	Aug 11, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">4.21e-06</a>	ClinVar; gnomAD			<a href="#">2141823</a>
c.468del	p.Ala157fs	Exon 5	Likely Pathogenic	May 04, 2024	GM1 gangliosidosis type 3;Infantile GM1 gangliosidosis;GM1 gangliosidosis type 2;Mucopolysaccharidosis; MPS-IV-B	<a href="#">7.02e-07</a>	ClinVar; gnomAD			<a href="#">3589136</a>
c.473del	p.Val158fs	Exon 5	Pathogenic	Jun 14, 2020	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar	<a href="#">18524657</a>		<a href="#">1069437</a>
c.479del	p.Lys160fs	Exon 5	Likely Pathogenic	Aug 16, 2024	Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">4818321</a>
c.481T>G	p.Trp161Gly	Exon 5	Conflicting	Dec 22, 2025	GM1 gangliosidosis type 2;Infantile GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis type 3; GM1 gangliosidosis; Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis; MPS-IV-B	<a href="#">5.60e-06</a>	ClinVar; LOVD; gnomAD; Cure GM1 AllStripes PIN	<a href="#">21491941</a> ; <a href="#">21497194</a> ; <a href="#">21520340</a> ; <a href="#">23337983</a> ; <a href="#">25600812</a>	2	<a href="#">92910</a>
c.482G>A	p.Trp161Ter	Exon 5	Pathogenic	Jan 26, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">7.00e-07</a>	ClinVar; gnomAD	<a href="#">18524657</a> ; <a href="#">20175788</a>		<a href="#">2157091</a>
c.482G>T	p.Trp161Leu	Exon 5	VUS	Jan 13, 2026		<a href="#">1.40e-06</a>	ClinVar; gnomAD			<a href="#">4689436</a>
c.485T>C	p.Leu162Ser	Exon 5	Likely Pathogenic	Oct 09, 2025	Mucopolysaccharidosis; MPS-IV-B		ClinVar	<a href="#">17309651</a> ; <a href="#">17664528</a>		<a href="#">4687404</a>
c.485del	p.Leu162fs	Exon 5	Pathogenic	Aug 20, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar; LOVD	<a href="#">18524657</a>		<a href="#">1911508</a>
c.486G>A	p.Leu162=	Exon 5	Likely Benign	May 14, 2025	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis; Inborn genetic diseases	<a href="#">3.49e-06</a>	ClinVar; gnomAD			<a href="#">1050398</a>
c.487_488del	p.Gly163fs	Exon 5	Likely Pathogenic	Mar 30, 2022	Infantile GM1 gangliosidosis;GM1 gangliosidosis type 3;GM1 gangliosidosis type 2		ClinVar			<a href="#">1725613</a>
c.488dup	p.Val164fs	Exon 5	Likely Pathogenic	May 10, 2024	GM1 gangliosidosis type 3;Infantile GM1 gangliosidosis;GM1 gangliosidosis type 2;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">3589135</a>
c.492C>G	p.Val164=	Exon 5	Likely Benign	Oct 21, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">2036420</a>
c.495_497del	p.Leu166del	Exon 5	Conflicting	Nov 10, 2025	GM1 gangliosidosis type 2; GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B; Infantile GM1 gangliosidosis; Mucopolysaccharidosis	<a href="#">5.58e-06</a>	ClinVar; gnomAD	<a href="#">20920281</a> ; <a href="#">30267299</a> ; <a href="#">33737400</a>		<a href="#">684406</a>

cDNA Change	Protein Change	Location	Pathogenicity	Classified	Phenotype	gnomAD AF	Sources	PMID(s)	PIN	ClinVar ID
c.496C>T	p.Leu166=	Exon 5	Likely Benign	Jan 24, 2024	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">1092407</a>
c.500_501del	p.Pro167fs	Exon 5	Likely Pathogenic	Mar 15, 2022	Infantile GM1 gangliosidosis;GM1 gangliosidosis type 3;GM1 gangliosidosis type 2		ClinVar			<a href="#">1725318</a>
c.501C>T	p.Pro167=	Exon 5	VUS	Feb 02, 2018	Infantile GM1 gangliosidosis;GM1 gangliosidosis type 2;Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis type 3		ClinVar			<a href="#">556506</a>
c.504G>A	p.Lys168=	Exon 5	Likely Benign	Sep 17, 2023	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">2951995</a>
c.510G>A	p.Lys170=	Exon 5	Likely Benign	Oct 06, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">2034344</a>
c.514CTC[1]	p.Leu173del	Exon 5	Pathogenic	Sep 19, 2023	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar	<a href="#">26108645</a>		<a href="#">2941466</a>
c.515_516del	p.Leu172fs	Exon 5	Likely Pathogenic	-	Infantile GM1 gangliosidosis		ClinVar			<a href="#">3362831</a>
c.516C>T	p.Leu172=	Exon 5	Likely Benign	Dec 24, 2023	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">2944111</a>
c.517C>G	p.Leu173Val	Exon 5	Likely Pathogenic	Jan 25, 2024	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">6.97e-06</a>	ClinVar; gnomAD; Santamaria 2006 (PMID:16941474)	<a href="#">16941474</a> ; <a href="#">17664528</a>		<a href="#">1523122</a>
c.517C>T	p.Leu173Phe	Exon 5	Likely Pathogenic	Nov 29, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">6.97e-07</a>	ClinVar; gnomAD; Santamaria 2006 (PMID:16941474)	<a href="#">16941474</a> ; <a href="#">17664528</a>		<a href="#">2931731</a>
c.518T>C	p.Leu173Pro	Exon 5	P/LP	Oct 10, 2025	GM1 gangliosidosis type 2; GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis type 3;Infantile GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis type 2; Mucopolysaccharidosis		ClinVar; Santamaria 2006 (PMID:16941474)	<a href="#">16941474</a> ; <a href="#">17664528</a>		<a href="#">68479</a>
c.519C>G	p.Leu173=	Exon 5	Likely Benign	Feb 22, 2024	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">1081737</a>
c.520T>C	p.Tyr174His	Exon 5	Likely Pathogenic	May 27, 2025	Mucopolysaccharidosis; MPS-IV-B		ClinVar; LOVD	<a href="#">25443580</a> ; <a href="#">29451896</a> ; <a href="#">35143101</a>		<a href="#">3902680</a>
c.520T>G	p.Tyr174Asp	Exon 5	Likely Pathogenic	Jul 25, 2023	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar	<a href="#">25443580</a> ; <a href="#">29451896</a>		<a href="#">2950242</a>
c.521A>C	p.Tyr174Ser	Exon 5	Likely Pathogenic	May 06, 2021	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar	<a href="#">25443580</a> ; <a href="#">29451896</a>		<a href="#">1519356</a>
c.521A>G	p.Tyr174Cys	Exon 5	Likely Pathogenic	Feb 09, 2024	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar	<a href="#">25443580</a> ; <a href="#">29451896</a>		<a href="#">3762067</a>
c.523C>T	p.Gln175Ter	Exon 5	Likely Pathogenic	Apr 03, 2017	GM1 gangliosidosis type 3;Mucopolysaccharidosis; MPS-IV-B;Infantile GM1 gangliosidosis;GM1 gangliosidosis type 2		ClinVar			<a href="#">551241</a>
c.524A>G	p.Gln175Arg	Exon 5	VUS	Oct 22, 2023	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">2.09e-06</a>	ClinVar; gnomAD			<a href="#">1357332</a>

cDNA Change	Protein Change	Location	Pathogenicity	Classified	Phenotype	gnomAD AF	Sources	PMID(s)	PIN	ClinVar ID
c.531_534del	p.Gly178fs	Exon 5	P/LP	Dec 12, 2025	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B; Mucopolysaccharidosis; MPS-IV-B;Infantile GM1 gangliosidosis;GM1 gangliosidosis type 3;GM1 gangliosidosis type 2		ClinVar	<a href="#">18524657</a>		<a href="#">837229</a>
c.532G>T	p.Gly178Trp	Exon 5	VUS	Apr 28, 2025	Inborn genetic diseases		ClinVar			<a href="#">4030001</a>
c.534G>A	p.Gly178=	Exon 5	Conflicting	Dec 10, 2024	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis; Mucopolysaccharidosis; Inborn genetic diseases		ClinVar			<a href="#">789413</a>
c.542T>A		Exon 5	Not Classified				LOVD			
c.545C>G	p.Thr182Arg	Exon 5	VUS	Jun 21, 2019	Infantile GM1 gangliosidosis		ClinVar			<a href="#">689485</a>
c.546A>G	p.Thr182=	Exon 5	Likely Benign	May 10, 2025	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis; Inborn genetic diseases		ClinVar			<a href="#">1102868</a>
c.550C>T	p.Gln184Ter	Exon 5	Pathogenic	Apr 11, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar	<a href="#">18524657</a> ; <a href="#">29451896</a>		<a href="#">2927180</a>
c.552+1G>A		Intron 5	Pathogenic	Jan 19, 2023	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar; Bidchol 2015 (PMID:25936995)	<a href="#">16199547</a> ; <a href="#">18524657</a> ; <a href="#">25936995</a>		<a href="#">2942918</a>
c.552+1G>T		Intron 5	Likely Pathogenic		GM1 gangliosidosis		Bidchol 2015 (PMID:25936995)	<a href="#">25936995</a>		
c.552+2T>C		Intron 5	Likely Pathogenic	Jul 22, 2022	GM1 gangliosidosis type 3;Mucopolysaccharidosis; MPS-IV-B;Infantile GM1 gangliosidosis;GM1 gangliosidosis type 2; Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar	<a href="#">16199547</a> ; <a href="#">18524657</a>		<a href="#">553673</a>
c.552+7C>G		Intron 5	Likely Benign	Feb 16, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">7.03e-07</a>	ClinVar; gnomAD			<a href="#">2933316</a>
c.552+8T>C		Intron 5	Likely Benign	Sep 29, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">2.11e-06</a>	ClinVar; gnomAD			<a href="#">1620099</a>
c.552+13T>A		Intron 5	Likely Benign	Mar 19, 2025	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">7.05e-07</a>	ClinVar; gnomAD			<a href="#">2931889</a>
c.552+18C>T		Intron 5	Likely Benign	Sep 02, 2025	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">0.00014</a>	ClinVar; gnomAD			<a href="#">1405579</a>
c.552+19A>G		Intron 5	Likely Benign	Jul 02, 2024	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">3.18e-05</a>	ClinVar; gnomAD			<a href="#">2081168</a>
c.552+21G>A		Intron 5	Likely Benign	Apr 19, 2023		<a href="#">0.00102</a>	ClinVar; gnomAD			<a href="#">496829</a>
c.553-119A>G		Intron 5	Benign	Jun 19, 2018			ClinVar			<a href="#">677897</a>
c.553-20T>C		Intron 5	Likely Benign	Apr 11, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">2928956</a>
c.553-17T>C		Intron 5	Likely Benign	Sep 23, 2025	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">4.10e-06</a>	ClinVar; gnomAD			<a href="#">1649040</a>
c.553-8A>G		Intron 5	VUS	Jan 13, 2018	Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis	<a href="#">1.37e-06</a>	ClinVar; gnomAD			<a href="#">344793</a>
c.553-2A>G		Intron 5	Pathogenic	Jul 02, 2024	Mucopolysaccharidosis; MPS-IV-B		ClinVar	<a href="#">27679996</a>		<a href="#">4818322</a>

cDNA Change	Protein Change	Location	Pathogenicity	Classified	Phenotype	gnomAD AF	Sources	PMID(s)	PIN	ClinVar ID
c.553-2A>T		Intron 5	P/LP	Apr 01, 2024	Infantile GM1 gangliosidosis; Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis; GLB1-related disorder		ClinVar	<a href="#">16199547</a> ; <a href="#">18524657</a> ; <a href="#">27679996</a>		<a href="#">1685330</a>
c.553-1G>C		Intron 5	Pathogenic	Nov 11, 2018	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar	<a href="#">16199547</a> ; <a href="#">18524657</a> ; <a href="#">27679996</a>		<a href="#">639100</a>
c.553G>T	p.Val185Phe	Exon 6	Not Classified	-	Pancreatic adenocarcinoma		ClinVar			<a href="#">4295488</a>
c.557A>C	p.Glu186Ala	Exon 6	Likely Pathogenic	Dec 05, 2025	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis; Mucopolysaccharidosis; Lysosomal storage disease	<a href="#">3.01e-05</a>	ClinVar; gnomAD; Cure GM1 AllStripes PIN	<a href="#">25600812</a>	1	<a href="#">1066585</a>
c.558A>G	p.Glu186=	Exon 6	Likely Benign	Aug 14, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">6.84e-07</a>	ClinVar; gnomAD			<a href="#">2010441</a>
c.560dup	p.Asn187fs	Exon 6	Pathogenic	Nov 13, 2023	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar	<a href="#">18524657</a>		<a href="#">2942613</a>
c.561T>C	p.Asn187=	Exon 6	Likely Benign	Aug 07, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">1.37e-06</a>	ClinVar; gnomAD			<a href="#">1134694</a>
c.562G>T	p.Glu188Ter	Exon 6	Pathogenic	Nov 21, 2023	Infantile GM1 gangliosidosis; Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar	<a href="#">18524657</a>		<a href="#">2507026</a>
c.569G>A	p.Gly190Asp	Exon 6	P/LP	Sep 13, 2024	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B; Infantile GM1 gangliosidosis; Mucopolysaccharidosis	<a href="#">4.79e-06</a>	ClinVar; LOVD; gnomAD; Hofer 2009 (PMID:19472408)	<a href="#">19472408</a> ; <a href="#">21520340</a> ; <a href="#">22234367</a> ; <a href="#">33737400</a>		<a href="#">1353182</a>
c.569G>T	p.Gly190Val	Exon 6	Likely Pathogenic	Aug 08, 2022	Infantile GM1 gangliosidosis		ClinVar			<a href="#">1710133</a>
c.570C>T	p.Gly190=	Exon 6	Not Classified	-	Familial prostate cancer	<a href="#">2.74e-06</a>	ClinVar; gnomAD			<a href="#">4295487</a>
c.572G>A	p.Ser191Asn	Exon 6	P/LP	Jan 21, 2026	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis type 2;GM1 gangliosidosis type 3;Infantile GM1 gangliosidosis; MPS-IV-B;GM1 gangliosidosis; MPS-IV-B	<a href="#">3.42e-06</a>	ClinVar; gnomAD	<a href="#">21497194</a> ; <a href="#">32779865</a>		<a href="#">549962</a>
c.572G>C	p.Ser191Thr	Exon 6	Conflicting	Apr 02, 2025	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar	<a href="#">21497194</a> ; <a href="#">32779865</a>		<a href="#">2945675</a>
c.572G>T	p.Ser191Ile	Exon 6	Likely Pathogenic	Jan 13, 2026	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar	<a href="#">21497194</a> ; <a href="#">32779865</a>		<a href="#">2953563</a>
c.573C>G	p.Ser191Arg	Exon 6	Not Classified	-	Thyroid cancer; nonmedullary; 1	<a href="#">8.90e-06</a>	ClinVar; gnomAD			<a href="#">4295486</a>
c.574T>C	p.Tyr192His	Exon 6	Conflicting	Jun 24, 2025	GM1 gangliosidosis; GLB1-related disorder	<a href="#">6.84e-07</a>	ClinVar; gnomAD; Cure GM1 AllStripes PIN	<a href="#">27159321</a> ; <a href="#">33240792</a> ; <a href="#">33558080</a> ; <a href="#">35029890</a>	1	<a href="#">918052</a>
c.576C>T	p.Tyr192=	Exon 6	Likely Benign	Dec 02, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">2924549</a>
c.579T>C	p.Phe193=	Exon 6	Likely Benign	Nov 20, 2023	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">2954068</a>
c.581C>T	p.Ala194Val	Exon 6	VUS	Aug 21, 2025	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">7.52e-06</a>	ClinVar; gnomAD			<a href="#">2142483</a>
c.586G>C	p.Asp196His	Exon 6	VUS	Nov 02, 2021	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">1401965</a>
c.591del	p.Phe197fs	Exon 6	Pathogenic	Jan 03, 2023	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar	<a href="#">18524657</a>		<a href="#">2942924</a>

cDNA Change	Protein Change	Location	Pathogenicity	Classified	Phenotype	gnomAD AF	Sources	PMID(s)	PIN	ClinVar ID
c.591dup	p.Asp198Ter	Exon 6	Pathogenic	Aug 28, 2025	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis; Infantile GM1 gangliosidosis; GM1 gangliosidosis type 3;Mucopolysaccharidosis; MPS-IV-B;Infantile GM1 gangliosidosis;GM1 gangliosidosis type 2; MPS-IV-B		ClinVar; Santamaria 2006 (PMID:16941474)	<a href="#">16941474</a> ; <a href="#">18524657</a>		<a href="#">1449094</a>
c.592G>C	p.Asp198His	Exon 6	VUS	Dec 09, 2024	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">2416124</a>
c.597C>T	p.Tyr199=	Exon 6	Likely Benign	Aug 23, 2021	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">1567912</a>
c.597_598del	p.Leu200fs	Exon 6	Likely Pathogenic	Mar 13, 2024	GM1 gangliosidosis type 3;Infantile GM1 gangliosidosis;GM1 gangliosidosis type 2;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">3589134</a>
c.601C>T	p.Arg201Cys	Exon 6	P/LP	Aug 12, 2025	GM1 gangliosidosis type 2; GM1 gangliosidosis type 3; Infantile GM1 gangliosidosis; GM1 gangliosidosis; Mucopolysaccharidosis; MPS-IV-B; MPS-IV-B;GM1 gangliosidosis; GLB1-related disorder; GM1 gangliosidosis type 3;Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis type 2;Infantile GM1 gangliosidosis	<a href="#">7.18e-05</a>	ClinVar; LOVD; gnomAD; NTSAD; Caciotti 2003 (PMID:12644936); Hofer 2009 (PMID:19472408); Cure GM1 AllStripes PIN	<a href="#">11511921</a> ; <a href="#">12644936</a> ; <a href="#">16617000</a> ; <a href="#">16626397</a> ; <a href="#">17309651</a> ; <a href="#">1907800</a> ; <a href="#">1909089</a> ; <a href="#">19472408</a> ; <a href="#">20175788</a> ; <a href="#">21520340</a> ; <a href="#">23430499</a> ; <a href="#">25443580</a> ; <a href="#">25557439</a> ; <a href="#">25600812</a> ; <a href="#">26646981</a> ; <a href="#">28554332</a> ; <a href="#">28716012</a> ; <a href="#">29439846</a> ; <a href="#">31367523</a> ; <a href="#">8068159</a> ; <a href="#">8112731</a> ; <a href="#">9203065</a>	8	<a href="#">925</a>
c.602G>A	p.Arg201His	Exon 6	P/LP	Jan 15, 2026	GM1 gangliosidosis; Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis type 2; MPS-IV-B;GM1 gangliosidosis; Infantile GM1 gangliosidosis; GM1 gangliosidosis type 2;Mucopolysaccharidosis; MPS-IV-B;Infantile GM1 gangliosidosis;GM1 gangliosidosis type 3	<a href="#">3.49e-05</a>	ClinVar; LOVD; gnomAD; OMIM; NTSAD; Santamaria 2006 (PMID:16941474); Hofer 2009 (PMID:19472408); Cure GM1 AllStripes PIN	<a href="#">10737981</a> ; <a href="#">11504597</a> ; <a href="#">11511921</a> ; <a href="#">16314480</a> ; <a href="#">16538002</a> ; <a href="#">16617000</a> ; <a href="#">16941474</a> ; <a href="#">17309651</a> ; <a href="#">17664528</a> ; <a href="#">19472408</a> ; <a href="#">20175788</a> ; <a href="#">21520340</a> ; <a href="#">22675082</a> ; <a href="#">23430499</a> ; <a href="#">24033266</a> ; <a href="#">32779865</a> ; <a href="#">33737400</a> ; <a href="#">9203065</a>	8	<a href="#">198077</a>
c.606C>T	p.Phe202=	Exon 6	Likely Benign	Nov 01, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">6.84e-07</a>	ClinVar; gnomAD			<a href="#">2928233</a>
c.616C>T	p.Arg206Cys	Exon 6	VUS	Apr 23, 2022	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B; Mucopolysaccharidosis; GM1 gangliosidosis	<a href="#">7.39e-05</a>	ClinVar; gnomAD			<a href="#">902394</a>
c.617G>A	p.Arg206His	Exon 6	VUS	Aug 02, 2022	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">6.16e-06</a>	ClinVar; gnomAD			<a href="#">2163947</a>
c.621T>C	p.Phe207=	Exon 6	Likely Benign	Oct 06, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">2034347</a>
c.622C>T	p.Arg208Cys	Exon 6	Pathogenic	Jan 29, 2026	Infantile GM1 gangliosidosis; GM1 gangliosidosis type 2; GM1 gangliosidosis type 3; Mucopolysaccharidosis; MPS-IV-B; MPS-IV-B;GM1 gangliosidosis; GM1 gangliosidosis; GM1 gangliosidosis type 3;Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis type 2;Infantile GM1 gangliosidosis; Inborn genetic diseases	<a href="#">1.44e-05</a>	ClinVar; LOVD; gnomAD; OMIM; NTSAD; Caciotti 2005 (PMID:15714521); Cure GM1 AllStripes PIN	<a href="#">10338095</a> ; <a href="#">15714521</a> ; <a href="#">17309651</a> ; <a href="#">20175788</a> ; <a href="#">22128166</a> ; <a href="#">23337983</a> ; <a href="#">31761138</a> ; <a href="#">8213816</a> ; <a href="#">8652017</a>	3	<a href="#">939</a>

cDNA Change	Protein Change	Location	Pathogenicity	Classified	Phenotype	gnomAD AF	Sources	PMID(s)	PIN	ClinVar ID
c.623G>A	p.Arg208His	Exon 6	Conflicting	Jan 19, 2026	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis; Mucopolysaccharidosis	<a href="#">0.00105</a>	ClinVar; LOVD; gnomAD; Caciotti 2005 (PMID:15714521)	<a href="#">15714521</a>		<a href="#">562201</a>
c.623G>C		Exon 6	Not Classified				LOVD			
c.623G>T	p.Arg208Leu	Exon 6	VUS	Jun 29, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">6.84e-07</a>	ClinVar; gnomAD; Caciotti 2005 (PMID:15714521)	<a href="#">10338095</a> ; <a href="#">15714521</a> ; <a href="#">17309651</a> ; <a href="#">23337983</a> ; <a href="#">8213816</a>		<a href="#">2060699</a>
c.624C>T	p.Arg208=	Exon 6	Likely Benign	Mar 19, 2024	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">1.37e-06</a>	ClinVar; gnomAD			<a href="#">1573972</a>
c.625C>T	p.His209Tyr	Exon 6	VUS	Nov 14, 2021	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">1443336</a>
c.626del	p.His209fs	Exon 6	Likely Pathogenic	Apr 05, 2018	Infantile GM1 gangliosidosis;GM1 gangliosidosis type 2;Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis type 3		ClinVar			<a href="#">557667</a>
c.627C>T	p.His209=	Exon 6	Likely Benign	Sep 14, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">2135448</a>
c.631C>T	p.Leu211=	Exon 6	Likely Benign	Sep 19, 2023	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">6.84e-07</a>	ClinVar; gnomAD			<a href="#">2951745</a>
c.632T>G		Exon 6	Not Classified				LOVD			
c.633G>A	p.Leu211=	Exon 6	Likely Benign	Mar 29, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">2935478</a>
c.636G>A	p.Gly212=	Exon 6	Likely Benign	Feb 08, 2024	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">2.05e-06</a>	ClinVar; gnomAD			<a href="#">1624700</a>
c.639T>C	p.Asp213=	Exon 6	Likely Benign	Jun 23, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">1.37e-06</a>	ClinVar; gnomAD			<a href="#">2009680</a>
c.645_648del	p.Val216fs	Exon 6	Likely Pathogenic	Feb 23, 2024	GM1 gangliosidosis type 3;Mucopolysaccharidosis; MPS-IV-B;Infantile GM1 gangliosidosis;GM1 gangliosidosis type 2; Mucopolysaccharidosis; MPS-IV-B		ClinVar; LOVD			<a href="#">554283</a>
c.647T>C	p.Val216Ala	Exon 6	VUS	Oct 21, 2015			ClinVar			<a href="#">284171</a>
c.659dup	p.Thr220_Asp221insTer	Exon 6	Pathogenic	Aug 07, 2024	Mucopolysaccharidosis; MPS-IV-B	<a href="#">6.84e-07</a>	ClinVar; gnomAD			<a href="#">3366437</a>
c.666A>G	p.Gly222=	Exon 6	Likely Benign	May 14, 2024	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">1910899</a>
c.668C>G	p.Ala223Gly	Exon 6	VUS	Apr 11, 2025	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B; Inborn genetic diseases	<a href="#">7.52e-06</a>	ClinVar; gnomAD			<a href="#">3748336</a>
c.669A>G	p.Ala223=	Exon 6	Likely Benign	Feb 23, 2024	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">6.84e-07</a>	ClinVar; gnomAD			<a href="#">1079895</a>
c.670C>G	p.His224Asp	Exon 6	VUS	Nov 04, 2024	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">1981116</a>
c.672_673del		Exon 6	Not Classified				LOVD			
c.672_673delAT		Exon 6	Pathogenic	-	Infantile GM1 gangliosidosis		ClinVar			<a href="#">100726</a>
c.677del	p.Thr226fs	Exon 6	Pathogenic	Jul 22, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar	<a href="#">18524657</a>		<a href="#">2018917</a>

cDNA Change	Protein Change	Location	Pathogenicity	Classified	Phenotype	gnomAD AF	Sources	PMID(s)	PIN	ClinVar ID
c.678A>G	p.Thr226=	Exon 6	Likely Benign	Mar 20, 2024	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">1.71e-05</a>	ClinVar; gnomAD			<a href="#">1084431</a>
c.678A>T	p.Thr226=	Exon 6	Likely Benign	Mar 16, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">6.84e-07</a>	ClinVar; gnomAD			<a href="#">1976491</a>
c.681C>T	p.Phe227=	Exon 6	Likely Benign	Jul 29, 2025	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis; GLB1-related disorder	<a href="#">1.50e-05</a>	ClinVar; gnomAD			<a href="#">1159142</a>
c.689G>A	p.Cys230Tyr	Exon 6	Likely Pathogenic	Aug 24, 2024	Mucopolysaccharidosis; MPS-IV-B		ClinVar	<a href="#">16314480</a>		<a href="#">4818323</a>
c.693G>A	p.Gly231=	Exon 6	Likely Benign	Oct 27, 2024	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">6.84e-07</a>	ClinVar; gnomAD			<a href="#">1937289</a>
c.693G>C	p.Gly231=	Exon 6	Likely Benign	Jul 07, 2025	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">4.10e-06</a>	ClinVar; gnomAD			<a href="#">726678</a>
c.693G>T	p.Gly231=	Exon 6	Likely Benign	Mar 25, 2024	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">3.42e-06</a>	ClinVar; gnomAD			<a href="#">1128507</a>
c.694dup	p.Ala232fs	Exon 6	P/LP	Oct 02, 2025	GM1 gangliosidosis type 3;Mucopolysaccharidosis; MPS-IV-B;Infantile GM1 gangliosidosis;GM1 gangliosidosis type 2; Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis; MPS-IV-B		ClinVar	<a href="#">18524657</a>		<a href="#">552535</a>
c.696C>T	p.Ala232=	Exon 6	Likely Benign	Mar 27, 2023	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">6.84e-07</a>	ClinVar; gnomAD			<a href="#">2945891</a>
c.697C>A	p.Leu233Met	Exon 6	VUS	May 30, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">1.30e-05</a>	ClinVar; gnomAD			<a href="#">2082728</a>
c.699G>T	p.Leu233=	Exon 6	Likely Benign	May 08, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">3.42e-06</a>	ClinVar; gnomAD			<a href="#">1911712</a>
c.699del	p.Gln234fs	Exon 6	P/LP	Jan 19, 2026	GM1 gangliosidosis; GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis type 2;Infantile GM1 gangliosidosis;GM1 gangliosidosis type 3;Mucopolysaccharidosis; Mucopolysaccharidosis	<a href="#">2.74e-06</a>	ClinVar; LOVD; gnomAD	<a href="#">18524657</a> ; <a href="#">28476546</a>		<a href="#">928700</a>
c.703G>A	p.Gly235Ser	Exon 6	VUS	Nov 01, 2022	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">2.05e-06</a>	ClinVar; gnomAD			<a href="#">1419496</a>
c.708C>T	p.Leu236=	Exon 6	Likely Benign	Oct 27, 2020	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">1146191</a>
c.710A>G	p.Tyr237Cys	Exon 6	P/LP	Nov 24, 2025	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis; GM1 gangliosidosis type 2;Mucopolysaccharidosis; MPS-IV-B;Infantile GM1 gangliosidosis;GM1 gangliosidosis type 3		ClinVar	<a href="#">30267299</a>		<a href="#">2927179</a>
c.711C>T	p.Tyr237=	Exon 6	Conflicting	Dec 14, 2025	GM1 gangliosidosis; Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis;Mucopolysaccharidosis	<a href="#">6.84e-06</a>	ClinVar; gnomAD			<a href="#">900563</a>
c.712A>G	p.Thr238Ala	Exon 6	VUS	Jun 02, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">4.79e-06</a>	ClinVar; gnomAD			<a href="#">1502493</a>
c.713C>T	p.Thr238Ile	Exon 6	VUS	-	GM1 gangliosidosis type 2		ClinVar			<a href="#">2429471</a>

cDNA Change	Protein Change	Location	Pathogenicity	Classified	Phenotype	gnomAD AF	Sources	PMID(s)	PIN	ClinVar ID
c.716C>T	p.Thr239Met	Exon 6	P/LP	Feb 26, 2025	GM1 gangliosidosis type 2; Infantile GM1 gangliosidosis; Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis; GM1 gangliosidosis type 2; GM1 gangliosidosis type 3; Infantile GM1 gangliosidosis; Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis; Lysosomal storage disease	<a href="#">1.03e-05</a>	ClinVar; LOVD; gnomAD; Caciotti 2005 (PMID:15714521); Hofer 2009 (PMID:19472408)	<a href="#">15714521</a> ; <a href="#">19472408</a> ; <a href="#">33737400</a>		<a href="#">807421</a>
c.717G>A		Exon 6	Not Classified			1.85e-05	LOVD; gnomAD			
c.717G>T	p.Thr239=	Exon 6	Likely Benign	Mar 25, 2024	Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis		ClinVar			<a href="#">1080440</a>
c.718G>A	p.Val240Met	Exon 6	Pathogenic	Feb 25, 2024	GM1 gangliosidosis; Mucopolysaccharidosis; MPS-IV-B		ClinVar	<a href="#">10338095</a>		<a href="#">3759083</a>
c.729A>G	p.Gly243=	Exon 6	Likely Benign	Sep 19, 2023	GM1 gangliosidosis; Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">2003529</a>
c.733+1G>A		Intron 6	Pathogenic	Jan 14, 2026	GM1 gangliosidosis type 3; Mucopolysaccharidosis; MPS-IV-B; Infantile GM1 gangliosidosis; GM1 gangliosidosis type 2; Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis; MPS-IV-B	<a href="#">4.04e-05</a>	ClinVar; gnomAD; Caciotti 2005 (PMID:15714521)	<a href="#">15714521</a> ; <a href="#">16199547</a> ; <a href="#">18524657</a>		<a href="#">553125</a>
c.733+2T>C		Intron 6	P/LP	Oct 21, 2025	Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis; Mucopolysaccharidosis		ClinVar; Caciotti 2005 (PMID:15714521)	<a href="#">15714521</a> ; <a href="#">16199547</a> ; <a href="#">18524657</a> ; <a href="#">31905715</a>		<a href="#">987846</a>
c.733+2_733+4del		Intron 6	Likely Pathogenic	Apr 13, 2018	Infantile GM1 gangliosidosis; GM1 gangliosidosis type 2; Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis type 3		ClinVar			<a href="#">557859</a>
c.733+6T>C		Intron 6	Pathogenic	Feb 02, 2022	GM1 gangliosidosis		ClinVar			<a href="#">2500729</a>
c.733+7G>A		Intron 6	Likely Benign	May 27, 2021	Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis		ClinVar			<a href="#">1621642</a>
c.733+10T>C		Intron 6	Likely Benign	Aug 02, 2023	GM1 gangliosidosis; Mucopolysaccharidosis; MPS-IV-B	<a href="#">6.85e-07</a>	ClinVar; gnomAD			<a href="#">2946968</a>
c.733+11G>A		Intron 6	Likely Benign	Dec 23, 2022	Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis	<a href="#">2.05e-06</a>	ClinVar; gnomAD			<a href="#">2932241</a>
c.733+13A>T		Intron 6	Likely Benign	Jun 23, 2022	Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis	<a href="#">1.37e-06</a>	ClinVar; gnomAD			<a href="#">2018036</a>
c.733+20G>A		Intron 6	Likely Benign	Dec 14, 2023	Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis		ClinVar			<a href="#">2921414</a>
c.733+42T>A		Intron 6	Likely Benign	-		<a href="#">2.06e-06</a>	ClinVar; gnomAD			<a href="#">256025</a>
c.733+78A>G		Intron 6	Likely Benign	Jun 15, 2021	Infantile GM1 gangliosidosis; Mucopolysaccharidosis; MPS-IV-B; Chronic lymphocytic leukemia/small lymphocytic lymphoma; Familial pancreatic carcinoma; Ovarian cancer; Malignant lymphoma; large B-cell; diffuse; Uterine carcinosarcoma; Nonpapillary renal cell carcinoma; Germ cell tumor of testis		ClinVar			<a href="#">1174057</a>
c.734-245G>T		Intron 6	Not Classified	-	Ovarian serous cystadenocarcinoma		ClinVar			<a href="#">4295485</a>

cDNA Change	Protein Change	Location	Pathogenicity	Classified	Phenotype	gnomAD AF	Sources	PMID(s)	PIN	ClinVar ID
c.734-116del		Intron 6	Likely Benign	Apr 20, 2019			ClinVar			<a href="#">1193900</a>
c.734-112T>C		Intron 6	Benign	Jun 15, 2021	Mucopolysaccharidosis; MPS-IV-B; Infantile GM1 gangliosidosis		ClinVar			<a href="#">1174056</a>
c.734-99G>T		Intron 6	Benign	Jun 15, 2021	Mucopolysaccharidosis; MPS-IV-B; Infantile GM1 gangliosidosis; Cholangiocarcinoma; Acute myeloid leukemia; Malignant lymphoma; large B-cell; diffuse; Thymoma		ClinVar			<a href="#">1174055</a>
c.734-80G>A		Intron 6	Likely Benign	Jun 15, 2021	Infantile GM1 gangliosidosis; Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">1174054</a>
c.734-17C>T		Intron 6	Likely Benign	Jan 18, 2026	Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis	<a href="#">1.37e-06</a>	ClinVar; gnomAD			<a href="#">2926247</a>
c.734-16C>T		Intron 6	Likely Benign	Aug 31, 2023	Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis	<a href="#">4.79e-06</a>	ClinVar; gnomAD			<a href="#">2934534</a>
c.734-14C>G		Intron 6	Likely Benign	Jul 19, 2023	GM1 gangliosidosis; Mucopolysaccharidosis; MPS-IV-B	<a href="#">1.97e-05</a>	ClinVar; gnomAD			<a href="#">2944891</a>
c.734-14C>T		Intron 6	Likely Benign	Jan 27, 2024	Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis	<a href="#">1.97e-05</a>	ClinVar; gnomAD			<a href="#">2921859</a>
c.734-11A>G		Intron 6	Likely Benign	Jan 03, 2024	Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis	<a href="#">2.05e-06</a>	ClinVar; gnomAD			<a href="#">2931019</a>
c.734-11dup		Intron 6	Likely Benign	Aug 29, 2023	Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis		ClinVar			<a href="#">2935815</a>
c.734-10C>A		Intron 6	Likely Benign	Jan 01, 2026	Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis	<a href="#">1.30e-05</a>	ClinVar; gnomAD			<a href="#">1114125</a>
c.734-10C>G		Intron 6	VUS	Jul 03, 2025	GM1 gangliosidosis; Mucopolysaccharidosis; MPS-IV-B	<a href="#">1.37e-06</a>	ClinVar; gnomAD	<a href="#">37645600</a>		<a href="#">4783619</a>
c.734-10C>T		Intron 6	Likely Benign	Jul 13, 2023	GM1 gangliosidosis; Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">2946897</a>
c.734-8A>G		Intron 6	Conflicting	Oct 08, 2025	GM1 gangliosidosis type 2; Infantile GM1 gangliosidosis; Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis type 3; GM1 gangliosidosis; Lung cancer; Mucopolysaccharidosis; MPS-IV-B	<a href="#">8.21e-06</a>	ClinVar; gnomAD	<a href="#">20175788</a> ; <a href="#">30408610</a> ; <a href="#">35614200</a> ; <a href="#">38730490</a>		<a href="#">92912</a>
c.734-6T>A		Intron 6	Conflicting	Aug 02, 2025	GM1 gangliosidosis; Mucopolysaccharidosis; MPS-IV-B	<a href="#">6.57e-06</a>	ClinVar; gnomAD			<a href="#">1518783</a>
c.734-2A>G		Intron 6	Pathogenic	Feb 24, 2024	GM1 gangliosidosis; Mucopolysaccharidosis; MPS-IV-B		ClinVar	<a href="#">16199547</a> ; <a href="#">18524657</a>		<a href="#">946866</a>
c.739A>G	p.Asn247Asp	Exon 7	VUS	-	Infantile GM1 gangliosidosis	<a href="#">2.05e-06</a>	ClinVar; gnomAD			<a href="#">2585573</a>
c.747A>T	p.Thr249=	Exon 7	Likely Benign	Dec 12, 2022	GM1 gangliosidosis; Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">2942233</a>
c.750T>C	p.Asp250=	Exon 7	Likely Benign	Jan 12, 2023	GM1 gangliosidosis; Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">2943131</a>
c.751G>A	p.Ala251Thr	Exon 7	VUS	Jul 25, 2022	GM1 gangliosidosis; Mucopolysaccharidosis; MPS-IV-B	<a href="#">1.37e-06</a>	ClinVar; gnomAD			<a href="#">1350099</a>
c.756C>T	p.Phe252=	Exon 7	Likely Benign	Feb 02, 2026	GM1 gangliosidosis; Mucopolysaccharidosis; MPS-IV-B; Mucopolysaccharidosis; GM1 gangliosidosis	<a href="#">0.00095</a>	ClinVar; gnomAD			<a href="#">256026</a>

cDNA Change	Protein Change	Location	Pathogenicity	Classified	Phenotype	gnomAD AF	Sources	PMID(s)	PIN	ClinVar ID
c.765G>C	p.Gln255His	Exon 7	Likely Pathogenic	Sep 09, 2025	Infantile GM1 gangliosidosis;GM1 gangliosidosis type 2;Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis type 3; GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B; Mucopolysaccharidosis	<a href="#">1.37e-06</a>	ClinVar; gnomAD; Hofer 2009 (PMID:19472408); Cure GM1 AllStripes PIN	<a href="#">16617000</a> ; <a href="#">19472408</a> ; <a href="#">21520340</a> ; <a href="#">22128166</a> ; <a href="#">23337983</a> ; <a href="#">25600812</a> ; <a href="#">27750150</a> ; <a href="#">31720227</a> ; <a href="#">37871851</a>	1	<a href="#">554850</a>
c.765G>T	p.Gln255His	Exon 7	Likely Pathogenic	Dec 08, 2023	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar; Hofer 2009 (PMID:19472408)	<a href="#">16617000</a> ; <a href="#">19472408</a> ; <a href="#">25600812</a>		<a href="#">2946183</a>
c.768G>A	p.Arg256=	Exon 7	Likely Benign	Nov 29, 2022	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">6.84e-07</a>	ClinVar; gnomAD			<a href="#">2101395</a>
c.769_792+13del		Exon 7	P/LP	Jun 19, 2025	Infantile GM1 gangliosidosis;GM1 gangliosidosis type 2;Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis type 3; GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">6.84e-06</a>	ClinVar; gnomAD; Bidchol 2015 (PMID:25936995)	<a href="#">16199547</a> ; <a href="#">18524657</a> ; <a href="#">25936995</a>		<a href="#">556646</a>
c.771G>A	p.Lys257=	Exon 7	Likely Benign	Jan 24, 2023	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">6.84e-07</a>	ClinVar; gnomAD			<a href="#">2943575</a>
c.773G>A	p.Cys258Tyr	Exon 7	VUS	Nov 26, 2025	Inborn genetic diseases		ClinVar			<a href="#">4620831</a>
c.774T>A	p.Cys258Ter	Exon 7	Pathogenic	Apr 30, 2022	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar	<a href="#">18524657</a>		<a href="#">2132074</a>
c.775G>T	p.Glu259Ter	Exon 7	Likely Pathogenic	Jan 01, 2020	GM1 gangliosidosis type 2;Infantile GM1 gangliosidosis;GM1 gangliosidosis type 3		ClinVar			<a href="#">983743</a>
c.777G>A	p.Glu259=	Exon 7	Likely Benign	Dec 16, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">9.58e-06</a>	ClinVar; gnomAD			<a href="#">755658</a>
c.779C>A	p.Pro260His	Exon 7	VUS	Aug 15, 2022	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">2.74e-06</a>	ClinVar; gnomAD			<a href="#">1417955</a>
c.780C>G	p.Pro260=	Exon 7	Likely Benign	Jan 31, 2026	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">3.83e-05</a>	ClinVar; gnomAD			<a href="#">752013</a>
c.780C>T	p.Pro260=	Exon 7	Likely Benign	Nov 24, 2022	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">2941817</a>
c.782A>G	p.Lys261Arg	Exon 7	Conflicting	Jan 27, 2026	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis; GLB1-related disorder	<a href="#">2.53e-05</a>	ClinVar; gnomAD			<a href="#">1105606</a>
c.785G>A	p.Gly262Glu	Exon 7	P/LP	Jan 29, 2026	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis; GM1 gangliosidosis; MPS-IV-B	<a href="#">4.79e-06</a>	ClinVar; gnomAD; Bidchol 2015 (PMID:25936995)	<a href="#">25936995</a> ; <a href="#">38313286</a>		<a href="#">2203326</a>
c.785G>C	p.Gly262Ala	Exon 7	P/LP	Jun 20, 2025	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B; Mucopolysaccharidosis	<a href="#">1.31e-05</a>	ClinVar; gnomAD; Bidchol 2015 (PMID:25936995)	<a href="#">25936995</a>		<a href="#">933199</a>
c.785G>T	p.Gly262Val	Exon 7	P/LP	Oct 23, 2024	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis; MPS-IV-B		ClinVar	<a href="#">22675082</a> ; <a href="#">26646981</a>		<a href="#">2203325</a>
c.786A>C	p.Gly262=	Exon 7	Likely Benign	Oct 08, 2023	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">2943811</a>
c.787C>T	p.Pro263Ser	Exon 7	Likely Pathogenic	Jan 31, 2025	GM1 gangliosidosis	<a href="#">1.37e-06</a>	ClinVar; LOVD; gnomAD	<a href="#">10571006</a> ; <a href="#">21520340</a> ; <a href="#">23337983</a>		<a href="#">3769334</a>
c.789C>G	p.Pro263=	Exon 7	Likely Benign	Mar 28, 2019	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">1133188</a>
c.789C>T	p.Pro263=	Exon 7	Likely Benign	Aug 30, 2021	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">6.84e-07</a>	ClinVar; gnomAD			<a href="#">1601931</a>

cDNA Change	Protein Change	Location	Pathogenicity	Classified	Phenotype	gnomAD AF	Sources	PMID(s)	PIN	ClinVar ID
c.792+9C>A		Intron 7	Likely Benign	Oct 27, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">6.84e-07</a>	ClinVar; gnomAD			<a href="#">1152039</a>
c.792+9C>T		Intron 7	Likely Benign	Aug 27, 2025	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">4.10e-06</a>	ClinVar; gnomAD			<a href="#">2171397</a>
c.792+10G>A		Intron 7	Likely Benign	Jan 13, 2026	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis; GLB1-related disorder	<a href="#">0.00016</a>	ClinVar; LOVD; gnomAD			<a href="#">1151063</a>
c.792+10G>C		Intron 7	Likely Benign	Feb 24, 2025	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">1144894</a>
c.792+10G>T		Intron 7	Likely Benign	Feb 04, 2026	Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis; Infantile GM1 gangliosidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">0.03530</a>	ClinVar; gnomAD			<a href="#">92914</a>
c.792+16G>C		Intron 7	Likely Benign	Nov 17, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">6.84e-07</a>	ClinVar; gnomAD			<a href="#">2930473</a>
c.792+18G>A		Intron 7	Benign	Jan 28, 2026	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">0.00040</a>	ClinVar; gnomAD			<a href="#">1166108</a>
c.792+18G>C		Intron 7	Likely Benign	Jul 17, 2024	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">6.84e-07</a>	ClinVar; gnomAD			<a href="#">3763162</a>
c.792+19G>A		Intron 7	Likely Benign	Dec 13, 2025	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">4.38e-05</a>	ClinVar; gnomAD			<a href="#">1607480</a>
c.792+187C>G		Intron 7	Likely Benign	Apr 12, 2019			ClinVar			<a href="#">1189208</a>
c.792+231G>A		Intron 7	Benign	Aug 28, 2018			ClinVar			<a href="#">1282223</a>
c.792+250G>C		Intron 7	Benign	Aug 28, 2018			ClinVar			<a href="#">1263301</a>
c.793-246A>G		Intron 7	Likely Benign	Apr 20, 2019			ClinVar			<a href="#">1199113</a>
c.793-185C>G		Intron 7	Benign	Oct 17, 2018			ClinVar			<a href="#">1231856</a>
c.793-132T>G		Intron 7	Benign	Jun 19, 2018			ClinVar			<a href="#">678118</a>
c.793-69T>C		Intron 7	Likely Benign	Nov 13, 2019		<a href="#">0.00096</a>	ClinVar; gnomAD			<a href="#">1216855</a>
c.793-20A>G		Intron 7	Likely Benign	Feb 17, 2025	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">1532292</a>
c.793-19C>T		Intron 7	Benign	Jan 28, 2026	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">0.00061</a>	ClinVar; LOVD; gnomAD			<a href="#">558921</a>
c.793-18G>A		Intron 7	Likely Benign	Nov 04, 2024	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">2.12e-05</a>	ClinVar; gnomAD			<a href="#">1916741</a>
c.793-14dup		Intron 7	Benign	Jul 14, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">1166391</a>
c.793-13T>G		Intron 7	Likely Benign	Nov 06, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">3.42e-06</a>	ClinVar; gnomAD			<a href="#">2933855</a>
c.793-9T>A		Intron 7	Likely Benign	Mar 31, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">6.16e-06</a>	ClinVar; gnomAD			<a href="#">1665182</a>
c.797A>G	p.Asn266Ser	Exon 8	VUS	Dec 05, 2017	Infantile GM1 gangliosidosis;GM1 gangliosidosis type 2;Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis type 3	<a href="#">1.37e-06</a>	ClinVar; gnomAD	<a href="#">21520340</a> ; <a href="#">22128166</a> ; <a href="#">23337983</a> ; <a href="#">9203065</a>		<a href="#">555028</a>
c.802G>C	p.Glu268Gln	Exon 8	Pathogenic	Oct 13, 2023	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">2.74e-06</a>	ClinVar; gnomAD			<a href="#">2089605</a>
c.804A>G	p.Glu268=	Exon 8	Likely Benign	Jan 08, 2026	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">6.84e-07</a>	ClinVar; gnomAD			<a href="#">2042744</a>

cDNA Change	Protein Change	Location	Pathogenicity	Classified	Phenotype	gnomAD AF	Sources	PMID(s)	PIN	ClinVar ID
c.806T>C	p.Phe269Ser	Exon 8	VUS	Jun 28, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">2013625</a>
c.808T>C	p.Tyr270His	Exon 8	Not Classified	-	Infantile GM1 gangliosidosis		ClinVar			<a href="#">1330373</a>
c.808T>G	p.Tyr270Asp	Exon 8	P/LP	Dec 21, 2025	Inborn genetic diseases; Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis; GM1 gangliosidosis type 2;Infantile GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis type 3; MPS-IV-B; GLB1-related disorder	<a href="#">1.98e-05</a>	ClinVar; LOVD; gnomAD; Hofer 2009 (PMID:19472408); Cure GM1 AllStripes PIN	<a href="#">11511921</a> ; <a href="#">19472408</a> ; <a href="#">21520340</a>	1	<a href="#">284172</a>
c.809A>C	p.Tyr270Ser	Exon 8	VUS	Oct 27, 2014			ClinVar	<a href="#">23151865</a>		<a href="#">198727</a>
c.809A>G	p.Tyr270Cys	Exon 8	Conflicting	Jun 10, 2025	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis; GM1 gangliosidosis	<a href="#">6.16e-06</a>	ClinVar; LOVD; gnomAD; Hofer 2009 (PMID:19472408)	<a href="#">11511921</a> ; <a href="#">19472408</a> ; <a href="#">21520340</a>		<a href="#">198726</a>
c.813T>C	p.Thr271=	Exon 8	Likely Benign	Sep 01, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">2.05e-06</a>	ClinVar; gnomAD			<a href="#">1154879</a>
c.815G>T	p.Gly272Val	Exon 8	Likely Pathogenic	Oct 23, 2025	GM1 gangliosidosis type 2		ClinVar; Santamaria 2006 (PMID:16941474)	<a href="#">16941474</a>		<a href="#">4819894</a>
c.817_818delinsCT	p.Trp273Leu	Exon 8	P/LP	Jan 28, 2026	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis; MPS-IV-B; Infantile GM1 gangliosidosis; MPS-IV-B;Infantile GM1 gangliosidosis;GM1 gangliosidosis type 3;GM1 gangliosidosis type 2		ClinVar; LOVD; Hofer 2009 (PMID:19472408)	<a href="#">11511921</a> ; <a href="#">18546276</a> ; <a href="#">1928092</a> ; <a href="#">19472408</a> ; <a href="#">21497194</a> ; <a href="#">30809705</a> ; <a href="#">33266180</a> ; <a href="#">33737400</a>		<a href="#">568792</a>
c.817_818inv		Exon 8	Not Classified				LOVD			
c.818G>T	p.Trp273Leu	Exon 8	Likely Pathogenic	Feb 01, 2024	Mucopolysaccharidosis; MPS-IV-B; Infantile GM1 gangliosidosis	<a href="#">3.56e-05</a>	ClinVar; LOVD; gnomAD	<a href="#">11511921</a> ; <a href="#">1928092</a>		<a href="#">931</a>
c.819G>A	p.Trp273Ter	Exon 8	P/LP	Sep 24, 2025	GM1 gangliosidosis type 3;GM1 gangliosidosis type 2;Infantile GM1 gangliosidosis; GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis type 3;GM1 gangliosidosis type 2;Infantile GM1 gangliosidosis;Mucopolysaccharidosis	<a href="#">1.37e-06</a>	ClinVar; gnomAD	<a href="#">18524657</a> ; <a href="#">20175788</a>		<a href="#">983742</a>
c.819G>T	p.Trp273Cys	Exon 8	VUS	May 12, 2025			ClinVar			<a href="#">3902324</a>
c.821T>C	p.Leu274Pro	Exon 8	VUS	Mar 29, 2024	Mucopolysaccharidosis; MPS-IV-B; Infantile GM1 gangliosidosis; GM1 gangliosidosis type 2; GM1 gangliosidosis type 3	<a href="#">6.84e-07</a>	ClinVar; gnomAD			<a href="#">1210418</a>
c.825T>A	p.Asp275Glu	Exon 8	VUS	Sep 22, 2025	Inborn genetic diseases		ClinVar			<a href="#">2662239</a>
c.825T>C	p.Asp275=	Exon 8	Likely Benign	Mar 30, 2024	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">3762395</a>
c.827A>C	p.His276Pro	Exon 8	Likely Pathogenic	Jan 21, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">663086</a>
c.829T>C		Exon 8	Not Classified				LOVD			

cDNA Change	Protein Change	Location	Pathogenicity	Classified	Phenotype	gnomAD AF	Sources	PMID(s)	PIN	ClinVar ID
c.833del	p.Gly278fs	Exon 8	Pathogenic	Dec 13, 2025	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis; Infantile GM1 gangliosidosis;GM1 gangliosidosis type 2;Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis type 3	<a href="#">6.84e-07</a>	ClinVar; gnomAD	<a href="#">18524657</a>		<a href="#">2043978</a>
c.835C>T	p.Gln279Ter	Exon 8	Pathogenic	Dec 27, 2024	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar	<a href="#">18524657</a>		<a href="#">3760049</a>
c.835del	p.Gln279fs	Exon 8	Likely Pathogenic	Nov 11, 2021	Infantile GM1 gangliosidosis;GM1 gangliosidosis type 3;GM1 gangliosidosis type 2	<a href="#">6.84e-07</a>	ClinVar; gnomAD			<a href="#">1724395</a>
c.836A>G	p.Gln279Arg	Exon 8	VUS	Aug 31, 2021	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">4.79e-06</a>	ClinVar; gnomAD			<a href="#">1346804</a>
c.837A>C	p.Gln279His	Exon 8	VUS	May 30, 2022	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">2001182</a>
c.837A>G	p.Gln279=	Exon 8	Likely Benign	Mar 06, 2021	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">6.84e-07</a>	ClinVar; gnomAD			<a href="#">1577932</a>
c.838C>G	p.Pro280Ala	Exon 8	VUS	Feb 17, 2024	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">1717336</a>
c.838C>T	p.Pro280Ser	Exon 8	VUS	Sep 27, 2022	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">2161836</a>
c.841C>T	p.His281Tyr	Exon 8	Pathogenic	Oct 22, 2025	Mucopolysaccharidosis; MPS-IV-B; MPS-IV-B;GM1 gangliosidosis; GLB1-related disorder; Infantile GM1 gangliosidosis;Mucopolysaccharidosis; Lysosomal storage disease	<a href="#">1.78e-05</a>	ClinVar; LOVD; gnomAD; Caciotti 2005 (PMID:15714521); Cure GM1 AllStripes PIN	<a href="#">11511921</a> ; <a href="#">15714521</a> ; <a href="#">18546276</a> ; <a href="#">21214877</a> ; <a href="#">21497194</a> ; <a href="#">22128166</a> ; <a href="#">28476546</a> ; <a href="#">31761138</a>	2	<a href="#">372371</a>
c.842A>G	p.His281Arg	Exon 8	Conflicting	Apr 18, 2025	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">2.74e-06</a>	ClinVar; gnomAD	<a href="#">11511921</a> ; <a href="#">18546276</a> ; <a href="#">21214877</a> ; <a href="#">21497194</a>		<a href="#">2173809</a>
c.843C>T	p.His281=	Exon 8	Likely Benign	Apr 20, 2024	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">2.05e-06</a>	ClinVar; gnomAD			<a href="#">1547054</a>
c.845C>T	p.Ser282Phe	Exon 8	VUS	Aug 27, 2021	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">3.42e-06</a>	ClinVar; gnomAD			<a href="#">1039218</a>
c.846del	p.Thr283fs	Exon 8	Pathogenic	Dec 30, 2025	Infantile GM1 gangliosidosis;GM1 gangliosidosis type 2;Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis type 3; GM1 gangliosidosis type 2; GM1 gangliosidosis; GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B; Mucopolysaccharidosis		ClinVar	<a href="#">17309651</a> ; <a href="#">18524657</a> ; <a href="#">21214877</a>		<a href="#">557052</a>
c.849A>C	p.Thr283=	Exon 8	Likely Benign	Feb 23, 2025	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">4.45e-05</a>	ClinVar; gnomAD			<a href="#">766435</a>
c.850A>G	p.Ile284Val	Exon 8	VUS	Jun 13, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">2.74e-06</a>	ClinVar; gnomAD			<a href="#">1421788</a>
c.856_862del		Exon 8	Not Classified				LOVD			
c.858C>T	p.Thr286=	Exon 8	Likely Benign	May 29, 2025	GM1 gangliosidosis type 2;Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis type 3;Infantile GM1 gangliosidosis; GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">2.67e-05</a>	ClinVar; gnomAD	<a href="#">21497194</a>		<a href="#">256027</a>

cDNA Change	Protein Change	Location	Pathogenicity	Classified	Phenotype	gnomAD AF	Sources	PMID(s)	PIN	ClinVar ID
c.859G>A	p.Glu287Lys	Exon 8	VUS	Aug 10, 2022	Intellectual disability; GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">4.79e-06</a>	ClinVar; gnomAD	<a href="#">21956720</a> ; <a href="#">25157020</a> ; <a href="#">34131312</a> ; <a href="#">34211152</a>		<a href="#">975401</a>
c.859G>T	p.Glu287Ter	Exon 8	Likely Pathogenic	Feb 27, 2024	GM1 gangliosidosis type 2		ClinVar	<a href="#">38703036</a>		<a href="#">3767206</a>
c.873C>T	p.Ser291=	Exon 8	Likely Benign	Oct 13, 2025	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">2.05e-06</a>	ClinVar; gnomAD			<a href="#">1157245</a>
c.875C>G	p.Ser292Cys	Exon 8	VUS	May 15, 2025	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B; Inborn genetic diseases	<a href="#">9.58e-06</a>	ClinVar; gnomAD			<a href="#">858357</a>
c.876C>G	p.Ser292=	Exon 8	Likely Benign	Oct 24, 2024	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">6.84e-07</a>	ClinVar; gnomAD			<a href="#">1078263</a>
c.876C>T	p.Ser292=	Exon 8	Likely Benign	Aug 14, 2025	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">6.84e-07</a>	ClinVar; gnomAD			<a href="#">1624061</a>
c.879C>G	p.Leu293=	Exon 8	Likely Benign	Sep 05, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">6.84e-07</a>	ClinVar; gnomAD			<a href="#">2029100</a>
c.879C>T	p.Leu293=	Exon 8	Likely Benign	Jul 31, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">1.37e-06</a>	ClinVar; gnomAD			<a href="#">1112333</a>
c.881_882del	p.Leu293_Tyr294insTer	Exon 8	Pathogenic	Mar 05, 2025	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis; GM1 gangliosidosis; GLB1-related disorder	<a href="#">7.52e-06</a>	ClinVar; gnomAD	<a href="#">18524657</a> ; <a href="#">30555092</a>		<a href="#">817580</a>
c.882T>C	p.Tyr294=	Exon 8	Likely Benign	Jan 25, 2024	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">2943030</a>
c.884A>G	p.Asp295Gly	Exon 8	VUS	Jun 03, 2024	GM1 gangliosidosis type 3; GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">3.69e-05</a>	ClinVar; gnomAD			<a href="#">1333279</a>
c.887T>C	p.Ile296Thr	Exon 8	VUS	Jun 21, 2024		<a href="#">6.84e-07</a>	ClinVar; gnomAD	<a href="#">31731261</a>		<a href="#">3339870</a>
c.890T>G	p.Leu297Arg	Exon 8	VUS	Aug 11, 2025	Infantile GM1 gangliosidosis; Inborn genetic diseases		ClinVar			<a href="#">3767249</a>
c.895C>T	p.Arg299Cys	Exon 8	VUS	Jul 13, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">4.10e-06</a>	ClinVar; gnomAD			<a href="#">2199198</a>
c.899G>T		Exon 8	Not Classified				LOVD			
c.900_903dup	p.Ser302fs	Exon 8	Pathogenic	Jul 05, 2019	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">948954</a>
c.901G>A	p.Ala301Thr	Exon 8	Likely Pathogenic	Mar 14, 2014			ClinVar			<a href="#">167148</a>
c.902C>G	p.Ala301Gly	Exon 8	VUS	Nov 14, 2025			ClinVar			<a href="#">4536702</a>
c.902C>T	p.Ala301Val	Exon 8	Pathogenic	Dec 29, 2025	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis; MPS-IV-B; GLB1-related disorder; GM1 gangliosidosis	<a href="#">1.64e-05</a>	ClinVar; gnomAD; Santamaria 2006 (PMID:16941474)	<a href="#">16941474</a> ; <a href="#">20175788</a> ; <a href="#">20409738</a> ; <a href="#">20920281</a> ; <a href="#">30408610</a> ; <a href="#">31761138</a> ; <a href="#">33240792</a>		<a href="#">381567</a>
c.902_914+17delinsAGGCAAGTATATACTTGCC		Exon 8	Likely Pathogenic	Mar 27, 2020			ClinVar			<a href="#">420844</a>
c.903G>A	p.Ala301=	Exon 8	Likely Benign	May 30, 2025	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">2.74e-06</a>	ClinVar; gnomAD			<a href="#">1101689</a>
c.903G>T	p.Ala301=	Exon 8	Likely Benign	Dec 30, 2025	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">1576697</a>
c.905G>A	p.Ser302Asn	Exon 8	Conflicting	Sep 05, 2025	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis; Inborn genetic diseases	<a href="#">3.42e-06</a>	ClinVar; gnomAD			<a href="#">1943059</a>

cDNA Change	Protein Change	Location	Pathogenicity	Classified	Phenotype	gnomAD AF	Sources	PMID(s)	PIN	ClinVar ID
c.905G>C	p.Ser302Thr	Exon 8	VUS	Sep 20, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">1977790</a>
c.905G>T	p.Ser302Ile	Exon 8	VUS	Jun 20, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">1525293</a>
c.909G>A	p.Val303=	Exon 8	Likely Benign	Jan 29, 2024	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">2.74e-06</a>	ClinVar; gnomAD			<a href="#">738925</a>
c.911A>G	p.Asn304Ser	Exon 8	VUS	Apr 22, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">2.05e-06</a>	ClinVar; gnomAD			<a href="#">2086295</a>
c.912C>T	p.Asn304=	Exon 8	Likely Benign	Jan 17, 2024	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">1.37e-06</a>	ClinVar; gnomAD			<a href="#">1128829</a>
c.914+4A>G		Intron 8	Likely Pathogenic	Oct 04, 2024	GM1 gangliosidosis		ClinVar			<a href="#">3384079</a>
c.914+5G>A		Intron 8	VUS	Jun 15, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">1.37e-06</a>	ClinVar; gnomAD	<a href="#">17576681</a> ; <a href="#">9536098</a>		<a href="#">2937281</a>
c.914+7G>C		Intron 8	Likely Benign	Jul 09, 2020	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">1096191</a>
c.914+15C>T		Intron 8	Likely Benign	Jul 25, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">1.31e-05</a>	ClinVar; gnomAD			<a href="#">1918241</a>
c.914+15_914+16delinsAGTGTGAAGTGTGAGTGT		Intron 8	VUS	Jun 19, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">2008534</a>
c.914+17T>C		Intron 8	Likely Benign	Jan 29, 2024	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">3.42e-06</a>	ClinVar; gnomAD			<a href="#">2924263</a>
c.914+18G>T		Intron 8	Likely Benign	May 31, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">2924326</a>
c.915-18T>C		Intron 8	Likely Benign	Jan 13, 2024	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">5.47e-06</a>	ClinVar; gnomAD			<a href="#">2946730</a>
c.915-18del		Intron 8	Likely Benign	Feb 07, 2024	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">3754491</a>
c.915-17C>A		Intron 8	Likely Benign	Jan 03, 2026	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">1.09e-05</a>	ClinVar; gnomAD			<a href="#">1645157</a>
c.915-14T>C		Intron 8	Likely Benign	Jul 30, 2023	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">5.47e-06</a>	ClinVar; gnomAD			<a href="#">2946689</a>
c.915-11T>G		Intron 8	Likely Benign	May 29, 2023	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">2948338</a>
c.915-10T>C		Intron 8	Likely Benign	Nov 15, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">4.79e-06</a>	ClinVar; gnomAD			<a href="#">1127399</a>
c.915-8C>G		Intron 8	Likely Benign	Dec 12, 2024	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">6.84e-07</a>	ClinVar; gnomAD			<a href="#">1659498</a>
c.915-4G>A		Intron 8	Likely Benign	Aug 10, 2022	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">1604319</a>
c.915-2A>G		Intron 8	Likely Pathogenic	Jan 07, 2024	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar	<a href="#">16199547</a> ; <a href="#">18524657</a>		<a href="#">2942110</a>
c.915-1G>A		Intron 8	Not Classified	-	Nonpapillary renal cell carcinoma		ClinVar			<a href="#">4295484</a>
c.915-1G>C		Intron 8	Not Classified	-	Nonpapillary renal cell carcinoma		ClinVar			<a href="#">4295483</a>
c.915-1G>T		Intron 8	Likely Pathogenic	Mar 11, 2024	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis; Thyroid cancer; nonmedullary; 1; Nonpapillary renal cell carcinoma; MPS-IV-B	<a href="#">6.84e-07</a>	ClinVar; gnomAD	<a href="#">16199547</a> ; <a href="#">18524657</a>		<a href="#">1480023</a>

cDNA Change	Protein Change	Location	Pathogenicity	Classified	Phenotype	gnomAD AF	Sources	PMID(s)	PIN	ClinVar ID
c.915G>C	p.Leu305Phe	Exon 9	VUS	Aug 22, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis; Cervical cancer	<a href="#">6.84e-06</a>	ClinVar; gnomAD			<a href="#">1921716</a>
c.915G>T	p.Leu305Phe	Exon 9	Likely Benign	-			ClinVar			<a href="#">256028</a>
c.918C>A	p.Tyr306Ter	Exon 9	Likely Pathogenic	Dec 21, 2019	GM1 gangliosidosis type 2;Infantile GM1 gangliosidosis;GM1 gangliosidosis type 3		ClinVar			<a href="#">983741</a>
c.921G>A	p.Met307Ile	Exon 9	Conflicting	Feb 27, 2023	See cases		ClinVar			<a href="#">809442</a>
c.922T>C	p.Phe308Leu	Exon 9	Pathogenic	-	Infantile GM1 gangliosidosis		ClinVar; LOVD			<a href="#">100727</a>
c.931G>A	p.Gly311Arg	Exon 9	P/LP	Feb 06, 2025	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis; GM1 gangliosidosis type 2;Mucopolysaccharidosis; MPS-IV-B;Infantile GM1 gangliosidosis;GM1 gangliosidosis type 3; Mucopolysaccharidosis	<a href="#">2.67e-05</a>	ClinVar; gnomAD; OMIM; NTSAD; Cure GM1 AllStripes PIN	<a href="#">23430499</a> ; <a href="#">26108645</a> ; <a href="#">33027564</a> ; <a href="#">33737400</a>	3	<a href="#">854615</a>
c.932G>A	p.Gly311Glu	Exon 9	Likely Pathogenic	Mar 26, 2022	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar	<a href="#">23430499</a> ; <a href="#">26108645</a>		<a href="#">2117837</a>
c.936C>A	p.Thr312=	Exon 9	Likely Benign	Mar 01, 2023	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">6.84e-07</a>	ClinVar; gnomAD			<a href="#">2939378</a>
c.936C>G	p.Thr312=	Exon 9	Likely Benign	Apr 27, 2024	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">1.37e-06</a>	ClinVar; gnomAD			<a href="#">2930921</a>
c.937A>G	p.Asn313Asp	Exon 9	Likely Pathogenic	Aug 15, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">1950819</a>
c.938A>G	p.Asn313Ser	Exon 9	Pathogenic	Oct 14, 2024	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">1.37e-06</a>	ClinVar; gnomAD			<a href="#">2151550</a>
c.941T>G	p.Phe314Cys	Exon 9	Likely Pathogenic	Jan 01, 2022			ClinVar			<a href="#">932729</a>
c.944C>A	p.Ala315Asp	Exon 9	VUS	Aug 08, 2022	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B; Inborn genetic diseases	<a href="#">2.05e-06</a>	ClinVar; gnomAD			<a href="#">1693865</a>
c.945C>T	p.Ala315=	Exon 9	Likely Benign	Jan 31, 2024	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">3.42e-06</a>	ClinVar; gnomAD			<a href="#">1663272</a>
c.947A>G	p.Tyr316Cys	Exon 9	P/LP	Mar 12, 2018	Infantile GM1 gangliosidosis; GM1 gangliosidosis type 3;Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis type 2;Infantile GM1 gangliosidosis	<a href="#">1.37e-06</a>	ClinVar; LOVD; gnomAD	<a href="#">1606711</a> ; <a href="#">1907800</a> ; <a href="#">21520340</a> ; <a href="#">22128166</a> ; <a href="#">23337983</a> ; <a href="#">24767253</a>		<a href="#">929</a>
c.948T>C	p.Tyr316=	Exon 9	Likely Benign	Nov 17, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">5.47e-06</a>	ClinVar; gnomAD			<a href="#">1109073</a>
c.950G>A	p.Trp317Ter	Exon 9	Likely Pathogenic	Sep 26, 2019	Infantile GM1 gangliosidosis		ClinVar			<a href="#">800928</a>
c.955+2T>A		Intron 9	Pathogenic	Dec 12, 2024	GLB1-related disorder	<a href="#">6.84e-07</a>	ClinVar; gnomAD			<a href="#">3765821</a>
c.955+2T>G		Intron 9	P/LP	Apr 02, 2024	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis; GM1 gangliosidosis type 2;Infantile GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis type 3; Nonpapillary renal cell carcinoma; MPS-IV-B	<a href="#">1.37e-06</a>	ClinVar; gnomAD	<a href="#">16199547</a> ; <a href="#">18524657</a> ; <a href="#">31776384</a>		<a href="#">953471</a>
c.955+5G>A		Intron 9	Pathogenic	Jun 11, 2025	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">2.05e-06</a>	ClinVar; gnomAD	<a href="#">17576681</a> ; <a href="#">9536098</a>		<a href="#">2186594</a>

cDNA Change	Protein Change	Location	Pathogenicity	Classified	Phenotype	gnomAD AF	Sources	PMID(s)	PIN	ClinVar ID
c.955+8C>T		Intron 9	Likely Benign	Jul 17, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">3.42e-06</a>	ClinVar; gnomAD			<a href="#">1102999</a>
c.955+10C>T		Intron 9	Likely Benign	Mar 03, 2024	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">3755093</a>
c.955+14A>C		Intron 9	Likely Benign	Jan 19, 2025	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">3.42e-06</a>	ClinVar; gnomAD			<a href="#">2931695</a>
c.955+24G>A		Intron 9	Likely Benign	Apr 28, 2025		<a href="#">6.57e-06</a>	ClinVar; gnomAD			<a href="#">4684684</a>
c.955+60A>G		Intron 9	Benign	Jun 15, 2021	Infantile GM1 gangliosidosis; Mucopolysaccharidosis; MPS-IV-B	<a href="#">0.87500</a>	ClinVar; gnomAD			<a href="#">678119</a>
c.955+93del		Intron 9	Benign	Aug 28, 2018			ClinVar			<a href="#">1283994</a>
c.955+123dup		Intron 9	Benign	Aug 26, 2019			ClinVar			<a href="#">1251633</a>
c.955+139dup		Intron 9	Benign	Sep 02, 2019			ClinVar			<a href="#">1272712</a>
c.955+142_955+143insTTTTCT		Intron 9	Likely Benign	Jun 16, 2020			ClinVar			<a href="#">1699664</a>
c.955+142_955+143insTTTTCTTTT		Intron 9	Benign	Jun 16, 2020			ClinVar			<a href="#">1177895</a>
c.955+144T>C		Intron 9	Benign	Aug 26, 2019			ClinVar			<a href="#">1242432</a>
c.955+154del		Intron 9	Benign	Oct 16, 2019			ClinVar			<a href="#">1231412</a>
c.955+206A>G		Intron 9	Likely Benign	May 15, 2019			ClinVar			<a href="#">1207803</a>
c.955+300G>A		Intron 9	Likely Benign	Apr 20, 2019			ClinVar			<a href="#">1186798</a>
c.956-190C>G		Intron 9	Benign	Aug 28, 2018			ClinVar			<a href="#">1276962</a>
c.956-118G>C		Intron 9	Benign	Apr 20, 2019			ClinVar			<a href="#">1277988</a>
c.956-19G>A		Intron 9	Likely Benign	Dec 04, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">6.84e-07</a>	ClinVar; gnomAD			<a href="#">2933134</a>
c.956-19G>T		Intron 9	Likely Benign	Nov 05, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">1.37e-06</a>	ClinVar; gnomAD			<a href="#">2933281</a>
c.956-18C>T		Intron 9	Likely Benign	Jun 08, 2025	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">6.57e-06</a>	ClinVar; gnomAD			<a href="#">4785214</a>
c.956-14T>C		Intron 9	Likely Benign	Feb 01, 2024	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">6.57e-06</a>	ClinVar; gnomAD			<a href="#">2041644</a>
c.956-6T>C		Intron 9	Likely Benign	Jan 25, 2026	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">7.53e-06</a>	ClinVar; gnomAD			<a href="#">1158387</a>
c.956-4C>A		Intron 9	Likely Benign	Apr 28, 2023	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">2947256</a>
c.956-2A>G		Intron 9	Likely Pathogenic		GM1 gangliosidosis	2.05e-06	gnomAD; Bidchol 2015 (PMID:25936995)	<a href="#">25936995</a>		
c.956-1G>T		Intron 9	Not Classified	-	Papillary renal cell carcinoma type 1		ClinVar	<a href="#">24319509</a> ; <a href="#">25394175</a>		<a href="#">4295482</a>
c.956G>A	p.Gly319Glu	Exon 10	VUS	Aug 02, 2021	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">1408595</a>
c.958G>A	p.Ala320Thr	Exon 10	Likely Pathogenic	Jul 01, 2023			ClinVar			<a href="#">916259</a>
c.963C>T	p.Asn321=	Exon 10	Likely Benign	Jun 25, 2021	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">1.37e-06</a>	ClinVar; gnomAD			<a href="#">1622433</a>
c.964_965del	p.Ser322fs	Exon 10	P/LP	Sep 27, 2024	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis; MPS-IV-B		ClinVar	<a href="#">18524657</a>		<a href="#">1075170</a>

cDNA Change	Protein Change	Location	Pathogenicity	Classified	Phenotype	gnomAD AF	Sources	PMID(s)	PIN	ClinVar ID
c.967C>G	p.Pro323Ala	Exon 10	VUS	Nov 02, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis; GM1 gangliosidosis		ClinVar; Cure GM1 AllStripes PIN		1	<a href="#">639303</a>
c.971A>C	p.Tyr324Ser	Exon 10	Likely Pathogenic	Aug 29, 2023	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar	<a href="#">23337983</a>		<a href="#">2951339</a>
c.971A>G	p.Tyr324Cys	Exon 10	Conflicting	Nov 14, 2025	Infantile GM1 gangliosidosis;GM1 gangliosidosis type 2;Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis type 3; GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">6.84e-07</a>	ClinVar; gnomAD	<a href="#">16617000</a> ; <a href="#">23337983</a>		<a href="#">556658</a>
c.971A>T	p.Tyr324Phe	Exon 10	VUS	Apr 29, 2025		<a href="#">1.97e-05</a>	ClinVar; gnomAD			<a href="#">4540848</a>
c.972T>C	p.Tyr324=	Exon 10	Likely Benign	Dec 16, 2025	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">1.31e-05</a>	ClinVar; gnomAD			<a href="#">1101357</a>
c.975A>G	p.Ala325=	Exon 10	Likely Benign	Jul 03, 2021	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">1.37e-06</a>	ClinVar; gnomAD			<a href="#">1096477</a>
c.977C>T	p.Ala326Val	Exon 10	VUS	Apr 01, 2021		<a href="#">6.84e-07</a>	ClinVar; gnomAD			<a href="#">932728</a>
c.981G>A	p.Gln327=	Exon 10	Likely Benign	Jul 19, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">6.84e-07</a>	ClinVar; gnomAD			<a href="#">2937528</a>
c.982C>T	p.Pro328Ser	Exon 10	Conflicting	Nov 27, 2025	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">6.84e-07</a>	ClinVar; gnomAD			<a href="#">1393979</a>
c.987C>T	p.Thr329=	Exon 10	Likely Benign	Dec 13, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">2.74e-06</a>	ClinVar; gnomAD			<a href="#">1134186</a>
c.990C>T	p.Ser330=	Exon 10	Likely Benign	Dec 09, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">6.84e-07</a>	ClinVar; gnomAD			<a href="#">1915624</a>
c.993C>T	p.Tyr331=	Exon 10	Likely Benign	Oct 26, 2025	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">1.09e-05</a>	ClinVar; gnomAD			<a href="#">1125944</a>
c.994G>A	p.Asp332Asn	Exon 10	P/LP	Jun 11, 2025	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis; GM1 gangliosidosis; GM1 gangliosidosis type 2;Mucopolysaccharidosis; MPS-IV-B;Infantile GM1 gangliosidosis;GM1 gangliosidosis type 3; MPS-IV-B	<a href="#">1.16e-05</a>	ClinVar; gnomAD; Hofer 2009 (PMID:19472408); Bidchol 2015 (PMID:25936995)	<a href="#">10839995</a> ; <a href="#">10841810</a> ; <a href="#">18353697</a> ; <a href="#">19472408</a> ; <a href="#">21497194</a> ; <a href="#">23337983</a> ; <a href="#">25936995</a>		<a href="#">1071414</a>
c.994_995del	p.Asp332fs	Exon 10	Pathogenic	Jun 17, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar	<a href="#">18524657</a>		<a href="#">2007683</a>
c.996C>G		Exon 10	Not Classified				LOVD			
c.998A>G	p.Tyr333Cys	Exon 10	P/LP	Oct 04, 2024	Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis; GM1 gangliosidosis type 3;Infantile GM1 gangliosidosis;GM1 gangliosidosis type 2;Mucopolysaccharidosis	<a href="#">1.37e-06</a>	ClinVar; gnomAD	<a href="#">19091613</a> ; <a href="#">3121219</a> ; <a href="#">32071837</a>		<a href="#">1301844</a>
c.998A>T	p.Tyr333Phe	Exon 10	VUS	Apr 05, 2024		<a href="#">1.78e-05</a>	ClinVar; gnomAD			<a href="#">3251493</a>
c.1004C>T	p.Ala335Val	Exon 10	Likely Pathogenic	Oct 22, 2012		<a href="#">6.84e-07</a>	ClinVar; gnomAD			<a href="#">92892</a>
c.1008A>G	p.Pro336=	Exon 10	Likely Benign	Jul 12, 2020	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">1137882</a>
c.1009C>G	p.Leu337Val	Exon 10	Likely Pathogenic	Oct 09, 2021	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">6.84e-07</a>	ClinVar; gnomAD; Bidchol 2015 (PMID:25936995)	<a href="#">23151865</a> ; <a href="#">25936995</a>		<a href="#">1491024</a>
c.1009C>T	p.Leu337=	Exon 10	Likely Benign	Sep 25, 2025	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">2.74e-06</a>	ClinVar; gnomAD			<a href="#">785180</a>

cDNA Change	Protein Change	Location	Pathogenicity	Classified	Phenotype	gnomAD AF	Sources	PMID(s)	PIN	ClinVar ID
c.1010T>A	p.Leu337Gln	Exon 10	VUS	Apr 04, 2025			ClinVar			<a href="#">3896480</a>
c.1010T>C	p.Leu337Pro	Exon 10	P/LP	Jul 27, 2023	Infantile GM1 gangliosidosis; GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis	<a href="#">1.37e-06</a>	ClinVar; gnomAD; Bidchol 2015 (PMID:25936995)	<a href="#">23151865</a> ; <a href="#">25936995</a>		<a href="#">1251981</a>
c.1011G>C	p.Leu337=	Exon 10	Likely Benign	Aug 21, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">2025791</a>
c.1017G>A	p.Glu339=	Exon 10	Likely Benign	Oct 30, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">1671105</a>
c.1018G>A	p.Ala340Thr	Exon 10	VUS	Aug 01, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis; Inborn genetic diseases	<a href="#">2.74e-06</a>	ClinVar; LOVD; gnomAD			<a href="#">2063405</a>
c.1020T>C	p.Ala340=	Exon 10	Likely Benign	Sep 05, 2019	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">6.84e-07</a>	ClinVar; gnomAD			<a href="#">1106309</a>
c.1022G>T	p.Gly341Val	Exon 10	VUS	May 20, 2023	GM1 gangliosidosis type 3	<a href="#">6.84e-07</a>	ClinVar; gnomAD			<a href="#">3367000</a>
c.1023_1024del	p.Asp342fs	Exon 10	Likely Pathogenic	Dec 31, 2021	Infantile GM1 gangliosidosis;GM1 gangliosidosis type 2;GM1 gangliosidosis type 3		ClinVar			<a href="#">1726739</a>
c.1024G>A		Exon 10	Not Classified				LOVD			
c.1026C>A	p.Asp342Glu	Exon 10	VUS	Jan 22, 2024	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">1525326</a>
c.1027C>T	p.Leu343Phe	Exon 10	VUS	Mar 25, 2024	Infantile GM1 gangliosidosis	<a href="#">6.84e-07</a>	ClinVar; gnomAD			<a href="#">3064524</a>
c.1029C>T	p.Leu343=	Exon 10	Likely Benign	Apr 09, 2024	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">3762993</a>
c.1030A>G	p.Thr344Ala	Exon 10	VUS	Oct 17, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">2045556</a>
c.1032T>C	p.Thr344=	Exon 10	Conflicting	Dec 26, 2025	Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">0.00131</a>	ClinVar; LOVD; gnomAD			<a href="#">344792</a>
c.1038G>A	p.Lys346=	Exon 10	Likely Benign	Mar 13, 2023	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">6.84e-07</a>	ClinVar; gnomAD			<a href="#">2944910</a>
c.1038G>C	p.Lys346Asn	Exon 10	P/LP	Dec 04, 2025	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis type 2;GM1 gangliosidosis type 3;Infantile GM1 gangliosidosis; MPS-IV-B;GM1 gangliosidosis; GLB1-related disorder; Infantile GM1 gangliosidosis	<a href="#">7.53e-06</a>	ClinVar; gnomAD; Santamaria 2006 (PMID:16941474); Hofer 2009 (PMID:19472408); Cure GM1 AllStripes PIN	<a href="#">16941474</a> ; <a href="#">19472408</a> ; <a href="#">21520340</a> ; <a href="#">24777551</a> ; <a href="#">33737400</a>	1	<a href="#">550856</a>
c.1038G>T	p.Lys346Asn	Exon 10	P/LP	Oct 09, 2023	Infantile GM1 gangliosidosis;GM1 gangliosidosis type 2;Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis type 3; GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis	<a href="#">2.74e-06</a>	ClinVar; gnomAD; Santamaria 2006 (PMID:16941474); Hofer 2009 (PMID:19472408)	<a href="#">16941474</a> ; <a href="#">19472408</a> ; <a href="#">21520340</a> ; <a href="#">22128166</a> ; <a href="#">23337983</a> ; <a href="#">24777551</a>		<a href="#">558213</a>
c.1040A>G	p.Tyr347Cys	Exon 10	Pathogenic	Oct 11, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar; Santamaria 2006 (PMID:16941474)	<a href="#">16941474</a> ; <a href="#">21497194</a> ; <a href="#">21520340</a>		<a href="#">1475998</a>
c.1047T>A	p.Ala349=	Exon 10	Likely Benign	Sep 09, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">2061599</a>

cDNA Change	Protein Change	Location	Pathogenicity	Classified	Phenotype	gnomAD AF	Sources	PMID(s)	PIN	ClinVar ID
c.1051C>T	p.Arg351Ter	Exon 10	Pathogenic	Dec 14, 2025	GM1-gangliosidosis; type I; with cardiac involvement; GM1 gangliosidosis type 3;Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis type 2;Infantile GM1 gangliosidosis; Mucopolysaccharidosis; MPS-IV-B; Spastic ataxia; MPS-IV-B;GM1 gangliosidosis; Infantile GM1 gangliosidosis	<a href="#">1.16e-05</a>	ClinVar; gnomAD; OMIM; Caciotti 2005 (PMID:15714521); Cure GM1 AllStripes PIN	<a href="#">10841810</a> ; <a href="#">15714521</a> ; <a href="#">18353697</a> ; <a href="#">18524657</a> ; <a href="#">21497194</a> ; <a href="#">8922281</a>	1	<a href="#">941</a>
c.1052G>A	p.Arg351Gln	Exon 10	VUS	Nov 07, 2022	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B; Inborn genetic diseases	<a href="#">7.53e-06</a>	ClinVar; LOVD; gnomAD			<a href="#">1062142</a>
c.1056C>T	p.Asn352=	Exon 10	Likely Benign	Feb 19, 2023	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">2944524</a>
c.1060A>G	p.Ile354Val	Exon 10	Likely Pathogenic	Jan 25, 2023	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">6.85e-07</a>	ClinVar; gnomAD	<a href="#">26108645</a> ; <a href="#">34258138</a>		<a href="#">2941220</a>
c.1061T>G	p.Ile354Ser	Exon 10	Not Classified	-	GM1 gangliosidosis type 2		ClinVar			<a href="#">66109</a>
c.1062C>T	p.Ile354=	Exon 10	Likely Benign	Nov 08, 2023	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">6.85e-07</a>	ClinVar; gnomAD			<a href="#">1563143</a>
c.1063C>T	p.Gln355Ter	Exon 10	Pathogenic	Mar 01, 2024	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar	<a href="#">18524657</a>		<a href="#">3749750</a>
c.1068G>A	p.Lys356=	Exon 10	VUS	Jan 28, 2015		<a href="#">6.85e-07</a>	ClinVar; gnomAD			<a href="#">193584</a>
c.1068+1G>T		Intron 10	VUS	Apr 04, 2024	Infantile GM1 gangliosidosis		ClinVar			<a href="#">3068264</a>
c.1068+4G>A		Intron 10	VUS	Nov 05, 2024	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">5.48e-06</a>	ClinVar; LOVD; gnomAD	<a href="#">17576681</a> ; <a href="#">9536098</a>		<a href="#">3761308</a>
c.1068+7G>A		Intron 10	Likely Benign	Mar 23, 2021	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">3.43e-06</a>	ClinVar; gnomAD			<a href="#">1584861</a>
c.1068+14A>G		Intron 10	Likely Benign	Oct 06, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">1.37e-06</a>	ClinVar; gnomAD			<a href="#">2926104</a>
c.1068+18G>A		Intron 10	Likely Benign	Apr 18, 2023	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">6.85e-07</a>	ClinVar; gnomAD			<a href="#">2946874</a>
c.1068+18G>C		Intron 10	Likely Benign	Dec 13, 2022	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">2942117</a>
c.1068+18G>T		Intron 10	Likely Benign	Feb 01, 2024	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">1.37e-06</a>	ClinVar; gnomAD			<a href="#">1968377</a>
c.1068+20T>C		Intron 10	Likely Benign	Aug 04, 2023	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">1605087</a>
c.1068+217G>T		Intron 10	Benign	Aug 28, 2018			ClinVar			<a href="#">1231794</a>
c.1068+264G>A		Intron 10	Benign	Aug 28, 2018			ClinVar			<a href="#">1246406</a>
c.1068+291A>C		Intron 10	Likely Benign	Nov 13, 2019			ClinVar			<a href="#">1188742</a>
c.1068+2341_1068+2342ins GCTTGGTATTTTTCTGAG GGTATCTTTGCTTGGTATT TTTCTGAGTTAGCTTTTGC TTGATTTTTTTTCTGATG TACTTT		Intron 10	VUS	Nov 11, 2022	Schizophrenia		ClinVar			<a href="#">1801444</a>
c.1069-231G>A		Intron 10	Benign	Mar 10, 2019	Malignant tumor of esophagus		ClinVar			<a href="#">1234200</a>
c.1069-20T>C		Intron 10	Likely Benign	Jan 03, 2024	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">1.10e-05</a>	ClinVar; gnomAD			<a href="#">2935558</a>

cDNA Change	Protein Change	Location	Pathogenicity	Classified	Phenotype	gnomAD AF	Sources	PMID(s)	PIN	ClinVar ID
c.1069-19T>C		Intron 10	Likely Benign	Dec 30, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">1.38e-06</a>	ClinVar; gnomAD			<a href="#">2044458</a>
c.1069-17T>G		Intron 10	Likely Benign	Oct 04, 2023	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">6.88e-07</a>	ClinVar; gnomAD			<a href="#">2939107</a>
c.1069-8G>A		Intron 10	Likely Benign	Jul 06, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">1.17e-05</a>	ClinVar; gnomAD			<a href="#">2935507</a>
c.1069-8G>T		Intron 10	Likely Benign	Feb 07, 2024	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">1.37e-06</a>	ClinVar; gnomAD			<a href="#">1637152</a>
c.1069-4A>G		Intron 10	Conflicting	Nov 13, 2024	GM1 gangliosidosis; Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis;Mucopolysaccharidosis	<a href="#">0.00011</a>	ClinVar; LOVD; gnomAD			<a href="#">899441</a>
c.1069_1233dup		Exon 11	Pathogenic	Aug 01, 1991	Infantile GM1 gangliosidosis		ClinVar	<a href="#">1907800</a>		<a href="#">927</a>
c.1071T>G	p.Phe357Leu	Exon 11	VUS	Oct 18, 2025	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis; GM1 gangliosidosis	<a href="#">8.90e-06</a>	ClinVar; gnomAD	<a href="#">20175788</a> ; <a href="#">29396849</a>		<a href="#">167147</a>
c.1071_1073delinsGG	p.Phe357fs	Exon 11	P/LP	Mar 14, 2025	Infantile GM1 gangliosidosis; Mucopolysaccharidosis; MPS-IV-B		ClinVar	<a href="#">30408610</a>		<a href="#">2585128</a>
c.1074A>G	p.Glu358=	Exon 11	Likely Benign	Jan 08, 2024	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">5.48e-06</a>	ClinVar; gnomAD			<a href="#">1134116</a>
c.1077A>G	p.Lys359=	Exon 11	Likely Benign	Aug 29, 2025	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">4.80e-06</a>	ClinVar; gnomAD			<a href="#">758587</a>
c.1077del	p.Val360fs	Exon 11	Pathogenic	Oct 18, 2025	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis; MPS-IV-B; Infantile GM1 gangliosidosis; GM1 gangliosidosis	<a href="#">8.93e-06</a>	ClinVar; gnomAD; Bidchol 2015 (PMID:25936995)	<a href="#">18524657</a> ; <a href="#">20175788</a> ; <a href="#">25936995</a> ; <a href="#">30408610</a>		<a href="#">167146</a>
c.1079T>C	p.Val360Ala	Exon 11	VUS	Oct 02, 2024	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">4.79e-06</a>	ClinVar; gnomAD			<a href="#">3750804</a>
c.1083A>G	p.Pro361=	Exon 11	Likely Benign	Jan 10, 2023	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">2942540</a>
c.1084G>A	p.Glu362Lys	Exon 11	VUS	Jun 29, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">1957397</a>
c.1086A>G	p.Glu362=	Exon 11	Likely Benign	Feb 22, 2021	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">1636050</a>
c.1088G>A	p.Gly363Asp	Exon 11	VUS	Feb 24, 2024	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">1.10e-05</a>	ClinVar; gnomAD			<a href="#">1517651</a>
c.1092T>C	p.Pro364=	Exon 11	Likely Benign	Jan 24, 2026	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">3.97e-05</a>	ClinVar; gnomAD			<a href="#">719889</a>
c.1095C>G	p.Ile365Met	Exon 11	VUS	May 25, 2022	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">2.74e-06</a>	ClinVar; gnomAD			<a href="#">1985638</a>
c.1095C>T	p.Ile365=	Exon 11	Conflicting	Jan 25, 2026	GM1 gangliosidosis; GLB1-related disorder; Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis;Mucopolysaccharidosis	<a href="#">9.52e-05</a>	ClinVar; LOVD; gnomAD			<a href="#">899440</a>
c.1097C>A	p.Pro366His	Exon 11	Not Classified	-	-		ClinVar			<a href="#">242560</a>
c.1099C>T	p.Pro367Ser	Exon 11	VUS	-	Infantile GM1 gangliosidosis	<a href="#">6.87e-07</a>	ClinVar; gnomAD			<a href="#">1332870</a>
c.1101A>G	p.Pro367=	Exon 11	Likely Benign	Dec 16, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">3.42e-06</a>	ClinVar; gnomAD			<a href="#">1119663</a>

cDNA Change	Protein Change	Location	Pathogenicity	Classified	Phenotype	gnomAD AF	Sources	PMID(s)	PIN	ClinVar ID
c.1110A>C	p.Pro370=	Exon 11	Likely Benign	Oct 26, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">2036837</a>
c.1110A>G	p.Pro370=	Exon 11	Likely Benign	Mar 12, 2024	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">5.48e-06</a>	ClinVar; gnomAD			<a href="#">761756</a>
c.1113G>A	p.Lys371=	Exon 11	Likely Benign	Nov 17, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">1.37e-06</a>	ClinVar; gnomAD			<a href="#">2922956</a>
c.1119A>T	p.Ala373=	Exon 11	Likely Benign	Sep 10, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">6.57e-06</a>	ClinVar; gnomAD			<a href="#">2933312</a>
c.1120T>C	p.Tyr374His	Exon 11	VUS	Jan 07, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">1973604</a>
c.1121A>T	p.Tyr374Phe	Exon 11	VUS	Jun 03, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">2.74e-06</a>	ClinVar; gnomAD			<a href="#">2157464</a>
c.1122T>A	p.Tyr374Ter	Exon 11	Likely Pathogenic	Feb 28, 2022	Infantile GM1 gangliosidosis;GM1 gangliosidosis type 3;GM1 gangliosidosis type 2		ClinVar			<a href="#">1724830</a>
c.1122T>C	p.Tyr374=	Exon 11	Likely Benign	Feb 05, 2024	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">3.42e-06</a>	ClinVar; gnomAD			<a href="#">735429</a>
c.1122T>G	p.Tyr374Ter	Exon 11	Pathogenic	Dec 24, 2022	Infantile GM1 gangliosidosis		ClinVar			<a href="#">1810758</a>
c.1135T>G	p.Leu379Val	Exon 11	VUS	May 08, 2024			ClinVar			<a href="#">3379448</a>
c.1142del	p.Lys381fs	Exon 11	P/LP	Dec 22, 2021	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B; Infantile GM1 gangliosidosis;GM1 gangliosidosis type 3;GM1 gangliosidosis type 2		ClinVar	<a href="#">18524657</a>		<a href="#">1454377</a>
c.1143G>C	p.Lys381Asn	Exon 11	Not Classified	-	Hepatocellular carcinoma		ClinVar			<a href="#">4295481</a>
c.1143+1G>T		Intron 11	Likely Pathogenic	Feb 22, 2024	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar	<a href="#">16199547</a> ; <a href="#">18524657</a>		<a href="#">3761539</a>
c.1143+3A>C		Intron 11	VUS	Jul 14, 2021	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">1.10e-05</a>	ClinVar; gnomAD	<a href="#">17576681</a> ; <a href="#">9536098</a>		<a href="#">1430958</a>
c.1143+5G>A		Intron 11	VUS	Jul 09, 2025	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar	<a href="#">17576681</a> ; <a href="#">9536098</a>		<a href="#">4794273</a>
c.1143+11A>G		Intron 11	Likely Benign	Oct 20, 2023	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">6.87e-07</a>	ClinVar; gnomAD			<a href="#">2940062</a>
c.1143+14del		Intron 11	Benign	Jan 31, 2026	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">0.00011</a>	ClinVar; gnomAD			<a href="#">1600411</a>
c.1143+14dup		Intron 11	Benign	Jan 19, 2026	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">0.00021</a>	ClinVar; gnomAD			<a href="#">1641451</a>
c.1143+15del		Intron 11	Likely Benign	Dec 23, 2024	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">3.44e-06</a>	ClinVar; gnomAD			<a href="#">2041650</a>
c.1143+16A>T		Intron 11	Likely Benign	Jul 22, 2023	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">6.89e-07</a>	ClinVar; gnomAD			<a href="#">2939847</a>
c.1143+20_1143+21del		Intron 11	Likely Benign	Sep 01, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">3.45e-06</a>	ClinVar; gnomAD			<a href="#">2923346</a>
c.1143+28G>C		Intron 11	Likely Benign	Mar 10, 2019		<a href="#">0.01320</a>	ClinVar; gnomAD			<a href="#">558920</a>
c.1143+47G>A		Intron 11	Likely Benign	Sep 26, 2018		<a href="#">0.01220</a>	ClinVar; gnomAD			<a href="#">1199012</a>
c.1144-256C>G		Intron 11	Benign	Mar 10, 2019			ClinVar			<a href="#">1290141</a>
c.1144-18G>C		Intron 11	Likely Benign	Jul 26, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">1.37e-06</a>	ClinVar; gnomAD			<a href="#">2924205</a>

cDNA Change	Protein Change	Location	Pathogenicity	Classified	Phenotype	gnomAD AF	Sources	PMID(s)	PIN	ClinVar ID
c.1144-12G>A		Intron 11	Likely Benign	Mar 19, 2022	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">6.85e-07</a>	ClinVar; gnomAD			<a href="#">2109232</a>
c.1144-8C>G		Intron 11	Likely Benign	Aug 23, 2019	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">764447</a>
c.1144-4T>C		Intron 11	Likely Benign	Nov 05, 2024	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">2044234</a>
c.1144-2A>G		Intron 11	P/LP	Sep 22, 2023	Infantile GM1 gangliosidosis;GM1 gangliosidosis type 2;Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis type 3; GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B; Mucopolysaccharidosis	<a href="#">1.37e-06</a>	ClinVar; gnomAD	<a href="#">16199547</a> ; <a href="#">18524657</a> ; <a href="#">30408610</a>		<a href="#">557226</a>
c.1146A>G	p.Leu382=	Exon 12	Likely Benign	Oct 10, 2022	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">3.42e-06</a>	ClinVar; gnomAD			<a href="#">1453354</a>
c.1147A>T	p.Lys383Ter	Exon 12	P/LP	Apr 04, 2024	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis type 2;Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis type 3;Infantile GM1 gangliosidosis	<a href="#">6.84e-07</a>	ClinVar; gnomAD	<a href="#">18524657</a>		<a href="#">2164182</a>
c.1147_1148del	p.Lys383fs	Exon 12	Pathogenic	Dec 24, 2021	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar	<a href="#">18524657</a>		<a href="#">2058116</a>
c.1150A>T	p.Thr384Ser	Exon 12	Not Classified	-	GM1 gangliosidosis type 2		ClinVar			<a href="#">66110</a>
c.1152A>C	p.Thr384=	Exon 12	Likely Benign	Feb 16, 2024	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">6.57e-06</a>	ClinVar; gnomAD			<a href="#">3762578</a>
c.1158A>G	p.Gly386=	Exon 12	Likely Benign	Mar 24, 2025	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">4793927</a>
c.1169A>G	p.Asp390Gly	Exon 12	Likely Benign	Jan 11, 2024	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">6.16e-06</a>	ClinVar; gnomAD			<a href="#">2144396</a>
c.1174C>T	p.Leu392=	Exon 12	Likely Benign	Jul 07, 2025	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">4.10e-06</a>	ClinVar; gnomAD			<a href="#">1602203</a>
c.1174_1175del	p.Leu392fs	Exon 12	P/LP	Dec 01, 2025	GM1 gangliosidosis type 2;Infantile GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis type 3; Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis; MPS-IV-B	<a href="#">7.53e-06</a>	ClinVar; gnomAD	<a href="#">18524657</a>		<a href="#">92893</a>
c.1175T>G	p.Leu392Arg	Exon 12	VUS	May 01, 2024		<a href="#">2.05e-06</a>	ClinVar; gnomAD			<a href="#">3239286</a>
c.1176G>A	p.Leu392=	Exon 12	Likely Benign	Jul 06, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">6.84e-06</a>	ClinVar; gnomAD			<a href="#">1622269</a>
c.1176G>C	p.Leu392=	Exon 12	Likely Benign	Jan 04, 2024	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">1965864</a>
c.1176G>T	p.Leu392=	Exon 12	Likely Benign	Jan 06, 2025	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">4.10e-06</a>	ClinVar; gnomAD			<a href="#">2168393</a>
c.1184C>G	p.Ser395Cys	Exon 12	VUS	Jun 16, 2024	Inborn genetic diseases	<a href="#">2.60e-05</a>	ClinVar; gnomAD			<a href="#">3281503</a>
c.1184C>T	p.Ser395Phe	Exon 12	VUS	Aug 05, 2022	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">6.84e-07</a>	ClinVar; gnomAD			<a href="#">1992580</a>
c.1188_1189delinsCT	p.Pro397Ser	Exon 12	Likely Pathogenic	Nov 24, 2022	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">2941816</a>
c.1188dup	p.Pro397fs	Exon 12	Pathogenic	-	Infantile GM1 gangliosidosis		ClinVar; LOVD			<a href="#">100725</a>

cDNA Change	Protein Change	Location	Pathogenicity	Classified	Phenotype	gnomAD AF	Sources	PMID(s)	PIN	ClinVar ID
c.1189C>T	p.Pro397Ser	Exon 12	VUS	Jul 25, 2025			ClinVar			<a href="#">3339178</a>
c.1191del	p.Ile398fs	Exon 12	Pathogenic	Apr 07, 2021	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">6.84e-07</a>	ClinVar; gnomAD	<a href="#">18524657</a>		<a href="#">591620</a>
c.1192A>G	p.Ile398Val	Exon 12	Likely Benign	Feb 01, 2026	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis; Inborn genetic diseases	<a href="#">5.81e-05</a>	ClinVar; gnomAD			<a href="#">1546896</a>
c.1198A>C	p.Ser400Arg	Exon 12	VUS	Oct 16, 2024	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">6.57e-06</a>	ClinVar; gnomAD			<a href="#">2094626</a>
c.1201C>A	p.Leu401Ile	Exon 12	VUS	Jan 02, 2021	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">1500957</a>
c.1202T>C	p.Leu401Pro	Exon 12	VUS	Sep 24, 2021	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">2.74e-06</a>	ClinVar; gnomAD			<a href="#">1491187</a>
c.1202T>G	p.Leu401Arg	Exon 12	VUS	Oct 14, 2024	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">4.79e-06</a>	ClinVar; gnomAD			<a href="#">1415908</a>
c.1207C>T	p.Pro403Ser	Exon 12	VUS	Jul 03, 2025	Inborn genetic diseases; Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">2228306</a>
c.1208C>T	p.Pro403Leu	Exon 12	VUS	Nov 23, 2024	Inborn genetic diseases	<a href="#">4.10e-06</a>	ClinVar; gnomAD			<a href="#">3520385</a>
c.1209C>G	p.Pro403=	Exon 12	Benign	Jan 31, 2026	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">5.95e-05</a>	ClinVar; gnomAD			<a href="#">779088</a>
c.1215A>C	p.Thr405=	Exon 12	Likely Benign	Sep 30, 2019	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">2.05e-06</a>	ClinVar; gnomAD			<a href="#">1138461</a>
c.1215A>G	p.Thr405=	Exon 12	Likely Benign	Jun 18, 2021	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">1566921</a>
c.1223A>C	p.Gln408Pro	Exon 12	Likely Pathogenic	Jan 13, 2026	Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis type 3;Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis type 2;Infantile GM1 gangliosidosis		ClinVar; Hofer 2009 (PMID:19472408)	<a href="#">11511921</a> ; <a href="#">19472408</a> ; <a href="#">21520340</a>		<a href="#">943</a>
c.1223A>G	p.Gln408Arg	Exon 12	VUS	Jul 01, 2024	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">6.84e-07</a>	ClinVar; gnomAD			<a href="#">1399203</a>
c.1224G>A	p.Gln408=	Exon 12	Likely Benign	Jul 17, 2023	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">2941769</a>
c.1225del	p.Gln408_Val409insTer	Exon 12	Pathogenic	May 16, 2023	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar	<a href="#">18524657</a>		<a href="#">2947807</a>
c.1228A>C	p.Lys410Gln	Exon 12	VUS	Feb 12, 2024	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">1.98e-05</a>	ClinVar; gnomAD			<a href="#">2051796</a>
c.1231C>T	p.Gln411Ter	Exon 12	Pathogenic	Aug 02, 2023	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar	<a href="#">18524657</a>		<a href="#">2950588</a>
c.1232A>C	p.Gln411Pro	Exon 12	VUS	Jul 10, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">6.84e-07</a>	ClinVar; gnomAD			<a href="#">2050376</a>
c.1233+1G>A		Intron 12	P/LP	Mar 29, 2025	Infantile GM1 gangliosidosis;GM1 gangliosidosis type 2;Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis type 3; GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B; GLB1-related disorder; Mucopolysaccharidosis	<a href="#">6.84e-07</a>	ClinVar; gnomAD; OMIM; NTSAD; Cure GM1 AllStripes PIN	<a href="#">16199547</a> ; <a href="#">18524657</a> ; <a href="#">31761138</a>	1	<a href="#">556286</a>
c.1233+1G>T		Intron 12	Likely Pathogenic	Mar 29, 2025	Mucopolysaccharidosis; MPS-IV-B		ClinVar	<a href="#">31761138</a>		<a href="#">4818310</a>
c.1233+5G>A		Intron 12	Not Classified			2.05e-06	LOVD; gnomAD			

cDNA Change	Protein Change	Location	Pathogenicity	Classified	Phenotype	gnomAD AF	Sources	PMID(s)	PIN	ClinVar ID
c.1233+8T>C		Intron 12	Likely Benign	Feb 04, 2026	GM1 gangliosidosis; Mucopolysaccharidosis; MPS-IV-B; MPS-IV-B;GM1 gangliosidosis; Infantile GM1 gangliosidosis	<a href="#">0.21300</a>	ClinVar; LOVD; gnomAD			<a href="#">92894</a>
c.1233+10del		Intron 12	Likely Benign	Mar 30, 2022	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">2119080</a>
c.1233+12A>G		Intron 12	Likely Benign	Dec 02, 2023	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">1.37e-06</a>	ClinVar; gnomAD			<a href="#">2954366</a>
c.1233+13A>C		Intron 12	Likely Benign	Nov 09, 2023	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">2.05e-06</a>	ClinVar; gnomAD			<a href="#">2953718</a>
c.1233+15G>A		Intron 12	Likely Benign	May 19, 2025	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">1.37e-06</a>	ClinVar; gnomAD			<a href="#">2161217</a>
c.1233+15del		Intron 12	Likely Benign	Oct 26, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">2937262</a>
c.1233+19T>C		Intron 12	Likely Benign	Dec 23, 2024	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">4.80e-06</a>	ClinVar; gnomAD			<a href="#">2168322</a>
c.1233+36A>G		Intron 12	Likely Benign	Jan 29, 2019		<a href="#">0.00333</a>	ClinVar; gnomAD			<a href="#">1198799</a>
c.1233+91G>C		Intron 12	Likely Benign	Jun 15, 2021	Mucopolysaccharidosis; MPS-IV-B; Infantile GM1 gangliosidosis		ClinVar			<a href="#">1174036</a>
c.1233+174G>T		Intron 12	Benign	Aug 28, 2018			ClinVar			<a href="#">1280342</a>
c.1233+218C>T		Intron 12	Benign	Mar 10, 2019			ClinVar			<a href="#">1298258</a>
c.1234-307G>A		Intron 12	Likely Benign	Jan 12, 2020			ClinVar			<a href="#">1220424</a>
c.1234-177C>T		Intron 12	Benign	Mar 10, 2019			ClinVar			<a href="#">1244312</a>
c.1234-20T>G		Intron 12	Likely Benign	Nov 28, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">2035797</a>
c.1234-19T>A		Intron 12	Likely Benign	Dec 05, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">1.10e-05</a>	ClinVar; gnomAD			<a href="#">2052703</a>
c.1234-17A>T		Intron 12	Benign	Jan 31, 2026	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">0.00015</a>	ClinVar; gnomAD			<a href="#">1584848</a>
c.1234-13C>A		Intron 12	Likely Benign	May 16, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">2931644</a>
c.1234-13C>G		Intron 12	Likely Benign	Jun 02, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">2935132</a>
c.1234-11T>A		Intron 12	Conflicting	Jan 09, 2026	GM1 gangliosidosis; Mucopolysaccharidosis; MPS-IV-B; MPS-IV-B;GM1 gangliosidosis; Chronic lymphocytic leukemia/small lymphocytic lymphoma	<a href="#">0.00042</a>	ClinVar; gnomAD			<a href="#">344791</a>
c.1234-10T>G		Intron 12	Likely Benign	Jan 13, 2026	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">5.40e-05</a>	ClinVar; gnomAD			<a href="#">1148229</a>
c.1234-8C>G		Intron 12	Likely Benign	Jul 13, 2025	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">2.05e-06</a>	ClinVar; gnomAD			<a href="#">1519260</a>
c.1234-8C>T		Intron 12	Likely Benign	Sep 10, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">2923157</a>
c.1234-4C>T		Intron 12	Not Classified				LOVD			
c.1234-3C>A		Intron 12	VUS	Jun 29, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis; Uterine carcinosarcoma	<a href="#">1.64e-05</a>	ClinVar; gnomAD	<a href="#">17576681</a> ; <a href="#">9536098</a>		<a href="#">2191325</a>

cDNA Change	Protein Change	Location	Pathogenicity	Classified	Phenotype	gnomAD AF	Sources	PMID(s)	PIN	ClinVar ID
c.1234-1G>T		Intron 12	Likely Pathogenic	Mar 14, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">7.52e-06</a>	ClinVar; gnomAD	<a href="#">16199547</a> ; <a href="#">18524657</a>		<a href="#">2934239</a>
c.1238A>G	p.Tyr413Cys	Exon 13	VUS	Oct 26, 2022	Inborn genetic diseases	<a href="#">6.58e-06</a>	ClinVar; gnomAD			<a href="#">2319585</a>
c.1241G>A	p.Gly414Glu	Exon 13	VUS	Sep 01, 2021	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">1.37e-06</a>	ClinVar; gnomAD			<a href="#">1388621</a>
c.1242del	p.Phe415fs	Exon 13	Likely Pathogenic	Jun 12, 2024	GM1 gangliosidosis type 3;Infantile GM1 gangliosidosis;GM1 gangliosidosis type 2;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">3589132</a>
c.1250T>C	p.Leu417Pro	Exon 13	VUS	Feb 16, 2025			ClinVar			<a href="#">3910314</a>
c.1251G>A	p.Leu417=	Exon 13	Likely Benign	Feb 16, 2025	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">5.47e-06</a>	ClinVar; gnomAD			<a href="#">1674235</a>
c.1254C>G	p.Tyr418Ter	Exon 13	Pathogenic	-	GM1 gangliosidosis	<a href="#">6.84e-07</a>	ClinVar; gnomAD			<a href="#">973577</a>
c.1254C>T	p.Tyr418=	Exon 13	Likely Benign	Jan 04, 2024	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">6.84e-07</a>	ClinVar; gnomAD			<a href="#">1983286</a>
c.1255C>T	p.Arg419Trp	Exon 13	P/LP	Jan 06, 2025	GM1 gangliosidosis; GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B; Mucopolysaccharidosis	<a href="#">8.21e-06</a>	ClinVar; gnomAD	<a href="#">31761138</a> ; <a href="#">33083013</a>		<a href="#">973576</a>
c.1256G>A	p.Arg419Gln	Exon 13	Conflicting	Oct 15, 2025	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis type 2;GM1 gangliosidosis type 3;Mucopolysaccharidosis; MPS-IV-B;Infantile GM1 gangliosidosis	<a href="#">4.10e-05</a>	ClinVar; LOVD; gnomAD	<a href="#">31761138</a> ; <a href="#">33083013</a>		<a href="#">1516780</a>
c.1256G>C	p.Arg419Pro	Exon 13	Likely Pathogenic	Dec 31, 2022	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar	<a href="#">31761138</a> ; <a href="#">33083013</a>		<a href="#">2942833</a>
c.1257G>A	p.Arg419=	Exon 13	Likely Benign	Sep 24, 2025	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">1.03e-05</a>	ClinVar; gnomAD			<a href="#">1135711</a>
c.1258A>C	p.Thr420Pro	Exon 13	P/LP	Sep 13, 2024	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis; MPS-IV-B	<a href="#">1.31e-05</a>	ClinVar; gnomAD; Santamaria 2006 (PMID:16941474)	<a href="#">16941474</a> ; <a href="#">17664528</a> ; <a href="#">23337983</a>		<a href="#">1070505</a>
c.1258A>G	p.Thr420Ala	Exon 13	Conflicting	Jun 03, 2025	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">6.84e-07</a>	ClinVar; gnomAD; Santamaria 2006 (PMID:16941474)	<a href="#">16941474</a>		<a href="#">1483973</a>
c.1259C>A		Exon 13	Not Classified				LOVD			
c.1261A>G	p.Thr421Ala	Exon 13	VUS	Sep 01, 2021	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">6.16e-06</a>	ClinVar; gnomAD			<a href="#">1448206</a>
c.1263A>C	p.Thr421=	Exon 13	Likely Benign	May 01, 2023	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">2947388</a>
c.1263A>G	p.Thr421=	Exon 13	Likely Benign	Aug 30, 2020	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">1.37e-06</a>	ClinVar; gnomAD			<a href="#">1144994</a>
c.1264C>T	p.Leu422Phe	Exon 13	VUS	May 04, 2022			ClinVar			<a href="#">1684771</a>
c.1273G>C	p.Asp425His	Exon 13	VUS	Aug 30, 2021	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">1.37e-06</a>	ClinVar; gnomAD			<a href="#">2420802</a>
c.1276T>G	p.Cys426Gly	Exon 13	Pathogenic	Feb 24, 2024	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">2.67e-05</a>	ClinVar; gnomAD	<a href="#">26646981</a>		<a href="#">1372550</a>
c.1279A>G	p.Ser427Gly	Exon 13	VUS	Mar 22, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">3.42e-06</a>	ClinVar; gnomAD			<a href="#">2195747</a>
c.1279_1298del	p.Ser427fs	Exon 13	Pathogenic	Mar 19, 2022	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">6.84e-07</a>	ClinVar; gnomAD	<a href="#">18524657</a>		<a href="#">2114150</a>

cDNA Change	Protein Change	Location	Pathogenicity	Classified	Phenotype	gnomAD AF	Sources	PMID(s)	PIN	ClinVar ID
c.1282A>G	p.Asn428Asp	Exon 13	Likely Benign	Aug 30, 2025	Inborn genetic diseases	<a href="#">1.37e-06</a>	ClinVar; gnomAD			<a href="#">4263967</a>
c.1283A>G	p.Asn428Ser	Exon 13	VUS	Apr 20, 2021		<a href="#">2.05e-06</a>	ClinVar; gnomAD			<a href="#">1314582</a>
c.1285C>T	p.Pro429Ser	Exon 13	Conflicting	Jan 19, 2026	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis type 3;Infantile GM1 gangliosidosis;GM1 gangliosidosis type 2;Mucopolysaccharidosis	<a href="#">0.00011</a>	ClinVar; LOVD; gnomAD	<a href="#">28332257</a>		<a href="#">242561</a>
c.1286C>T	p.Pro429Leu	Exon 13	Conflicting	Jan 26, 2026	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B; Inborn genetic diseases	<a href="#">3.22e-05</a>	ClinVar; gnomAD	<a href="#">28518168</a> ; <a href="#">32461654</a>		<a href="#">839524</a>
c.1293T>C	p.Pro431=	Exon 13	Likely Benign	Apr 22, 2021	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">1.31e-05</a>	ClinVar; gnomAD			<a href="#">1143435</a>
c.1298_1299del	p.Ser433fs	Exon 13	P/LP	Apr 01, 2022	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis type 3;Infantile GM1 gangliosidosis;GM1 gangliosidosis type 2	<a href="#">6.84e-07</a>	ClinVar; gnomAD	<a href="#">18524657</a>		<a href="#">842879</a>
c.1300T>A	p.Ser434Thr	Exon 13	VUS	May 15, 2025	Inborn genetic diseases	<a href="#">6.58e-06</a>	ClinVar; gnomAD			<a href="#">4030002</a>
c.1306C>G	p.Leu436Val	Exon 13	VUS	Sep 27, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">4.79e-06</a>	ClinVar; gnomAD			<a href="#">1444178</a>
c.1306C>T	p.Leu436Phe	Exon 13	Likely Benign	Feb 04, 2026	GM1 gangliosidosis type 2;Infantile GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis type 3; GM1 gangliosidosis; Mucopolysaccharidosis; MPS-IV-B; MPS-IV-B;GM1 gangliosidosis; GLB1-related disorder	<a href="#">0.01320</a>	ClinVar; LOVD; gnomAD; Caciotti 2003 (PMID:12644936); Santamaria 2006 (PMID:16941474)	<a href="#">12644936</a> ; <a href="#">16941474</a>		<a href="#">92895</a>
c.1310A>G	p.Asn437Ser	Exon 13	Conflicting	Feb 02, 2026	GM1 gangliosidosis type 3;Mucopolysaccharidosis; MPS-IV-B;Infantile GM1 gangliosidosis;GM1 gangliosidosis type 2; Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis; MPS-IV-B; GM1 gangliosidosis; GLB1-related disorder	<a href="#">0.00035</a>	ClinVar; LOVD; gnomAD	<a href="#">26990548</a> ; <a href="#">28332257</a> ; <a href="#">34426522</a>		<a href="#">554101</a>
c.1310A>T	p.Asn437Ile	Exon 13	P/LP	Mar 07, 2024	GM1 gangliosidosis type 2;Infantile GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis type 3; Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">6.84e-07</a>	ClinVar; gnomAD			<a href="#">92896</a>
c.1310del	p.Asn437fs	Exon 13	Pathogenic	Jul 02, 2021	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">6.84e-07</a>	ClinVar; gnomAD; Caciotti 2005 (PMID:15714521)	<a href="#">15714521</a> ; <a href="#">18524657</a>		<a href="#">1457739</a>
c.1311T>C	p.Asn437=	Exon 13	Likely Benign	Sep 05, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">6.84e-07</a>	ClinVar; gnomAD			<a href="#">2153157</a>
c.1312G>T	p.Gly438Ter	Exon 13	Likely Pathogenic	Mar 08, 2019	GM1 gangliosidosis type 2;Infantile GM1 gangliosidosis;GM1 gangliosidosis type 3		ClinVar			<a href="#">983740</a>

cDNA Change	Protein Change	Location	Pathogenicity	Classified	Phenotype	gnomAD AF	Sources	PMID(s)	PIN	ClinVar ID
c.1313G>A	p.Gly438Glu	Exon 13	Pathogenic	Jan 28, 2026	Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis type 3;Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis type 2;Infantile GM1 gangliosidosis; MPS-IV-B;GM1 gangliosidosis; Spondyloepiphyseal dysplasia; GM1 gangliosidosis	<a href="#">6.84e-07</a>	ClinVar; gnomAD; Hofer 2009 (PMID:19472408)	<a href="#">10841810</a> ; <a href="#">15986423</a> ; <a href="#">19472408</a> ; <a href="#">21497194</a> ; <a href="#">22033734</a> ; <a href="#">23831247</a> ; <a href="#">26646981</a> ; <a href="#">31216405</a> ; <a href="#">8922281</a>		<a href="#">940</a>
c.1313G>C	p.Gly438Ala	Exon 13	Likely Pathogenic	Dec 19, 2025	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar; Hofer 2009 (PMID:19472408)	<a href="#">10841810</a> ; <a href="#">19472408</a> ; <a href="#">21497194</a> ; <a href="#">23831247</a> ; <a href="#">26646981</a>		<a href="#">4789512</a>
c.1313G>T	p.Gly438Val	Exon 13	Likely Pathogenic	Nov 25, 2024	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar; Hofer 2009 (PMID:19472408)	<a href="#">10841810</a> ; <a href="#">19472408</a> ; <a href="#">21497194</a> ; <a href="#">23831247</a> ; <a href="#">26646981</a>		<a href="#">3753905</a>
c.1314A>C	p.Gly438=	Exon 13	Likely Benign	Dec 30, 2025	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">1.37e-06</a>	ClinVar; gnomAD			<a href="#">2139010</a>
c.1314A>G	p.Gly438=	Exon 13	Likely Benign	Nov 17, 2023	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">2953983</a>
c.1317C>G	p.Val439=	Exon 13	Likely Benign	Jan 12, 2026	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">6.84e-07</a>	ClinVar; gnomAD			<a href="#">1632034</a>
c.1317C>T	p.Val439=	Exon 13	Likely Benign	Apr 28, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">2935582</a>
c.1318C>T	p.His440Tyr	Exon 13	Pathogenic	Aug 15, 2019	Infantile GM1 gangliosidosis		ClinVar			<a href="#">827672</a>
c.1320C>T	p.His440=	Exon 13	Likely Benign	Feb 09, 2024	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">1.30e-05</a>	ClinVar; gnomAD			<a href="#">1622544</a>
c.1321G>A	p.Asp441Asn	Exon 13	Pathogenic	Dec 18, 2025	GM1 gangliosidosis type 3;Mucopolysaccharidosis; MPS-IV-B;Infantile GM1 gangliosidosis;GM1 gangliosidosis type 2; Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis; MPS-IV-B	<a href="#">4.79e-06</a>	ClinVar; LOVD; gnomAD; Santamaria 2006 (PMID:16941474)	<a href="#">16941474</a> ; <a href="#">17664528</a> ; <a href="#">21497194</a> ; <a href="#">22128166</a> ; <a href="#">28476546</a>		<a href="#">551496</a>
c.1322A>G	p.Asp441Gly	Exon 13	VUS	Apr 23, 2023	Infantile GM1 gangliosidosis;GM1 gangliosidosis type 2;GM1 gangliosidosis type 3;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">3893128</a>
c.1324C>T	p.Arg442Ter	Exon 13	P/LP	Mar 24, 2025	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis; GM1 gangliosidosis type 2;Mucopolysaccharidosis; MPS-IV-B;Infantile GM1 gangliosidosis;GM1 gangliosidosis type 3; MPS-IV-B	<a href="#">9.58e-06</a>	ClinVar; gnomAD	<a href="#">18524657</a>		<a href="#">1069817</a>
c.1325G>A	p.Arg442Gln	Exon 13	P/LP	Jan 01, 2026	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B; Infantile GM1 gangliosidosis;GM1 gangliosidosis type 3;GM1 gangliosidosis type 2;Mucopolysaccharidosis; Infantile GM1 gangliosidosis; GLB1-related disorder; GM1 gangliosidosis type 2; GM1 gangliosidosis type 3; Mucopolysaccharidosis	<a href="#">2.53e-05</a>	ClinVar; LOVD; gnomAD; Hofer 2009 (PMID:19472408)	<a href="#">16314480</a> ; <a href="#">18571950</a> ; <a href="#">19472408</a> ; <a href="#">20175788</a> ; <a href="#">21497194</a> ; <a href="#">21520340</a> ; <a href="#">27679996</a> ; <a href="#">31069529</a> ; <a href="#">37541188</a>		<a href="#">528328</a>
c.1326A>G	p.Arg442=	Exon 13	Likely Benign	Oct 29, 2025	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">1.16e-05</a>	ClinVar; gnomAD			<a href="#">705794</a>

cDNA Change	Protein Change	Location	Pathogenicity	Classified	Phenotype	gnomAD AF	Sources	PMID(s)	PIN	ClinVar ID
c.1329A>T	p.Ala443=	Exon 13	Likely Benign	May 26, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">6.84e-06</a>	ClinVar; gnomAD			<a href="#">2936963</a>
c.1333G>A	p.Val445Ile	Exon 13	VUS	Jul 25, 2022	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">1713712</a>
c.1336dup	p.Ala446fs	Exon 13	P/LP	Dec 10, 2025	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B; Mucopolysaccharidosis		ClinVar	<a href="#">18524657</a>		<a href="#">1405477</a>
c.1338T>C	p.Ala446=	Exon 13	Likely Benign	Feb 19, 2024	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">7.52e-06</a>	ClinVar; gnomAD			<a href="#">1975051</a>
c.1340_1341dup	p.Asp448fs	Exon 13	Pathogenic	Feb 16, 2024	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar	<a href="#">18524657</a>		<a href="#">2938645</a>
c.1342G>A	p.Asp448Asn	Exon 13	VUS	Sep 10, 2025	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">4.79e-06</a>	ClinVar; gnomAD	<a href="#">20175788</a> ; <a href="#">29439846</a>		<a href="#">4782561</a>
c.1343A>T	p.Asp448Val	Exon 13	P/LP	Oct 23, 2025	GM1 gangliosidosis; Mucopolysaccharidosis; MPS-IV-B; MPS-IV-B;GM1 gangliosidosis; Infantile GM1 gangliosidosis; GM1 gangliosidosis type 2;Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis type 3;Infantile GM1 gangliosidosis; GM1 gangliosidosis type 2	<a href="#">7.53e-06</a>	ClinVar; LOVD; gnomAD	<a href="#">20175788</a> ; <a href="#">21520340</a> ; <a href="#">23151865</a> ; <a href="#">23337983</a> ; <a href="#">29439846</a> ; <a href="#">33176815</a> ; <a href="#">35598274</a>		<a href="#">344790</a>
c.1345G>A		Exon 13	Not Classified			6.84e-07	LOVD; gnomAD			
c.1347+1G>A		Intron 13	Likely Pathogenic	Apr 07, 2023	GLB1-related disorder	<a href="#">1.37e-06</a>	ClinVar; gnomAD			<a href="#">2572285</a>
c.1347+1G>T		Intron 13	Likely Pathogenic	Feb 27, 2024	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis; GM1 gangliosidosis type 3;Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis type 2;Infantile GM1 gangliosidosis	<a href="#">6.84e-07</a>	ClinVar; gnomAD	<a href="#">16199547</a> ; <a href="#">18524657</a>		<a href="#">1943565</a>
c.1347+2del		Intron 13	Likely Pathogenic	Oct 11, 2025	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar	<a href="#">16199547</a> ; <a href="#">18524657</a>		<a href="#">4786382</a>
c.1347+7A>G		Intron 13	Likely Benign	Jan 14, 2023	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">2943193</a>
c.1347+9C>A		Intron 13	Likely Benign	Dec 12, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">6.84e-07</a>	ClinVar; gnomAD			<a href="#">2934467</a>
c.1347+9C>T		Intron 13	Likely Benign	Jul 18, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">4.11e-06</a>	ClinVar; gnomAD			<a href="#">1140500</a>
c.1347+10G>A		Intron 13	Conflicting	Jan 26, 2026	Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis; MPS-IV-B;GM1 gangliosidosis; GLB1-related disorder	<a href="#">0.00037</a>	ClinVar; gnomAD			<a href="#">344789</a>
c.1347+12C>T		Intron 13	Likely Benign	Apr 25, 2024	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">1.37e-06</a>	ClinVar; gnomAD			<a href="#">3751865</a>
c.1347+13T>G		Intron 13	Likely Benign	Jun 25, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">1.23e-05</a>	ClinVar; gnomAD			<a href="#">2924146</a>
c.1347+14C>T		Intron 13	Likely Benign	Jun 08, 2022	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">2002959</a>
c.1347+16G>C		Intron 13	Likely Benign	Mar 12, 2024	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">6.61e-06</a>	ClinVar; gnomAD			<a href="#">3751248</a>
c.1347+18A>C		Intron 13	Likely Benign	Jan 14, 2025	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">2.74e-06</a>	ClinVar; gnomAD			<a href="#">2924605</a>
c.1347+19C>T		Intron 13	Likely Benign	Feb 04, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">6.86e-07</a>	ClinVar; gnomAD			<a href="#">2932592</a>

cDNA Change	Protein Change	Location	Pathogenicity	Classified	Phenotype	gnomAD AF	Sources	PMID(s)	PIN	ClinVar ID
c.1347+20T>C		Intron 13	Likely Benign	Jan 16, 2024	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">2935113</a>
c.1347+194G>T		Intron 13	Benign	Mar 10, 2019			ClinVar			<a href="#">1245393</a>
c.1347+208A>G		Intron 13	Benign	Jul 15, 2018	Cholangiocarcinoma; Lung cancer		ClinVar			<a href="#">1231097</a>
c.1348-190G>T		Intron 13	Not Classified	-	Malignant tumor of esophagus		ClinVar			<a href="#">4295480</a>
c.1348-156C>A		Intron 13	Not Classified	-	Malignant lymphoma; large B-cell; diffuse		ClinVar			<a href="#">4295479</a>
c.1348-16G>A		Intron 13	Likely Benign	Jan 28, 2026	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">2.26e-05</a>	ClinVar; gnomAD			<a href="#">1535542</a>
c.1348-16del		Intron 13	Likely Benign	Jan 24, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">2934551</a>
c.1348-7C>G		Intron 13	Likely Benign	Dec 03, 2023	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">1.37e-06</a>	ClinVar; gnomAD			<a href="#">2954399</a>
c.1348-7C>T		Intron 13	Conflicting	Apr 18, 2025	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis; Mucopolysaccharidosis	<a href="#">3.42e-05</a>	ClinVar; gnomAD			<a href="#">902181</a>
c.1348-5dup		Intron 13	Benign	Nov 17, 2023	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">2953746</a>
c.1348-3C>G		Intron 13	Likely Pathogenic	Mar 11, 2024	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar	<a href="#">17576681</a> ; <a href="#">9536098</a>		<a href="#">948665</a>
c.1348-2A>G		Intron 13	Pathogenic	Oct 24, 2024	Mucopolysaccharidosis; MPS-IV-B	<a href="#">1.37e-06</a>	ClinVar; gnomAD	<a href="#">32219895</a>		<a href="#">4818311</a>
c.1348-1G>C		Intron 13	Not Classified				LOVD			
c.1350C>G	p.Ile450Met	Exon 14	VUS	Oct 09, 2024			ClinVar			<a href="#">4540847</a>
c.1350C>T	p.Ile450=	Exon 14	Likely Benign	Jan 26, 2023	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">4.79e-06</a>	ClinVar; gnomAD			<a href="#">2938798</a>
c.1351C>T	p.Pro451Ser	Exon 14	VUS	Oct 22, 2024	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">6.84e-07</a>	ClinVar; gnomAD			<a href="#">1922240</a>
c.1356G>A	p.Gln452=	Exon 14	Likely Benign	Jul 01, 2025	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">2.05e-05</a>	ClinVar; gnomAD			<a href="#">744135</a>
c.1356G>C	p.Gln452His	Exon 14	VUS	Feb 15, 2023	Inborn genetic diseases		ClinVar			<a href="#">2485304</a>
c.1360G>A	p.Val454Ile	Exon 14	VUS	Jun 24, 2025	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">8.21e-06</a>	ClinVar; gnomAD			<a href="#">1390459</a>
c.1363del	p.Glu456fs	Exon 14	Pathogenic	Nov 13, 2025	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar	<a href="#">18524657</a>		<a href="#">4786703</a>
c.1368G>A	p.Glu456=	Exon 14	Likely Benign	Apr 13, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">1.37e-06</a>	ClinVar; gnomAD			<a href="#">1078659</a>
c.1369C>T	p.Arg457Ter	Exon 14	Pathogenic	Oct 29, 2024	Infantile GM1 gangliosidosis; GM1 gangliosidosis type 3;Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis type 2;Infantile GM1 gangliosidosis; Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis; GM1 gangliosidosis; GM1 gangliosidosis type 2; MPS-IV-B	<a href="#">8.21e-06</a>	ClinVar; gnomAD; Bidchol 2015 (PMID:25936995)	<a href="#">17309651</a> ; <a href="#">18524657</a> ; <a href="#">1909089</a> ; <a href="#">21520340</a> ; <a href="#">25936995</a> ; <a href="#">26169295</a> ; <a href="#">28976722</a> ; <a href="#">30548430</a> ; <a href="#">33038107</a>		<a href="#">924</a>

cDNA Change	Protein Change	Location	Pathogenicity	Classified	Phenotype	gnomAD AF	Sources	PMID(s)	PIN	ClinVar ID
c.1370G>A	p.Arg457Gln	Exon 14	P/LP	Jan 30, 2026	GM1 gangliosidosis type 3; GM1 gangliosidosis type 3; GM1 gangliosidosis type 2; Infantile GM1 gangliosidosis; GM1 gangliosidosis; Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis; MPS-IV-B	<a href="#">6.84e-06</a>	ClinVar; gnomAD	<a href="#">11504597</a> ; <a href="#">1353343</a> ; <a href="#">16617000</a> ; <a href="#">1907800</a> ; <a href="#">20826101</a> ; <a href="#">21520340</a> ; <a href="#">31761138</a> ; <a href="#">33737400</a>		<a href="#">930</a>
c.1370_1372dup	p.Arg457dup	Exon 14	VUS	Dec 02, 2021	GM1 gangliosidosis; Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">1417992</a>
c.1375_1377del	p.Asn459del	Exon 14	VUS	Dec 08, 2022	GM1 gangliosidosis; Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">2941656</a>
c.1376A>G	p.Asn459Ser	Exon 14	VUS	Apr 28, 2021	Inborn genetic diseases	<a href="#">2.33e-05</a>	ClinVar; gnomAD			<a href="#">2365863</a>
c.1377T>C	p.Asn459=	Exon 14	Likely Benign	Mar 08, 2021	Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis	<a href="#">4.10e-06</a>	ClinVar; gnomAD			<a href="#">1621901</a>
c.1379_1380del	p.Val460fs	Exon 14	Likely Pathogenic	Dec 16, 2021	Infantile GM1 gangliosidosis; GM1 gangliosidosis type 2; GM1 gangliosidosis type 3		ClinVar			<a href="#">1726335</a>
c.1380G>A	p.Val460=	Exon 14	Likely Benign	Aug 30, 2023	Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis	<a href="#">1.31e-05</a>	ClinVar; gnomAD			<a href="#">2933756</a>
c.1384A>G	p.Thr462Ala	Exon 14	VUS	Feb 09, 2022	Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis		ClinVar			<a href="#">2141465</a>
c.1387_1388del	p.Leu463fs	Exon 14	Pathogenic	Jan 24, 2023	GM1 gangliosidosis; Mucopolysaccharidosis; MPS-IV-B		ClinVar	<a href="#">18524657</a>		<a href="#">2943569</a>
c.1388T>C	p.Leu463Pro	Exon 14	Pathogenic	Mar 10, 2023	GM1 gangliosidosis; Mucopolysaccharidosis; MPS-IV-B		ClinVar	<a href="#">33240792</a>		<a href="#">1057305</a>
c.1389G>T	p.Leu463=	Exon 14	Likely Benign	Mar 27, 2022	GM1 gangliosidosis; Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">2112803</a>
c.1393A>G	p.Ile465Val	Exon 14	VUS	Sep 26, 2025	Inborn genetic diseases	<a href="#">4.79e-06</a>	ClinVar; gnomAD			<a href="#">4620829</a>
c.1396delinsGGG	p.Thr466fs	Exon 14	Likely Pathogenic	Apr 28, 2025	Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">4818313</a>
c.1398_1399del	p.Gly467fs	Exon 14	P/LP	Jan 18, 2026	GM1 gangliosidosis; Mucopolysaccharidosis; MPS-IV-B; Mucopolysaccharidosis		ClinVar	<a href="#">18524657</a>		<a href="#">2089127</a>
c.1416T>A	p.Thr472=	Exon 14	Likely Benign	Dec 09, 2021	Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis	<a href="#">6.84e-07</a>	ClinVar; gnomAD			<a href="#">2039061</a>
c.1417del	p.Leu473fs	Exon 14	Pathogenic	Oct 03, 2023	Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis		ClinVar	<a href="#">18524657</a>		<a href="#">2052850</a>
c.1421A>G	p.Asp474Gly	Exon 14	VUS	-	Infantile GM1 gangliosidosis	<a href="#">6.84e-07</a>	ClinVar; gnomAD			<a href="#">1878580</a>
c.1423C>T	p.Leu475Phe	Exon 14	VUS	Dec 11, 2023	Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis	<a href="#">4.79e-06</a>	ClinVar; gnomAD			<a href="#">2077647</a>
c.1426C>G	p.Leu476Val	Exon 14	VUS	Apr 24, 2023	GM1 gangliosidosis; Mucopolysaccharidosis; MPS-IV-B	<a href="#">2.05e-06</a>	ClinVar; gnomAD			<a href="#">1383281</a>
c.1426C>T	p.Leu476=	Exon 14	Conflicting	Feb 01, 2026	Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis; MPS-IV-B; GM1 gangliosidosis; Infantile GM1 gangliosidosis; GLB1-related disorder	<a href="#">0.00039</a>	ClinVar; LOVD; gnomAD			<a href="#">92897</a>
c.1428G>A	p.Leu476=	Exon 14	Likely Benign	May 14, 2024	GM1 gangliosidosis; Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">3756341</a>
c.1428G>T	p.Leu476=	Exon 14	Not Classified	-	Malignant tumor of urinary bladder		ClinVar			<a href="#">4295478</a>
c.1430T>G		Exon 14	Not Classified				LOVD			

cDNA Change	Protein Change	Location	Pathogenicity	Classified	Phenotype	gnomAD AF	Sources	PMID(s)	PIN	ClinVar ID
c.1437C>A	p.Asn479Lys	Exon 14	VUS	Feb 24, 2024	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">6.84e-07</a>	ClinVar; gnomAD			<a href="#">2192335</a>
c.1438A>G	p.Met480Val	Exon 14	Likely Pathogenic	May 22, 2024	GM1 gangliosidosis type 3;Mucopolysaccharidosis; MPS-IV-B;Infantile GM1 gangliosidosis;GM1 gangliosidosis type 2; Mucopolysaccharidosis; MPS-IV-B	<a href="#">4.10e-06</a>	ClinVar; LOVD; gnomAD	<a href="#">21520340</a> ; <a href="#">23337983</a>		<a href="#">553636</a>
c.1441G>T	p.Gly481Ter	Exon 14	Pathogenic	Oct 25, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">2.74e-06</a>	ClinVar; LOVD; gnomAD; Santamaria 2006 (PMID:16941474)	<a href="#">16941474</a> ; <a href="#">18524657</a>		<a href="#">2925350</a>
c.1442G>A	p.Gly481Glu	Exon 14	Conflicting	Aug 28, 2025	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">8.21e-06</a>	ClinVar; gnomAD; OMIM; NTSAD; Cure GM1 AllStripes PIN		1	<a href="#">1430566</a>
c.1444C>T	p.Arg482Cys	Exon 14	P/LP	Mar 27, 2025	Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis type 3;Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis type 2;Infantile GM1 gangliosidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">8.21e-06</a>	ClinVar; gnomAD	<a href="#">15943552</a> ; <a href="#">1928092</a> ; <a href="#">22128166</a> ; <a href="#">23337983</a> ; <a href="#">32005694</a> ; <a href="#">7586649</a>		<a href="#">938</a>
c.1445G>A	p.Arg482His	Exon 14	Pathogenic	Nov 16, 2025	Infantile GM1 gangliosidosis; Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis; MPS-IV-B;GM1 gangliosidosis; GM1 gangliosidosis type 3;Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis type 2;Infantile GM1 gangliosidosis	<a href="#">1.92e-05</a>	ClinVar; LOVD; gnomAD; Caciotti 2005 (PMID:15714521); Bidchol 2015 (PMID:25936995)	<a href="#">10737981</a> ; <a href="#">1487238</a> ; <a href="#">15365997</a> ; <a href="#">15714521</a> ; <a href="#">15943552</a> ; <a href="#">17221873</a> ; <a href="#">1928092</a> ; <a href="#">21497194</a> ; <a href="#">21520340</a> ; <a href="#">22128166</a> ; <a href="#">25936995</a> ; <a href="#">7586649</a> ; <a href="#">8500799</a>		<a href="#">932</a>
c.1445G>T	p.Arg482Leu	Exon 14	VUS	Oct 15, 2021	Inborn genetic diseases	<a href="#">3.42e-06</a>	ClinVar; gnomAD			<a href="#">2245552</a>
c.1449G>T	p.Val483=	Exon 14	Likely Benign	Feb 04, 2021	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">1128621</a>
c.1451A>G	p.Asn484Ser	Exon 14	Not Classified	-	Nonpapillary renal cell carcinoma		ClinVar			<a href="#">4295477</a>
c.1452C>G	p.Asn484Lys	Exon 14	P/LP	Apr 21, 2025	GM1 gangliosidosis type 3;Mucopolysaccharidosis; MPS-IV-B;Infantile GM1 gangliosidosis;GM1 gangliosidosis type 2; Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis; MPS-IV-B	<a href="#">1.03e-05</a>	ClinVar; gnomAD	<a href="#">10841810</a> ; <a href="#">12393180</a> ; <a href="#">22128166</a> ; <a href="#">23337983</a>		<a href="#">553506</a>
c.1454A>C		Exon 14	Not Classified				LOVD			
c.1454A>G	p.Tyr485Cys	Exon 14	P/LP	Oct 16, 2025	Infantile GM1 gangliosidosis;GM1 gangliosidosis type 2;Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis type 3; GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B; Mucopolysaccharidosis	<a href="#">6.84e-07</a>	ClinVar; gnomAD	<a href="#">23151865</a> ; <a href="#">28595941</a> ; <a href="#">29396849</a> ; <a href="#">34539759</a> ; <a href="#">38313286</a> ; <a href="#">38702915</a>		<a href="#">555363</a>
c.1455T>C	p.Tyr485=	Exon 14	Likely Benign	Nov 05, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">6.84e-07</a>	ClinVar; gnomAD			<a href="#">2933744</a>
c.1456G>A		Exon 14	Not Classified			6.84e-07	LOVD; gnomAD			
c.1456_1466dup	p.Ile489fs	Exon 14	P/LP	Nov 25, 2025	GM1 gangliosidosis type 2;Infantile GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis type 3; Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar	<a href="#">18524657</a> ; <a href="#">23757202</a>		<a href="#">92898</a>

cDNA Change	Protein Change	Location	Pathogenicity	Classified	Phenotype	gnomAD AF	Sources	PMID(s)	PIN	ClinVar ID
c.1465_1466del	p.Ile489fs	Exon 14	Likely Pathogenic	Jan 11, 2021	Infantile GM1 gangliosidosis		ClinVar			<a href="#">1324475</a>
c.1467C>A	p.Ile489=	Exon 14	Likely Benign	Jul 08, 2023	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">2087508</a>
c.1468_1470del	p.Asn490del	Exon 14	Likely Pathogenic	Mar 24, 2025	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B; Mucopolysaccharidosis	<a href="#">6.84e-07</a>	ClinVar; gnomAD	<a href="#">26646981</a> ; <a href="#">30187681</a>		<a href="#">835718</a>
c.1470C>T	p.Asn490=	Exon 14	Likely Benign	Apr 18, 2025	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">6.16e-06</a>	ClinVar; gnomAD			<a href="#">1154103</a>
c.1471G>A	p.Asp491Asn	Exon 14	Conflicting	Oct 19, 2025	Infantile GM1 gangliosidosis;GM1 gangliosidosis type 2;GM1 gangliosidosis type 3;Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis;Mucopolysaccharidosis; Mucopolysaccharidosis; Infantile GM1 gangliosidosis	<a href="#">8.89e-06</a>	ClinVar; LOVD; gnomAD	<a href="#">10338095</a> ; <a href="#">17309651</a> ; <a href="#">21520340</a> ; <a href="#">23337983</a> ; <a href="#">31761138</a> ; <a href="#">33737400</a>		<a href="#">558272</a>
c.1471G>T	p.Asp491Tyr	Exon 14	Likely Pathogenic	Jun 07, 2024	GM1 gangliosidosis; GM1 gangliosidosis type 3;Mucopolysaccharidosis; MPS-IV-B;Infantile GM1 gangliosidosis;GM1 gangliosidosis type 2		ClinVar	<a href="#">17309651</a> ; <a href="#">23337983</a> ; <a href="#">31761138</a>		<a href="#">3339727</a>
c.1479G>T	p.Lys493Asn	Exon 14	Conflicting	Sep 04, 2025	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis; Infantile GM1 gangliosidosis	<a href="#">6.84e-07</a>	ClinVar; gnomAD; Bidchol 2015 (PMID:25936995); Cure GM1 AllStripes PIN	<a href="#">17576681</a> ; <a href="#">25936995</a> ; <a href="#">32506457</a> ; <a href="#">9536098</a>	1	<a href="#">654251</a>
c.1479+1G>A		Intron 14	Pathogenic	May 22, 2023	Infantile GM1 gangliosidosis;GM1 gangliosidosis type 2;Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis type 3; GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">6.16e-06</a>	ClinVar; gnomAD; Santamaria 2006 (PMID:16941474)	<a href="#">16199547</a> ; <a href="#">16941474</a> ; <a href="#">18524657</a>		<a href="#">557669</a>
c.1479+1G>C		Intron 14	Pathogenic	Aug 30, 2021	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar; Santamaria 2006 (PMID:16941474)	<a href="#">16941474</a>		<a href="#">1364568</a>
c.1479+2T>C		Intron 14	Not Classified	-	Thyroid cancer; nonmedullary; 1		ClinVar			<a href="#">4295476</a>
c.1479+2T>G		Intron 14	Not Classified	-	Thyroid cancer; nonmedullary; 1		ClinVar			<a href="#">4295475</a>
c.1479+7C>T		Intron 14	Likely Benign	Mar 18, 2023	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">2945432</a>
c.1479+11C>T		Intron 14	Conflicting	Jan 14, 2026	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B; Mucopolysaccharidosis; GM1 gangliosidosis	<a href="#">0.00030</a>	ClinVar; gnomAD			<a href="#">902180</a>
c.1479+13A>G		Intron 14	Likely Benign	Oct 27, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">6.84e-07</a>	ClinVar; gnomAD			<a href="#">2937606</a>
c.1479+17T>C		Intron 14	Likely Benign	Feb 18, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">5.48e-06</a>	ClinVar; gnomAD			<a href="#">2926665</a>
c.1479+19T>C		Intron 14	Likely Benign	Dec 16, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">2923541</a>
c.1479+20C>G		Intron 14	Likely Benign	Jun 17, 2023	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">2941257</a>
c.1479+128C>T		Intron 14	Not Classified	-	Malignant tumor of urinary bladder		ClinVar			<a href="#">4295474</a>
c.1479+204A>G		Intron 14	Benign	Sep 06, 2018			ClinVar			<a href="#">1246205</a>
c.1480-320C>A		Intron 14	Benign	Apr 12, 2019			ClinVar			<a href="#">1222508</a>
c.1480-295A>G		Intron 14	Likely Benign	Sep 06, 2018			ClinVar			<a href="#">1216917</a>
c.1480-272T>C		Intron 14	Benign	Jan 12, 2020			ClinVar			<a href="#">1221492</a>

cDNA Change	Protein Change	Location	Pathogenicity	Classified	Phenotype	gnomAD AF	Sources	PMID(s)	PIN	ClinVar ID
c.1480-149T>C		Intron 14	Benign	Oct 17, 2018			ClinVar			<a href="#">1248379</a>
c.1480-74C>G		Intron 14	Benign	Sep 04, 2018		<a href="#">0.05560</a>	ClinVar; gnomAD			<a href="#">1238885</a>
c.1480-20G>C		Intron 14	Likely Benign	May 26, 2025	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">4784966</a>
c.1480-16T>C		Intron 14	Likely Benign	Feb 01, 2024	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">2.05e-06</a>	ClinVar; gnomAD			<a href="#">2932591</a>
c.1480-12T>G		Intron 14	Likely Benign	Feb 22, 2024	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">6.84e-06</a>	ClinVar; gnomAD			<a href="#">2923102</a>
c.1480-9G>A		Intron 14	Likely Benign	Oct 24, 2021	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">1578791</a>
c.1480-8C>G		Intron 14	Conflicting	Jun 27, 2025	Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">9.79e-05</a>	ClinVar; gnomAD			<a href="#">344788</a>
c.1480-8del		Intron 14	Likely Benign	Mar 14, 2024	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">6.16e-06</a>	ClinVar; gnomAD			<a href="#">1085585</a>
c.1480-5T>C		Intron 14	Likely Benign	Feb 07, 2024	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">1.37e-06</a>	ClinVar; gnomAD			<a href="#">1137352</a>
c.1480-4G>T		Intron 14	Likely Benign	Feb 03, 2024	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">2.12e-05</a>	ClinVar; gnomAD			<a href="#">1138305</a>
c.1480-2A>G		Intron 14	Pathogenic	Sep 27, 2024	GM1-gangliosidosis; type I; with cardiac involvement; Mucopolysaccharidosis; MPS-IV-B; MPS-IV-B;GM1 gangliosidosis; Infantile GM1 gangliosidosis; GM1 gangliosidosis type 3;Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis type 2;Infantile GM1 gangliosidosis; GM1 gangliosidosis; Uveal melanoma	<a href="#">6.16e-06</a>	ClinVar; gnomAD; Santamaria 2006 (PMID:16941474)	<a href="#">10737981</a> ; <a href="#">16941474</a> ; <a href="#">21497194</a> ; <a href="#">25557439</a> ; <a href="#">30809705</a>		<a href="#">946</a>
c.1480-2A>T		Intron 14	Pathogenic	Sep 27, 2024	Mucopolysaccharidosis; MPS-IV-B		ClinVar; Santamaria 2006 (PMID:16941474)	<a href="#">10737981</a> ; <a href="#">16941474</a>		<a href="#">4818315</a>
c.1480-1G>A		Intron 14	Likely Pathogenic	Apr 02, 2024	Mucopolysaccharidosis; MPS-IV-B	<a href="#">6.84e-07</a>	ClinVar; gnomAD	<a href="#">31776384</a>		<a href="#">4818314</a>
c.1480-1G>C		Intron 14	Pathogenic	Sep 17, 2024	GM1 gangliosidosis type 2		ClinVar			<a href="#">4079395</a>
c.1481G>A	p.Gly494Asp	Exon 15	VUS	Aug 06, 2024			ClinVar			<a href="#">3366464</a>
c.1481G>T	p.Gly494Val	Exon 15	P/LP	Aug 01, 2025	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis type 2;GM1 gangliosidosis type 3;Infantile GM1 gangliosidosis; MPS-IV-B;GM1 gangliosidosis; MPS-IV-B	<a href="#">1.37e-06</a>	ClinVar; gnomAD; Santamaria 2006 (PMID:16941474)	<a href="#">16941474</a> ; <a href="#">20920281</a> ; <a href="#">30267299</a>		<a href="#">550166</a>
c.1491_1492insTTTAAATTTATTTATTGATAATCTTGGTGTTCACACAGAGGGGATTTGGCAGGGTCATGGACNNNNNNNNNTTTTATTTTTTGAGACGGAGTCTGCTCTGTTGCTGGGTGAGTGCAGTGGTGCAATCTCGGCTCACTGCAACCGAGTTTGGTTTCTT	p.Ser497_Asn498insPheLysPhelleLeuLeulleLeuGlyCysPheSerGlnArgGlylleTrpGlnGlyHisGlyThrXaaXaaXaaPheTyrPheLeuArgArgSerLeuAlaLeuLeuSerGlyTrpSerAlaValValGlnSerArgLeuThrAlaThrArgValTrpPheLeu	Exon 15	Pathogenic	Mar 08, 2022	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar	<a href="#">18524657</a> ; <a href="#">19763152</a> ; <a href="#">20307669</a> ; <a href="#">22406018</a>		<a href="#">2107697</a>
c.1495C>T	p.Leu499=	Exon 15	Likely Benign	Jun 24, 2021	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">1082445</a>

cDNA Change	Protein Change	Location	Pathogenicity	Classified	Phenotype	gnomAD AF	Sources	PMID(s)	PIN	ClinVar ID
c.1498A>G	p.Thr500Ala	Exon 15	Pathogenic	Jan 18, 2026	Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis type 3; Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis type 2; Infantile GM1 gangliosidosis; MPS-IV-B; GM1 gangliosidosis; GM1 gangliosidosis; GM1 gangliosidosis type 3	<a href="#">1.64e-05</a>	ClinVar; gnomAD; Hofer 2009 (PMID:19472408)	<a href="#">11511921</a> ; <a href="#">12393180</a> ; <a href="#">17664528</a> ; <a href="#">19472408</a> ; <a href="#">21520340</a> ; <a href="#">22128166</a> ; <a href="#">23337983</a> ; <a href="#">26108645</a> ; <a href="#">33558080</a> ; <a href="#">8922281</a>		<a href="#">942</a>
c.1498A>T	p.Thr500Ser	Exon 15	Likely Pathogenic	Jul 31, 2023	Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis		ClinVar; Hofer 2009 (PMID:19472408)	<a href="#">12393180</a> ; <a href="#">17664528</a> ; <a href="#">19472408</a> ; <a href="#">26108645</a>		<a href="#">2052783</a>
c.1500T>G	p.Thr500=	Exon 15	Likely Benign	Jun 16, 2023	GM1 gangliosidosis; Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">2948930</a>
c.1500_1503del	p.Leu501fs	Exon 15	Likely Pathogenic	Jul 14, 2017	GM1 gangliosidosis type 3; Mucopolysaccharidosis; MPS-IV-B; Infantile GM1 gangliosidosis; GM1 gangliosidosis type 2		ClinVar			<a href="#">552879</a>
c.1501C>G	p.Leu501Val	Exon 15	VUS	Jul 17, 2023	Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis; GM1 gangliosidosis	<a href="#">4.65e-05</a>	ClinVar; gnomAD			<a href="#">2047843</a>
c.1505G>A	p.Ser502Asn	Exon 15	VUS	Feb 24, 2024	Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis		ClinVar			<a href="#">2197890</a>
c.1508C>A	p.Ser503Tyr	Exon 15	VUS	Aug 02, 2022	Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis	<a href="#">6.84e-07</a>	ClinVar; gnomAD			<a href="#">1962692</a>
c.1509C>G	p.Ser503=	Exon 15	Likely Benign	Dec 28, 2025	GM1 gangliosidosis; Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">4787150</a>
c.1510_1511insGA	p.Asn504fs	Exon 15	Pathogenic	Apr 21, 2025	Infantile GM1 gangliosidosis; GM1 gangliosidosis type 2; Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis type 3; GM1 gangliosidosis; Mucopolysaccharidosis; MPS-IV-B; GLB1-related disorder	<a href="#">6.84e-07</a>	ClinVar; gnomAD; Santamaria 2006 (PMID:16941474)	<a href="#">16941474</a> ; <a href="#">17221873</a> ; <a href="#">17309651</a> ; <a href="#">23430803</a>		<a href="#">555117</a>
c.1511A>G	p.Asn504Ser	Exon 15	Likely Benign	Jan 13, 2026	Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis	<a href="#">5.13e-05</a>	ClinVar; gnomAD			<a href="#">1106585</a>
c.1512T>C	p.Asn504=	Exon 15	Likely Benign	Oct 12, 2021	Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis	<a href="#">6.84e-07</a>	ClinVar; gnomAD			<a href="#">1109445</a>
c.1515C>T	p.Ile505=	Exon 15	Likely Benign	Jan 14, 2020	Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis		ClinVar			<a href="#">1143827</a>
c.1516C>A	p.Leu506Ile	Exon 15	VUS	Aug 27, 2021	Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis	<a href="#">6.84e-07</a>	ClinVar; gnomAD			<a href="#">1959278</a>
c.1516C>T	p.Leu506Phe	Exon 15	VUS	Jul 08, 2024			ClinVar			<a href="#">3572599</a>
c.1518C>G	p.Leu506=	Exon 15	Likely Benign	Aug 27, 2021	GM1 gangliosidosis; Mucopolysaccharidosis; MPS-IV-B	<a href="#">6.84e-07</a>	ClinVar; gnomAD			<a href="#">1580517</a>
c.1518C>T	p.Leu506=	Exon 15	Likely Benign	Jan 24, 2026	GM1 gangliosidosis; Mucopolysaccharidosis; MPS-IV-B	<a href="#">1.30e-05</a>	ClinVar; gnomAD			<a href="#">711278</a>
c.1520C>T	p.Thr507Met	Exon 15	VUS	Feb 11, 2025	Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis; Inborn genetic diseases	<a href="#">9.58e-06</a>	ClinVar; gnomAD			<a href="#">2044218</a>
c.1521G>A	p.Thr507=	Exon 15	Likely Benign	Dec 22, 2025	Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis; Melanoma; Colon adenocarcinoma	<a href="#">4.38e-05</a>	ClinVar; gnomAD			<a href="#">1128145</a>

cDNA Change	Protein Change	Location	Pathogenicity	Classified	Phenotype	gnomAD AF	Sources	PMID(s)	PIN	ClinVar ID
c.1527G>A	p.Trp509Ter	Exon 15	Likely Pathogenic	May 20, 2024	Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">4818316</a>
c.1527G>T	p.Trp509Cys	Exon 15	P/LP	Nov 20, 2025	Mucopolysaccharidosis; MPS-IV-B; MPS-IV-B;GM1 gangliosidosis	<a href="#">6.16e-06</a>	ClinVar; gnomAD	<a href="#">16538002</a> ; <a href="#">1928092</a> ; <a href="#">23337983</a> ; <a href="#">37152986</a> ; <a href="#">9203065</a>		<a href="#">933</a>
c.1529C>T	p.Thr510Met	Exon 15	Conflicting	Jan 19, 2026	GM1 gangliosidosis; Mucopolysaccharidosis; MPS-IV-B; MPS-IV-B;GM1 gangliosidosis; Inborn genetic diseases	<a href="#">6.57e-05</a>	ClinVar; gnomAD			<a href="#">344787</a>
c.1530G>A	p.Thr510=	Exon 15	Conflicting	Jan 28, 2026	Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">0.00026</a>	ClinVar; LOVD; gnomAD			<a href="#">344786</a>
c.1533C>G	p.Ile511Met	Exon 15	VUS	May 07, 2025		<a href="#">2.05e-06</a>	ClinVar; gnomAD			<a href="#">4540846</a>
c.1536T>C	p.Phe512=	Exon 15	Likely Benign	Dec 14, 2025	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">9.65e-05</a>	ClinVar; gnomAD			<a href="#">1095288</a>
c.1537C>T	p.Pro513Ser	Exon 15	VUS	Jul 01, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">2.05e-06</a>	ClinVar; gnomAD			<a href="#">2151807</a>
c.1545C>T	p.Asp515=	Exon 15	Likely Benign	Jan 27, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">1566997</a>
c.1556C>T	p.Ala519Val	Exon 15	VUS	Aug 15, 2022	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">1417767</a>
c.1557A>G	p.Ala519=	Exon 15	Likely Benign	Dec 21, 2025	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">4.10e-06</a>	ClinVar; gnomAD			<a href="#">2160273</a>
c.1561=	p.Cys521=	Exon 15	Benign	Feb 04, 2026	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar; LOVD			<a href="#">256024</a>
c.1561T>A	p.Cys521Ser	Exon 15	Not Classified	-	Nonpapillary renal cell carcinoma		ClinVar			<a href="#">4295473</a>
c.1561T>C	p.Cys521Arg	Exon 15	Benign	Feb 04, 2026	Infantile GM1 gangliosidosis; Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis; GM1 gangliosidosis type 2; MPS-IV-B; GM1 gangliosidosis type 3	<a href="#">0.99300</a>	ClinVar; gnomAD			<a href="#">167145</a>
c.1561_1562inv	p.Cys521His	Exon 15	VUS	Sep 27, 2022	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">1005326</a>
c.1562G>A	p.Cys521Tyr	Exon 15	VUS	May 12, 2024	Inborn genetic diseases; GM1 gangliosidosis; Mucopolysaccharidosis; MPS-IV-B	<a href="#">2.19e-05</a>	ClinVar; gnomAD			<a href="#">899399</a>
c.1566C>T	p.Ser522=	Exon 15	Likely Benign	May 14, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">1664749</a>
c.1568A>G	p.His523Arg	Exon 15	VUS	Dec 04, 2025	Inborn genetic diseases; GM1 gangliosidosis; Mucopolysaccharidosis; MPS-IV-B	<a href="#">5.47e-06</a>	ClinVar; gnomAD			<a href="#">899398</a>
c.1570C>T	p.Leu524=	Exon 15	Likely Benign	Aug 14, 2023	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">2950971</a>
c.1572G>C	p.Leu524=	Exon 15	Likely Benign	Dec 22, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">6.16e-06</a>	ClinVar; LOVD; gnomAD			<a href="#">1529144</a>
c.1574_1583del	p.Gly525fs	Exon 15	Pathogenic	Mar 09, 2021	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar	<a href="#">17309651</a>		<a href="#">1434135</a>
c.1576_1583dup	p.His529fs	Exon 15	Pathogenic	Aug 27, 2021	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar; Santamaria 2006 (PMID:16941474)	<a href="#">16941474</a> ; <a href="#">17309651</a> ; <a href="#">17664528</a> ; <a href="#">23430803</a>		<a href="#">1378674</a>

cDNA Change	Protein Change	Location	Pathogenicity	Classified	Phenotype	gnomAD AF	Sources	PMID(s)	PIN	ClinVar ID
c.1577G>A	p.Gly526Asp	Exon 15	Likely Benign	Feb 04, 2026	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B; Intellectual disability	<a href="#">0.00050</a>	ClinVar; LOVD; gnomAD	<a href="#">21956720</a> ; <a href="#">25157020</a> ; <a href="#">34131312</a> ; <a href="#">34211152</a>		<a href="#">558919</a>
c.1577del	p.Gly526fs	Exon 15	P/LP	Oct 30, 2025	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B; Mucopolysaccharidosis	<a href="#">1.37e-06</a>	ClinVar; gnomAD; Santamaria 2006 (PMID:16941474)	<a href="#">16941474</a> ; <a href="#">17309651</a> ; <a href="#">17664528</a> ; <a href="#">23430803</a>		<a href="#">2942836</a>
c.1577dup	p.Trp527fs	Exon 15	Pathogenic	Dec 14, 2025	Infantile GM1 gangliosidosis; Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis; GM1 gangliosidosis type 2; GM1 gangliosidosis; GM1 gangliosidosis type 3; GLB1-related disorder; GM1 gangliosidosis type 2;GM1 gangliosidosis type 3;Mucopolysaccharidosis; MPS-IV-B;Infantile GM1 gangliosidosis; MPS-IV-B	<a href="#">5.47e-06</a>	ClinVar; gnomAD; Santamaria 2006 (PMID:16941474); Bidchol 2015 (PMID:25936995)	<a href="#">10338095</a> ; <a href="#">15986423</a> ; <a href="#">16941474</a> ; <a href="#">17309651</a> ; <a href="#">18524657</a> ; <a href="#">21637542</a> ; <a href="#">25936995</a> ; <a href="#">26108645</a> ; <a href="#">31761138</a>		<a href="#">202191</a>
c.1580G>A	p.Trp527Ter	Exon 15	Pathogenic	Aug 26, 2025	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis; MPS-IV-B	<a href="#">4.79e-06</a>	ClinVar; gnomAD; Santamaria 2006 (PMID:16941474)	<a href="#">16941474</a> ; <a href="#">18524657</a> ; <a href="#">21214877</a>		<a href="#">1076248</a>
c.1582G>T	p.Gly528Ter	Exon 15	Pathogenic	Nov 21, 2023	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar	<a href="#">18524657</a> ; <a href="#">35614200</a>		<a href="#">2954097</a>
c.1583del	p.Gly528fs	Exon 15	Pathogenic	Nov 24, 2023	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar; Santamaria 2006 (PMID:16941474)	<a href="#">16941474</a> ; <a href="#">17309651</a> ; <a href="#">17664528</a> ; <a href="#">23430803</a>		<a href="#">2954179</a>
c.1584A>C	p.Gly528=	Exon 15	Likely Benign	Jun 17, 2019	GLB1-related disorder		ClinVar			<a href="#">3033373</a>
c.1584A>G	p.Gly528=	Exon 15	Likely Benign	Jul 25, 2023	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">6.84e-07</a>	ClinVar; gnomAD			<a href="#">2950237</a>
c.1585C>T	p.His529Tyr	Exon 15	VUS	Oct 02, 2023	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">9.58e-06</a>	ClinVar; gnomAD			<a href="#">2174616</a>
c.1587_1590dup	p.Asp531delinsProTer	Exon 15	Pathogenic	Nov 01, 2021	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar	<a href="#">18524657</a>		<a href="#">1457406</a>
c.1588C>T	p.Arg530Cys	Exon 15	Conflicting	Jan 28, 2026	GM1 gangliosidosis type 2;Infantile GM1 gangliosidosis;GM1 gangliosidosis type 3;Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis;Mucopolysaccharidosis; GLB1-related disorder; Inborn genetic diseases	<a href="#">9.99e-05</a>	ClinVar; LOVD; gnomAD			<a href="#">450616</a>
c.1589G>A	p.Arg530His	Exon 15	VUS	-		<a href="#">7.52e-06</a>	ClinVar; gnomAD			<a href="#">1050609</a>
c.1593C>T	p.Asp531=	Exon 15	Likely Benign	Sep 18, 2024	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">3.42e-06</a>	ClinVar; gnomAD			<a href="#">1101939</a>
c.1593_1594inv	p.Ser532Gly	Exon 15	Likely Benign	Mar 20, 2024	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">1546229</a>
c.1594A>G	p.Ser532Gly	Exon 15	Likely Benign	Feb 04, 2026	Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis; MPS-IV-B;GM1 gangliosidosis; Infantile GM1 gangliosidosis; GLB1-related disorder	<a href="#">0.05550</a>	ClinVar; LOVD; gnomAD; Santamaria 2006 (PMID:16941474)	<a href="#">10338095</a> ; <a href="#">10839995</a> ; <a href="#">16466959</a> ; <a href="#">16941474</a> ; <a href="#">17661814</a> ; <a href="#">17664528</a> ; <a href="#">21497194</a> ; <a href="#">21637542</a> ; <a href="#">22784478</a> ; <a href="#">23046582</a> ; <a href="#">25600812</a> ; <a href="#">27750150</a>		<a href="#">92899</a>
c.1598_1601dup	p.His534fs	Exon 15	Pathogenic	Feb 17, 2024	Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis type 2;GM1 gangliosidosis type 3;Mucopolysaccharidosis; MPS-IV-B;Infantile GM1 gangliosidosis	<a href="#">6.84e-07</a>	ClinVar; gnomAD; Hofer 2009 (PMID:19472408)	<a href="#">19472408</a>		<a href="#">1683247</a>

cDNA Change	Protein Change	Location	Pathogenicity	Classified	Phenotype	gnomAD AF	Sources	PMID(s)	PIN	ClinVar ID
c.1599C>A	p.Gly533=	Exon 15	Likely Benign	Sep 10, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">2.53e-05</a>	ClinVar; gnomAD			<a href="#">2147504</a>
c.1601A>G	p.His534Arg	Exon 15	Conflicting	Dec 29, 2025	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis; Mucopolysaccharidosis; Inborn genetic diseases	<a href="#">6.84e-05</a>	ClinVar; gnomAD			<a href="#">771632</a>
c.1602C>T	p.His534=	Exon 15	Likely Benign	Mar 16, 2022	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">2002609</a>
c.1604A>G	p.His535Arg	Exon 15	VUS	Oct 28, 2024	Inborn genetic diseases	<a href="#">1.64e-05</a>	ClinVar; gnomAD			<a href="#">3520384</a>
c.1605T>C	p.His535=	Exon 15	Likely Benign	Oct 22, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">6.84e-07</a>	ClinVar; gnomAD			<a href="#">1152734</a>
c.1609G>T	p.Glu537Ter	Exon 15	Likely Pathogenic	Jun 16, 2017	GM1 gangliosidosis type 3;Mucopolysaccharidosis; MPS-IV-B;Infantile GM1 gangliosidosis;GM1 gangliosidosis type 2		ClinVar			<a href="#">552455</a>
c.1612G>A	p.Ala538Thr	Exon 15	VUS	Mar 15, 2024	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis; Inborn genetic diseases	<a href="#">5.47e-06</a>	ClinVar; gnomAD			<a href="#">1919061</a>
c.1616G>A	p.Trp539Ter	Exon 15	Pathogenic	Mar 04, 2023	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar	<a href="#">18524657</a>		<a href="#">1382131</a>
c.1616G>C	p.Trp539Ser	Exon 15	VUS	Jan 30, 2025	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">3760697</a>
c.1617G>A	p.Trp539Ter	Exon 15	Pathogenic	Oct 15, 2020		<a href="#">6.58e-06</a>	ClinVar; gnomAD			<a href="#">1323014</a>
c.1620C>T	p.Ala540=	Exon 15	Likely Benign	Mar 17, 2025	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">1144060</a>
c.1633A>G	p.Asn545Asp	Exon 15	VUS	Feb 28, 2025	Inborn genetic diseases	<a href="#">3.42e-06</a>	ClinVar; gnomAD			<a href="#">3854130</a>
c.1634dup	p.Asn545fs	Exon 15	Pathogenic	Nov 05, 2025	GM1 gangliosidosis type 2;Mucopolysaccharidosis; MPS-IV-B;Infantile GM1 gangliosidosis;GM1 gangliosidosis type 3; GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B; Lysosomal storage disease	<a href="#">2.12e-05</a>	ClinVar; gnomAD			<a href="#">550413</a>
c.1635C>T	p.Asn545=	Exon 15	Likely Benign	Apr 09, 2025	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">2119868</a>
c.1638C>T	p.Tyr546=	Exon 15	Likely Benign	Dec 24, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">2.74e-06</a>	ClinVar; gnomAD			<a href="#">1669948</a>
c.1639A>C	p.Thr547Pro	Exon 15	VUS	May 25, 2025	Inborn genetic diseases	<a href="#">2.05e-06</a>	ClinVar; gnomAD			<a href="#">4029999</a>
c.1640C>T	p.Thr547Met	Exon 15	VUS	Oct 07, 2024	Inborn genetic diseases; GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">8.96e-05</a>	ClinVar; LOVD; gnomAD			<a href="#">1901214</a>
c.1641G>A	p.Thr547=	Exon 15	Likely Benign	Jan 28, 2026	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">0.00028</a>	ClinVar; gnomAD			<a href="#">704479</a>
c.1641G>T	p.Thr547=	Exon 15	Likely Benign	Jul 28, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">1.37e-06</a>	ClinVar; gnomAD			<a href="#">2149806</a>
c.1643T>A	p.Leu548His	Exon 15	VUS	Jul 18, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">2.74e-06</a>	ClinVar; gnomAD			<a href="#">1437570</a>
c.1644C>G		Exon 15	Not Classified			1.37e-06	LOVD; gnomAD			

cDNA Change	Protein Change	Location	Pathogenicity	Classified	Phenotype	gnomAD AF	Sources	PMID(s)	PIN	ClinVar ID
c.1644C>T	p.Leu548=	Exon 15	Likely Benign	Jan 19, 2026	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">4.79e-06</a>	ClinVar; gnomAD			<a href="#">729883</a>
c.1645C>T	p.Pro549Ser	Exon 15	Conflicting	Jan 02, 2026	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">6.84e-07</a>	ClinVar; gnomAD	<a href="#">17221873</a> ; <a href="#">17309651</a> ; <a href="#">22234367</a>		<a href="#">1517873</a>
c.1646C>T	p.Pro549Leu	Exon 15	Pathogenic	Sep 23, 2024	Mucopolysaccharidosis; MPS-IV-B; MPS-IV-B;GM1 gangliosidosis; Infantile GM1 gangliosidosis	<a href="#">7.52e-06</a>	ClinVar; LOVD; gnomAD	<a href="#">17221873</a> ; <a href="#">17309651</a> ; <a href="#">21520340</a> ; <a href="#">22128166</a> ; <a href="#">22234367</a>		<a href="#">426185</a>
c.1647G>A	p.Pro549=	Exon 15	Likely Benign	Feb 02, 2024	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">1.57e-05</a>	ClinVar; gnomAD			<a href="#">1412452</a>
c.1647G>C	p.Pro549=	Exon 15	Likely Benign	Mar 27, 2022	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">6.84e-07</a>	ClinVar; gnomAD			<a href="#">2087783</a>
c.1650C>G	p.Ala550=	Exon 15	Likely Benign	Aug 01, 2019	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">1115625</a>
c.1653T>C	p.Phe551=	Exon 15	Likely Benign	Apr 28, 2025	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">1.37e-06</a>	ClinVar; gnomAD			<a href="#">2419986</a>
c.1654dup	p.Tyr552fs	Exon 15	Pathogenic	Aug 09, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar; Santamaria 2006 (PMID:16941474)	<a href="#">16941474</a> ; <a href="#">17309651</a> ; <a href="#">17664528</a> ; <a href="#">23430803</a>		<a href="#">2023148</a>
c.1656T>C	p.Tyr552=	Exon 15	Likely Benign	Sep 02, 2025	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">2.74e-06</a>	ClinVar; gnomAD			<a href="#">1557632</a>
c.1657A>G	p.Met553Val	Exon 15	Likely Benign	Nov 26, 2025	Inborn genetic diseases	<a href="#">2.05e-06</a>	ClinVar; gnomAD			<a href="#">4620832</a>
c.1658T>C	p.Met553Thr	Exon 15	VUS	Jul 14, 2021	Infantile GM1 gangliosidosis; GM1 gangliosidosis type 2; GM1 gangliosidosis type 3; Mucopolysaccharidosis; MPS-IV-B	<a href="#">6.84e-07</a>	ClinVar; gnomAD			<a href="#">1199319</a>
c.1660G>A	p.Gly554Arg	Exon 15	Likely Pathogenic	Nov 28, 2023	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar	<a href="#">17309651</a>		<a href="#">2940375</a>
c.1662G>A	p.Gly554=	Exon 15	Likely Benign	Dec 02, 2024	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">2.05e-06</a>	ClinVar; gnomAD			<a href="#">2937331</a>
c.1662G>T	p.Gly554=	Exon 15	Likely Benign	Mar 18, 2022	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">2107547</a>
c.1665C>T	p.Asn555=	Exon 15	Likely Benign	Jun 19, 2021	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">6.84e-07</a>	ClinVar; gnomAD			<a href="#">1668745</a>
c.1667T>C	p.Phe556Ser	Exon 15	Likely Pathogenic	Sep 03, 2024	Mucopolysaccharidosis; MPS-IV-B; MPS-IV-B;GM1 gangliosidosis	<a href="#">6.84e-07</a>	ClinVar; LOVD; gnomAD; OMIM; NTSAD; Cure GM1 AllStripes PIN	<a href="#">33240792</a>	1	<a href="#">1963157</a>
c.1672A>G	p.Ile558Val	Exon 15	VUS	Jul 28, 2022	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">1990219</a>
c.1675C>T		Exon 15	Not Classified			2.05e-06	LOVD; gnomAD			
c.1679G>A		Exon 15	Not Classified			9.58e-06	LOVD; gnomAD			
c.1680T>C	p.Ser560=	Exon 15	Likely Benign	Oct 31, 2022	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">6.84e-07</a>	ClinVar; gnomAD			<a href="#">2108140</a>
c.1685dup	p.Asp564fs	Exon 15	Likely Pathogenic	Nov 15, 2019	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar; Santamaria 2006 (PMID:16941474)	<a href="#">16941474</a> ; <a href="#">17309651</a> ; <a href="#">23430803</a>		<a href="#">959418</a>
c.1689A>G	p.Pro563=	Exon 15	Likely Benign	Jul 28, 2023	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">2950407</a>
c.1692C>G	p.Asp564Glu	Exon 15	VUS	May 22, 2025			ClinVar			<a href="#">4530713</a>
c.1692C>T	p.Asp564=	Exon 15	Likely Benign	Jan 24, 2024	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">2937094</a>

cDNA Change	Protein Change	Location	Pathogenicity	Classified	Phenotype	gnomAD AF	Sources	PMID(s)	PIN	ClinVar ID
c.1696C>T	p.Pro566Ser	Exon 15	Likely Pathogenic	Feb 02, 2017			ClinVar			<a href="#">393276</a>
c.1697C>A	p.Pro566His	Exon 15	Pathogenic	Jan 11, 2020	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">645125</a>
c.1698C>T	p.Pro566=	Exon 15	Likely Benign	Feb 14, 2024	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">8.21e-06</a>	ClinVar; gnomAD			<a href="#">1674346</a>
c.1699C>T	p.Gln567Ter	Exon 15	P/LP	Oct 05, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">2.74e-06</a>	ClinVar; gnomAD	<a href="#">21497194</a> ; <a href="#">25557439</a> ; <a href="#">8213816</a>		<a href="#">1075413</a>
c.1701G>A	p.Gln567=	Exon 15	Likely Benign	Sep 12, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">1568820</a>
c.1703A>G	p.Asp568Gly	Exon 15	Pathogenic	Dec 04, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar	<a href="#">29451896</a>		<a href="#">2927178</a>
c.1704C>T	p.Asp568=	Exon 15	Likely Benign	Jun 17, 2021	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">1566754</a>
c.1707C>T	p.Thr569=	Exon 15	Likely Benign	Jul 07, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">2930127</a>
c.1711A>T	p.Ile571Phe	Exon 15	VUS	Jan 01, 2020			ClinVar			<a href="#">871698</a>
c.1714C>T	p.Gln572Ter	Exon 15	Pathogenic	Aug 20, 2022	Infantile GM1 gangliosidosis;GM1 gangliosidosis type 2;Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis type 3; GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">556285</a>
c.1715A>G	p.Gln572Arg	Exon 15	Likely Benign	Jan 26, 2026	GM1 gangliosidosis; Mucopolysaccharidosis; MPS-IV-B; MPS-IV-B;GM1 gangliosidosis	<a href="#">0.00030</a>	ClinVar; gnomAD			<a href="#">344784</a>
c.1716G>A	p.Gln572=	Exon 15	Likely Benign	Dec 30, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">6.84e-07</a>	ClinVar; gnomAD			<a href="#">2046743</a>
c.1721C>T	p.Pro574Leu	Exon 15	VUS	Apr 12, 2025	Inborn genetic diseases	<a href="#">2.74e-06</a>	ClinVar; gnomAD			<a href="#">4030000</a>
c.1724_1725delinsTG	p.Gly575Val	Exon 15	VUS	Jul 08, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">1940221</a>
c.1725A>G	p.Gly575=	Exon 15	Likely Benign	Sep 27, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">1.37e-06</a>	ClinVar; gnomAD			<a href="#">1962309</a>
c.1725A>T	p.Gly575=	Exon 15	Likely Benign	Sep 26, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">9.58e-06</a>	ClinVar; gnomAD			<a href="#">2428054</a>
c.1728G>A	p.Trp576Ter	Exon 15	Pathogenic	May 27, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar	<a href="#">17309651</a> ; <a href="#">21497194</a> ; <a href="#">25557439</a> ; <a href="#">8213816</a>		<a href="#">2925349</a>
c.1732A>C	p.Lys578Gln	Exon 15	VUS	Aug 09, 2016			ClinVar			<a href="#">495864</a>
c.1733A>G	p.Lys578Arg	Exon 15	P/LP	Nov 17, 2025	GM1 gangliosidosis type 2; GM1 gangliosidosis type 3;GM1 gangliosidosis type 2;Infantile GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis;Mucopolysaccharidosis; GM1 gangliosidosis; Inborn genetic diseases; Mucopolysaccharidosis	<a href="#">0.00012</a>	ClinVar; LOVD; gnomAD; OMIM; NTSAD; Cure GM1 AllStripes PIN	<a href="#">21497194</a> ; <a href="#">21520340</a> ; <a href="#">25557439</a> ; <a href="#">28476546</a> ; <a href="#">28554332</a> ; <a href="#">29352662</a> ; <a href="#">30267299</a> ; <a href="#">31367523</a> ; <a href="#">31761138</a> ; <a href="#">33240792</a> ; <a href="#">33737400</a> ; <a href="#">8213816</a>	2	<a href="#">252985</a>
c.1734G>T	p.Lys578Asn	Exon 15	Likely Pathogenic	May 08, 2025	Mucopolysaccharidosis; MPS-IV-B	<a href="#">6.84e-07</a>	ClinVar; gnomAD	<a href="#">31731261</a>		<a href="#">4818317</a>
c.1734+4C>T		Intron 15	VUS	Jul 17, 2022	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">6.16e-06</a>	ClinVar; gnomAD	<a href="#">17576681</a> ; <a href="#">9536098</a>		<a href="#">1908830</a>
c.1734+13A>G		Intron 15	Likely Benign	Aug 17, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">2933779</a>

cDNA Change	Protein Change	Location	Pathogenicity	Classified	Phenotype	gnomAD AF	Sources	PMID(s)	PIN	ClinVar ID
c.1734+13A>T		Intron 15	Likely Benign	Nov 20, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">3.42e-06</a>	ClinVar; gnomAD			<a href="#">2924250</a>
c.1734+14T>C		Intron 15	Likely Benign	Dec 02, 2024	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">6.84e-07</a>	ClinVar; gnomAD			<a href="#">2168640</a>
c.1734+169G>A		Intron 15	Likely Benign	Apr 20, 2019			ClinVar			<a href="#">1185796</a>
c.1734+188C>T		Intron 15	Benign	May 12, 2021			ClinVar			<a href="#">1291220</a>
c.1734+233C>T		Intron 15	Benign	Aug 28, 2018			ClinVar			<a href="#">1230485</a>
c.1735-16G>C		Intron 15	Likely Benign	Jun 27, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">6.85e-07</a>	ClinVar; gnomAD			<a href="#">1568898</a>
c.1735-16_1735-9dup		Intron 15	Likely Benign	Dec 23, 2021	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">6.85e-07</a>	ClinVar; gnomAD			<a href="#">1092402</a>
c.1735-15TC[4]		Intron 15	Likely Benign	Jul 08, 2019	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">1083468</a>
c.1735-15TC[5]		Intron 15	Likely Benign	Nov 11, 2023	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">1092032</a>
c.1735-15TC[7]		Intron 15	Likely Benign	Jan 26, 2026	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">344783</a>
c.1735-12C>G		Intron 15	Likely Benign	Oct 18, 2025	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">1.30e-05</a>	ClinVar; gnomAD			<a href="#">2156417</a>
c.1735-10C>G		Intron 15	Likely Benign	Jan 05, 2026	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">2.12e-05</a>	ClinVar; gnomAD			<a href="#">770075</a>
c.1735-10C>T		Intron 15	Likely Benign	Feb 24, 2025	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">4.25e-05</a>	ClinVar; gnomAD			<a href="#">1121003</a>
c.1735-8C>G		Intron 15	Likely Benign	Aug 28, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">4.11e-06</a>	ClinVar; gnomAD			<a href="#">1952156</a>
c.1735-8C>T		Intron 15	Likely Benign	Nov 02, 2020	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">1.37e-06</a>	ClinVar; gnomAD			<a href="#">1097217</a>
c.1735-5_1735-4dup		Intron 15	Not Classified			0.00037	LOVD; gnomAD			
c.1735-4C>G		Intron 15	Likely Benign	Aug 04, 2025	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">1611023</a>
c.1736G>A	p.Gly579Asp	Exon 16	Pathogenic	Apr 30, 2025	GM1 gangliosidosis	<a href="#">2.74e-06</a>	ClinVar; gnomAD; Caciotti 2005 (PMID:15714521)	<a href="#">10737981</a> ; <a href="#">15714521</a> ; <a href="#">21497194</a> ; <a href="#">35186388</a>		<a href="#">3896250</a>
c.1738C>G	p.Gln580Glu	Exon 16	VUS	Feb 29, 2024	GM1 gangliosidosis	<a href="#">6.85e-07</a>	ClinVar; gnomAD	<a href="#">23046582</a>		<a href="#">4759478</a>
c.1739A>G	p.Gln580Arg	Exon 16	Likely Pathogenic	Nov 14, 2021	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">6.85e-07</a>	ClinVar; gnomAD	<a href="#">23046582</a>		<a href="#">1475941</a>
c.1742T>G	p.Val581Gly	Exon 16	Likely Pathogenic	Dec 02, 2023	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">2951049</a>
c.1746G>A	p.Trp582Ter	Exon 16	Pathogenic	Sep 27, 2023	Infantile GM1 gangliosidosis;GM1 gangliosidosis type 2;Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis type 3; GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">1.37e-06</a>	ClinVar; LOVD; gnomAD; Santamaria 2006 (PMID:16941474)	<a href="#">10744681</a> ; <a href="#">16941474</a> ; <a href="#">17309651</a> ; <a href="#">17664528</a> ; <a href="#">20175788</a> ; <a href="#">21520340</a> ; <a href="#">23337983</a> ; <a href="#">23430803</a>		<a href="#">557709</a>
c.1746G>T		Exon 16	Not Classified			6.85e-07	LOVD; gnomAD			
c.1748T>G	p.Ile583Ser	Exon 16	VUS	Nov 11, 2024	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">3753904</a>
c.1752T>C	p.Asn584=	Exon 16	Likely Benign	Mar 20, 2024	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">1545895</a>

cDNA Change	Protein Change	Location	Pathogenicity	Classified	Phenotype	gnomAD AF	Sources	PMID(s)	PIN	ClinVar ID
c.1753G>A	p.Gly585Ser	Exon 16	VUS	Jul 26, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">1.71e-05</a>	ClinVar; gnomAD			<a href="#">1428950</a>
c.1761C>T	p.Asn587=	Exon 16	Likely Benign	Sep 27, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">2938128</a>
c.1764T>C	p.Leu588=	Exon 16	Likely Benign	May 29, 2021	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">1546324</a>
c.1767C>T	p.Gly589=	Exon 16	Likely Benign	Jul 29, 2025	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">3.42e-06</a>	ClinVar; gnomAD			<a href="#">707293</a>
c.1768C>A	p.Arg590Ser	Exon 16	Likely Pathogenic	Jun 10, 2022	GM1 gangliosidosis type 3;Mucopolysaccharidosis; MPS-IV-B;Infantile GM1 gangliosidosis;GM1 gangliosidosis type 2; Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">7.53e-06</a>	ClinVar; gnomAD; Santamaria 2006 (PMID:16941474)	<a href="#">16941474</a> ; <a href="#">17309651</a> ; <a href="#">23046582</a> ; <a href="#">23430803</a> ; <a href="#">26646981</a> ; <a href="#">8213816</a>		<a href="#">551476</a>
c.1768C>G	p.Arg590Gly	Exon 16	Conflicting	Dec 16, 2025	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar; Santamaria 2006 (PMID:16941474)	<a href="#">16941474</a> ; <a href="#">17309651</a> ; <a href="#">17664528</a> ; <a href="#">23430803</a>		<a href="#">2018030</a>
c.1768C>T	p.Arg590Cys	Exon 16	Pathogenic	Oct 18, 2025	Infantile GM1 gangliosidosis; Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis; MPS-IV-B; GLB1-related disorder; GM1 gangliosidosis	<a href="#">1.30e-05</a>	ClinVar; gnomAD; Santamaria 2006 (PMID:16941474)	<a href="#">16941474</a> ; <a href="#">17309651</a> ; <a href="#">17664528</a> ; <a href="#">23430803</a> ; <a href="#">25326635</a> ; <a href="#">37673932</a>		<a href="#">194596</a>
c.1769G>A	p.Arg590His	Exon 16	P/LP	Dec 16, 2025	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis; Infantile GM1 gangliosidosis; GM1 gangliosidosis type 2;Infantile GM1 gangliosidosis;GM1 gangliosidosis type 3; GM1 gangliosidosis; Progressive familial intrahepatic cholestasis; GM1 gangliosidosis type 2;Infantile GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis type 3; MPS-IV-B	<a href="#">2.67e-05</a>	ClinVar; LOVD; gnomAD; Santamaria 2006 (PMID:16941474)	<a href="#">16941474</a> ; <a href="#">17309651</a> ; <a href="#">21520340</a> ; <a href="#">23337983</a> ; <a href="#">23430803</a> ; <a href="#">26646981</a> ; <a href="#">27619815</a> ; <a href="#">33258288</a> ; <a href="#">33737400</a> ; <a href="#">38256219</a> ; <a href="#">8213816</a>		<a href="#">92901</a>
c.1771T>A	p.Tyr591Asn	Exon 16	Pathogenic	Jan 01, 2000	GM1-gangliosidosis; type I; with cardiac involvement		ClinVar	<a href="#">10737981</a>		<a href="#">947</a>
c.1771T>C	p.Tyr591His	Exon 16	Likely Pathogenic	Jul 06, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">6.84e-07</a>	ClinVar; gnomAD	<a href="#">10737981</a> ; <a href="#">29439846</a>		<a href="#">2151971</a>
c.1772A>C	p.Tyr591Ser	Exon 16	Likely Pathogenic	Mar 09, 2021	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar	<a href="#">10737981</a>		<a href="#">1472090</a>
c.1772A>G	p.Tyr591Cys	Exon 16	P/LP	Oct 13, 2023	GM1-gangliosidosis; type I; with cardiac involvement; GM1 gangliosidosis type 3;Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis type 2;Infantile GM1 gangliosidosis; Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis; GM1 gangliosidosis type 2	<a href="#">2.05e-06</a>	ClinVar; gnomAD; Caciotti 2005 (PMID:15714521)	<a href="#">10737981</a> ; <a href="#">15714521</a> ; <a href="#">21520340</a> ; <a href="#">23337983</a>		<a href="#">948</a>
c.1783C>T	p.Arg595Trp	Exon 16	VUS	Sep 09, 2024	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis type 2;GM1 gangliosidosis type 3;Infantile GM1 gangliosidosis; GM1 gangliosidosis type 2; MPS-IV-B;GM1 gangliosidosis; Inborn genetic diseases	<a href="#">6.02e-05</a>	ClinVar; gnomAD	<a href="#">17661814</a> ; <a href="#">30442161</a>		<a href="#">550288</a>

cDNA Change	Protein Change	Location	Pathogenicity	Classified	Phenotype	gnomAD AF	Sources	PMID(s)	PIN	ClinVar ID
c.1784G>A	p.Arg595Gln	Exon 16	VUS	Jun 10, 2024	GM1 gangliosidosis type 2; Infantile GM1 gangliosidosis; GM1 gangliosidosis type 3; Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis;Mucopolysaccharidosis; Inborn genetic diseases	<a href="#">2.60e-05</a>	ClinVar; gnomAD			<a href="#">1210419</a>
c.1784G>C	p.Arg595Pro	Exon 16	Likely Pathogenic	May 01, 2020		<a href="#">1.37e-06</a>	ClinVar; gnomAD			<a href="#">932727</a>
c.1785G>A	p.Arg595=	Exon 16	Likely Benign	Sep 15, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">6.84e-07</a>	ClinVar; gnomAD			<a href="#">1666810</a>
c.1787G>A	p.Gly596Asp	Exon 16	VUS	Jun 21, 2024	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis type 3;Infantile GM1 gangliosidosis;GM1 gangliosidosis type 2;Mucopolysaccharidosis		ClinVar			<a href="#">863780</a>
c.1792C>T	p.Gln598Ter	Exon 16	Likely Pathogenic	Jun 25, 2024	GM1 gangliosidosis type 3;Infantile GM1 gangliosidosis;GM1 gangliosidosis type 2;Mucopolysaccharidosis; MPS-IV-B; Mucopolysaccharidosis		ClinVar			<a href="#">3589131</a>
c.1794G>A	p.Gln598=	Exon 16	Likely Benign	Apr 05, 2021	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">6.84e-07</a>	ClinVar; gnomAD			<a href="#">1548025</a>
c.1801T>C	p.Leu601=	Exon 16	Likely Benign	Oct 28, 2023	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">2953343</a>
c.1803G>A	p.Leu601=	Exon 16	Likely Benign	Jul 22, 2025	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">1093710</a>
c.1810C>G	p.Pro604Ala	Exon 16	VUS	Nov 10, 2025	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">3750139</a>
c.1810C>T		Exon 16	Not Classified			1.37e-06	LOVD; gnomAD			
c.1812C>G	p.Pro604=	Exon 16	Likely Benign	Jul 02, 2020	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">1146315</a>
c.1812C>T	p.Pro604=	Exon 16	Likely Benign	Jun 20, 2023	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">4.10e-06</a>	ClinVar; gnomAD			<a href="#">2940077</a>
c.1813del	p.Gln605fs	Exon 16	Likely Pathogenic	Jun 08, 2025	Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">4818318</a>
c.1815G>A	p.Gln605=	Exon 16	Likely Benign	Jan 20, 2026	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">2.80e-05</a>	ClinVar; gnomAD			<a href="#">1099083</a>
c.1820T>C		Exon 16	Not Classified				LOVD			
c.1821C>A	p.Ile607=	Exon 16	Likely Benign	Aug 09, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">1553017</a>
c.1823T>C	p.Leu608Pro	Exon 16	Likely Pathogenic	Jun 13, 2024	Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis type 3;Mucopolysaccharidosis; MPS-IV-B;Infantile GM1 gangliosidosis;GM1 gangliosidosis type 2		ClinVar	<a href="#">21520340</a>		<a href="#">3339535</a>
c.1824G>C	p.Leu608=	Exon 16	Benign	Feb 04, 2026	Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis; MPS-IV-B;GM1 gangliosidosis; Infantile GM1 gangliosidosis	<a href="#">0.02000</a>	ClinVar; gnomAD			<a href="#">92902</a>
c.1825_1828del		Exon 16	Not Classified				LOVD			

cDNA Change	Protein Change	Location	Pathogenicity	Classified	Phenotype	gnomAD AF	Sources	PMID(s)	PIN	ClinVar ID
c.1830C>G	p.Thr610=	Exon 16	Likely Benign	Apr 29, 2025	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">1.09e-05</a>	ClinVar; gnomAD			<a href="#">724940</a>
c.1831T>G	p.Ser611Ala	Exon 16	VUS	Feb 27, 2025	Inborn genetic diseases	<a href="#">4.10e-06</a>	ClinVar; gnomAD			<a href="#">3854131</a>
c.1833G>A	p.Ser611=	Exon 16	Likely Benign	Jan 05, 2026	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B; GLB1-related disorder	<a href="#">8.48e-05</a>	ClinVar; gnomAD			<a href="#">705887</a>
c.1833G>C	p.Ser611=	Exon 16	Likely Benign	Jul 17, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">1.37e-06</a>	ClinVar; gnomAD			<a href="#">1083702</a>
c.1837_1838del	p.Pro613fs	Exon 16	Likely Pathogenic	Sep 25, 2025	GM1 gangliosidosis; Mucopolysaccharidosis; MPS-IV-B	<a href="#">6.84e-07</a>	ClinVar; gnomAD	<a href="#">17221873</a>		<a href="#">917629</a>
c.1842C>T	p.Asn614=	Exon 16	Likely Benign	Nov 29, 2022	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">2941912</a>
c.1844C>A	p.Thr615Asn	Exon 16	Likely Benign	Nov 28, 2023	Inborn genetic diseases	<a href="#">8.89e-05</a>	ClinVar; gnomAD			<a href="#">3100051</a>
c.1850C>G		Exon 16	Not Classified				LOVD			
c.1851C>T	p.Thr617=	Exon 16	Likely Benign	Nov 16, 2025	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">1.85e-05</a>	ClinVar; gnomAD			<a href="#">1155734</a>
c.1852G>A	p.Val618Met	Exon 16	VUS	Oct 31, 2022	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">8.89e-06</a>	ClinVar; gnomAD			<a href="#">1038644</a>
c.1854G>A	p.Val618=	Exon 16	Likely Benign	Aug 19, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">1.37e-06</a>	ClinVar; gnomAD			<a href="#">2019856</a>
c.1854G>C	p.Val618=	Exon 16	Likely Benign	Oct 03, 2025	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">2022065</a>
c.1855C>A	p.Leu619Met	Exon 16	VUS	Nov 27, 2023	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">2097996</a>
c.1855C>T	p.Leu619=	Exon 16	Likely Benign	Jan 06, 2026	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">2940463</a>
c.1861C>T	p.Leu621=	Exon 16	Likely Benign	Dec 30, 2025	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">1.98e-05</a>	ClinVar; gnomAD			<a href="#">767268</a>
c.1870G>A	p.Ala624Thr	Exon 16	VUS	Apr 29, 2024			ClinVar			<a href="#">3251341</a>
c.1873C>T	p.Pro625Ser	Exon 16	VUS	Aug 04, 2023	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">2.74e-06</a>	ClinVar; gnomAD			<a href="#">1414271</a>
c.1879A>C	p.Ser627Arg	Exon 16	VUS	May 08, 2023	Inborn genetic diseases	<a href="#">3.63e-05</a>	ClinVar; gnomAD			<a href="#">2509861</a>
c.1883G>A	p.Ser628Asn	Exon 16	VUS	May 23, 2024	Inborn genetic diseases	<a href="#">4.10e-06</a>	ClinVar; gnomAD			<a href="#">3281502</a>
c.1896A>G	p.Glu632=	Exon 16	Likely Benign	Jul 25, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">4.10e-06</a>	ClinVar; gnomAD			<a href="#">1122370</a>
c.1897C>G	p.Leu633Val	Exon 16	VUS	Dec 20, 2021	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">5.47e-06</a>	ClinVar; gnomAD			<a href="#">2063069</a>
c.1899A>G	p.Leu633=	Exon 16	Likely Benign	Jan 17, 2026	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">6.16e-06</a>	ClinVar; gnomAD			<a href="#">2153167</a>
c.1899A>T	p.Leu633=	Exon 16	Likely Benign	Jan 05, 2024	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">6.84e-07</a>	ClinVar; gnomAD			<a href="#">2921979</a>
c.1901G>A	p.Cys634Tyr	Exon 16	VUS	Sep 04, 2024		<a href="#">1.23e-05</a>	ClinVar; LOVD; gnomAD	<a href="#">33737400</a> ; <a href="#">37381921</a>		<a href="#">3375047</a>
c.1903G>C	p.Ala635Pro	Exon 16	VUS	Dec 29, 2017	Ependymoma		ClinVar			<a href="#">487803</a>
c.1903G>T	p.Ala635Ser	Exon 16	VUS	Jun 04, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">1974200</a>
c.1905T>C	p.Ala635=	Exon 16	Likely Benign	Dec 25, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">2.05e-06</a>	ClinVar; gnomAD			<a href="#">2185969</a>

cDNA Change	Protein Change	Location	Pathogenicity	Classified	Phenotype	gnomAD AF	Sources	PMID(s)	PIN	ClinVar ID
c.1910C>T	p.Thr637Met	Exon 16	VUS	Jun 03, 2025	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B; Inborn genetic diseases	<a href="#">1.50e-05</a>	ClinVar; gnomAD			<a href="#">1512979</a>
c.1911G>A	p.Thr637=	Exon 16	Likely Benign	Aug 21, 2025	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">9.58e-06</a>	ClinVar; gnomAD			<a href="#">1102658</a>
c.1911G>T	p.Thr637=	Exon 16	Likely Benign	Oct 23, 2023	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">2952169</a>
c.1914C>T	p.Phe638=	Exon 16	Likely Benign	Jan 20, 2026	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">1.44e-05</a>	ClinVar; gnomAD			<a href="#">1116502</a>
c.1915G>A	p.Val639Met	Exon 16	VUS	May 16, 2025	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis; Inborn genetic diseases	<a href="#">8.89e-06</a>	ClinVar; gnomAD			<a href="#">1309040</a>
c.1917G>C	p.Val639=	Exon 16	Likely Benign	Feb 05, 2024	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">2.05e-06</a>	ClinVar; gnomAD			<a href="#">2145474</a>
c.1918G>T	p.Asp640Tyr	Exon 16	VUS	Sep 10, 2013		<a href="#">1.37e-06</a>	ClinVar; gnomAD			<a href="#">92903</a>
c.1931T>C	p.Ile644Thr	Exon 16	VUS	Feb 07, 2023	Inborn genetic diseases	<a href="#">6.16e-06</a>	ClinVar; gnomAD			<a href="#">2481567</a>
c.1935C>A	p.Gly645=	Exon 16	Likely Benign	Sep 15, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">2.05e-06</a>	ClinVar; gnomAD			<a href="#">1549913</a>
c.1938A>G	p.Ser646=	Exon 16	Likely Benign	Feb 04, 2026	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">7.52e-05</a>	ClinVar; gnomAD			<a href="#">706556</a>
c.1947C>T	p.Thr649=	Exon 16	Likely Benign	Apr 25, 2024	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">1.97e-05</a>	ClinVar; gnomAD			<a href="#">2051810</a>
c.1950C>T	p.Tyr650=	Exon 16	Likely Benign	Aug 20, 2025	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">2.12e-05</a>	ClinVar; gnomAD			<a href="#">1157465</a>
c.1951G>A	p.Asp651Asn	Exon 16	VUS	Mar 20, 2024	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">5.47e-06</a>	ClinVar; gnomAD			<a href="#">1973950</a>
c.1959C>T	p.Pro653=	Exon 16	Likely Benign	Feb 06, 2024	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">1.37e-06</a>	ClinVar; gnomAD			<a href="#">1566048</a>
c.1960T>G	p.Ser654Ala	Exon 16	VUS	Jan 09, 2025	Inborn genetic diseases		ClinVar			<a href="#">3854132</a>
c.1969G>A	p.Val657Ile	Exon 16	VUS	May 28, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">1.09e-05</a>	ClinVar; gnomAD			<a href="#">2161080</a>
c.1978A>T	p.Arg660Ter	Exon 16	VUS	Dec 28, 2017	Infantile GM1 gangliosidosis;GM1 gangliosidosis type 2;Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis type 3		ClinVar			<a href="#">555675</a>
c.1979G>A	p.Arg660Lys	Exon 16	VUS	Jul 29, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">6.84e-07</a>	ClinVar; gnomAD			<a href="#">1957630</a>
c.1980A>G	p.Arg660=	Exon 16	Likely Benign	Apr 09, 2019	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">1091463</a>
c.1981C>T	p.Leu661Phe	Exon 16	VUS	Oct 07, 2022	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">1.23e-05</a>	ClinVar; gnomAD			<a href="#">1982914</a>
c.1983C>T	p.Leu661=	Exon 16	Likely Benign	May 17, 2024	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">6.16e-06</a>	ClinVar; gnomAD			<a href="#">1096185</a>
c.1989C>T	p.Pro663=	Exon 16	Likely Benign	Oct 07, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar			<a href="#">1136136</a>
c.1990C>T	p.Pro664Ser	Exon 16	VUS	Mar 11, 2022	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">5.47e-06</a>	ClinVar; gnomAD			<a href="#">2069213</a>

cDNA Change	Protein Change	Location	Pathogenicity	Classified	Phenotype	gnomAD AF	Sources	PMID(s)	PIN	ClinVar ID
c.1991del	p.Pro664fs	Exon 16	Likely Pathogenic	Mar 21, 2024	GM1 gangliosidosis type 3;Mucopolysaccharidosis; MPS-IV-B;Infantile GM1 gangliosidosis;GM1 gangliosidosis type 2		ClinVar	<a href="#">10744681</a>		<a href="#">551038</a>
c.1992A>C	p.Pro664=	Exon 16	Likely Benign	Feb 11, 2022	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">2096335</a>
c.1992A>G	p.Pro664=	Exon 16	Likely Benign	Oct 15, 2025	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">4.10e-06</a>	ClinVar; gnomAD			<a href="#">759076</a>
c.1995C>G	p.Pro665=	Exon 16	Likely Benign	Sep 14, 2021	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">1604484</a>
c.1995C>T	p.Pro665=	Exon 16	Likely Benign	Jan 19, 2026	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">1.09e-05</a>	ClinVar; gnomAD			<a href="#">1162004</a>
c.1997C>G		Exon 16	Not Classified			6.84e-07	LOVD; gnomAD			
c.1997C>T	p.Pro666Leu	Exon 16	VUS	Sep 07, 2022	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">2.19e-05</a>	ClinVar; gnomAD			<a href="#">1347335</a>
c.1998G>A	p.Pro666=	Exon 16	Likely Benign	Jan 31, 2026	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">1.23e-05</a>	ClinVar; gnomAD			<a href="#">708999</a>
c.2002A>T	p.Lys668Ter	Exon 16	VUS	Jan 27, 2025	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis type 2;GM1 gangliosidosis type 3;Infantile GM1 gangliosidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">1.03e-05</a>	ClinVar; gnomAD	<a href="#">33558080</a>		<a href="#">550693</a>
c.2006del	p.Asn669fs	Exon 16	VUS	Jun 19, 2021	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">1.37e-06</a>	ClinVar; gnomAD			<a href="#">1473458</a>
c.2006dup	p.Asn669fs	Exon 16	P/LP	Oct 31, 2024	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis; GM1 gangliosidosis type 2; GLB1-related disorder	<a href="#">2.74e-06</a>	ClinVar; gnomAD	<a href="#">30703229</a> ; <a href="#">31367523</a> ; <a href="#">32005694</a>		<a href="#">1067823</a>
c.2007C>T	p.Asn669=	Exon 16	Likely Benign	Feb 09, 2024	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">7.52e-06</a>	ClinVar; gnomAD			<a href="#">1127239</a>
c.2007_2010del	p.Asn669fs	Exon 16	VUS	Mar 20, 2017	GM1 gangliosidosis type 3;Mucopolysaccharidosis; MPS-IV-B;Infantile GM1 gangliosidosis;GM1 gangliosidosis type 2		ClinVar			<a href="#">551178</a>
c.2008A>G	p.Lys670Glu	Exon 16	VUS	Dec 02, 2021	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">4.10e-06</a>	ClinVar; gnomAD			<a href="#">1365634</a>
c.2009A>G	p.Lys670Arg	Exon 16	VUS	Feb 05, 2022	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B	<a href="#">1.37e-06</a>	ClinVar; gnomAD			<a href="#">1354608</a>
c.2009_2010del	p.Lys670fs	Exon 16	Conflicting	Jul 25, 2025	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis		ClinVar	<a href="#">33558080</a>		<a href="#">1961404</a>
c.2010dup	p.Asp671fs	Exon 16	VUS	Jun 24, 2022	Infantile GM1 gangliosidosis	<a href="#">1.37e-06</a>	ClinVar; gnomAD			<a href="#">1699503</a>
c.2011del	p.Asp671fs	Exon 16	VUS	Mar 12, 2018	Infantile GM1 gangliosidosis;GM1 gangliosidosis type 2;Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis type 3		ClinVar			<a href="#">557185</a>

cDNA Change	Protein Change	Location	Pathogenicity	Classified	Phenotype	gnomAD AF	Sources	PMID(s)	PIN	ClinVar ID
c.2012A>G	p.Asp671Gly	Exon 16	Conflicting	Sep 20, 2024	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis; MPS-IV-B;GM1 gangliosidosis type 3;GM1 gangliosidosis type 2;Infantile GM1 gangliosidosis	<a href="#">1.16e-05</a>	ClinVar; gnomAD			<a href="#">2188026</a>
c.2016A>G	p.Ser672=	Exon 16	Conflicting	Nov 27, 2025	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis; Mucopolysaccharidosis; Infantile GM1 gangliosidosis; GM1 gangliosidosis type 3; GM1 gangliosidosis type 2	<a href="#">5.13e-05</a>	ClinVar; gnomAD			<a href="#">733159</a>
c.2016A>T	p.Ser672=	Exon 16	Likely Benign	Nov 10, 2022	GM1 gangliosidosis;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">2941454</a>
c.2022G>A	p.Leu674=	Exon 16	Likely Benign	Jan 03, 2024	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">6.84e-07</a>	ClinVar; gnomAD			<a href="#">2923580</a>
c.2028T>C	p.His676=	Exon 16	Likely Benign	Jan 05, 2023	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">4.79e-06</a>	ClinVar; gnomAD			<a href="#">1135872</a>
c.2030T>G	p.Val677Gly	Exon 16	VUS	Oct 26, 2024	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis; MPS-IV-B	<a href="#">8.89e-06</a>	ClinVar; gnomAD	<a href="#">33558080</a>		<a href="#">968996</a>
c.2031A>G	p.Val677=	Exon 16	Likely Benign	May 21, 2021	Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis	<a href="#">6.57e-06</a>	ClinVar; gnomAD			<a href="#">1077701</a>
c.2032T>C	p.Ter678Arg	Exon 16	VUS	Jun 06, 2018	Infantile GM1 gangliosidosis;GM1 gangliosidosis type 2;GM1 gangliosidosis type 3;Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">558705</a>
c.2034A>C	p.Ter678Cys	Exon 16	VUS	Mar 28, 2018	Infantile GM1 gangliosidosis;GM1 gangliosidosis type 2;Mucopolysaccharidosis; MPS-IV-B;GM1 gangliosidosis type 3		ClinVar			<a href="#">557535</a>
c.*39C>T		3'UTR	VUS	Jan 12, 2018	Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis		ClinVar			<a href="#">903011</a>
c.*44C>T		3'UTR	VUS	Jan 13, 2018	Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis	<a href="#">6.23e-05</a>	ClinVar; gnomAD			<a href="#">902132</a>
c.*182A>G		3'UTR	Benign	Jan 13, 2018	GM1 gangliosidosis; Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">344782</a>
c.*184G>A		3'UTR	Likely Benign	Jan 13, 2018	Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis		ClinVar			<a href="#">344781</a>
c.*194C>A		3'UTR	VUS	Jan 13, 2018	Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis		ClinVar			<a href="#">344780</a>
c.*224G>T		3'UTR	VUS	Jan 13, 2018	GM1 gangliosidosis; Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">900465</a>
c.*319G>A		3'UTR	Conflicting	Mar 01, 2023	Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis		ClinVar			<a href="#">344779</a>
c.*354T>C		3'UTR	Benign	Jan 12, 2018	GM1 gangliosidosis; Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">344778</a>
c.*376C>G		3'UTR	Likely Benign	Jan 13, 2018	Mucopolysaccharidosis; MPS-IV-B; GM1 gangliosidosis		ClinVar			<a href="#">344777</a>
c.*423A>G		3'UTR	VUS	Jan 13, 2018	GM1 gangliosidosis; Mucopolysaccharidosis; MPS-IV-B		ClinVar			<a href="#">344776</a>

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## Appendix

### ClinVar

NCBI ClinVar was queried via the eSummary API for all *GLB1* variation IDs. Data extracted: variant name (HGVS cDNA and protein), variant type, classification, review status, associated phenotype(s), and linked PubMed IDs (via NCBI eLink API). All variants were extracted regardless of classification.

### Cure GM1 Patient Insights Network (PIN)

The Cure GM1 Patient Insights Network (PIN) captures real-world genotypic data from GM1 gangliosidosis patients. PIN data informed the identification of 24 variants observed in the GM1 patient community. Results were featured at the Cure GM1 World Symposium (January 2025).

### Cure GM1 World Symposium Poster

*Genotypic Heterogeneity in GM1 Gangliosidosis* — presented at the Cure GM1 World Symposium, January 2025. This poster summarizes genotypic diversity across PIN participants and literature-derived cases, and was a primary source for variant and phenotype cross-referencing in this catalog. Available at [curegm1.org/world2025](https://curegm1.org/world2025)

### LOVD

The LOVD *GLB1* locus-specific database was queried via the LOVD Atom/REST API ([databases.lovd.nl](https://databases.lovd.nl)).

### NTSAD Variant Database

The National Tay-Sachs & Allied Diseases Association Variant Database (May 2024 PDF edition) was ingested and the *GLB1* section extracted.

### OMIM

Online Mendelian Inheritance in Man entry 611458 (*GLB1* gene) and four phenotype entries (#230500, #230600, #230650, #253010) were reviewed for named allelic variants. Variants were extracted with their associated phenotypes and cited PMIDs.

### gnomAD

gnomAD v4 was queried via the GraphQL API for all *GLB1* variants. Variant records were retrieved with allele counts (AC) and allele frequencies (AF) from the exome and genome datasets. Frequencies were matched to catalog variants by cDNA nomenclature.

### Literature

Six landmark publications were reviewed: Caciotti et al. 2011 (PMID:21497194); Bidchol et al. 2015 (PMID:25936995); Santamaria et al. 2006 (PMID:16941474); Morrone et al. 2000 (PMID:10737981); Hofer et al. 2009 (PMID:19472408) and Hofer et al. 2010 (PMID:20175788).

### Nomenclature

All cDNA nomenclature follows NM\_000404.4. Exon/intron location was assigned based on the published *GLB1* gene structure with approximate exon boundaries.