

Variant: NM_001363576.1:c.60G>A

Version: 1.0

CA355961522 [↗](#)

Gene: IDUA ([HGNC:3425](#))

Condition: mucopolysaccharidosis type 1 ([MONDO:0001586](#))

Inheritance Mode: Autosomal recessive inheritance

UUID: cb234f5a-6fdd-43cb-aba6-2a8746cda859

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Published on: 2026/5/15

HGVS expressions

NM_001363576.1:c.60G>A

NC_000004.12:g.1000952G>A

CM000666.2:g.1000952G>A

NC_000004.11:g.994740G>A

CM000666.1:g.994740G>A

NC_000004.10:g.984740G>A

NG_008103.1:g.18956G>A

ENST00000247933.9:c.456G>A

ENST00000514224.2:c.456G>A

ENST00000652070.1:n.512G>A

ENST00000247933.8:c.456G>A

ENST00000502910.5:c.315G>A

ENST00000504568.5:c.416G>A

ENST00000506561.5:n.465G>A

ENST00000508168.5:n.334G>A

ENST00000509948.5:c.249G>A

ENST00000514192.5:c.273G>A

ENST00000514224.1:c.60G>A

ENST00000514698.5:n.356G>A

NM_000203.4:c.456G>A

NR_110313.1:n.544G>A

NM_000203.5:c.456G>A

Likely Pathogenic

Met criteria codes **2**

PM2_Supporting

PVS1_Strong

Evidence Links **0**

Expert Panel

Lysosomal Diseases VCEP [↗](#)

Criteria Specification Information

[↗](#) **Criteria Specification:** ClinGen Lysosomal Diseases
Expert Panel Specifications to the ACMG/AMP Variant
Interpretation Guidelines for IDUA Version 1.2.0

[↗](#) **Criteria Specification Approval History**

[↗](#) **Criteria Specifications for this VCEP**

Lysosomal Diseases VCEP

The NM_000203.5:c.456G>A p.(Trp152Ter) variant in IDUA is a nonsense variant predicted to cause a premature stop codon in biologically-relevant-exon 4 out of 14, leading to nonsense mediated decay in a gene in which loss-of-function is an established disease mechanism (PVS1). This variant has been detected in at least one individual with MPS I. This individual is compound heterozygous for this variant and c.1513C>G p.(Arg505Gly), which has been classified as pathogenic or likely pathogenic by the ClinGen Lysosomal Diseases VCEP (PMID 31194252; phase unconfirmed; 0.25 points) (PM3 unmet). This variant is absent in gnomAD v4.1.0. (PM2_Supporting). The classification of this variant has been upgraded from Variant of Uncertain Significance to Likely Pathogenic based on the recommendations of the ClinGen Sequence Variant Interpretation Working Group, that a variant meeting PVS1 and PM2_Supporting is classified as Likely Pathogenic (https://clinicalgenome.org/site/assets/files/5182/pm2_-_svi_recommendation_-_approved_sept2020.pdf). In summary, this variant meets the criteria to be classified as likely pathogenic for MPS I based on the IDUA-specific ACMG/AMP criteria applied, as specified by the ClinGen Lysosomal Diseases Variant Curation Expert Panel (Specifications Version 1.2.0): PVS1, PM2_supporting (Classification approved by the ClinGen Lysosomal Diseases Variant Curation Expert Panel on May 4, 2026)

Met criteria codes

PM2_Supporting			This variant is absent in gnomAD v4.0. (PM2_Supporting).
PVS1_Strong			The NM_000203.5:c.456G>A p.(Trp152Ter) variant in IDUA is a nonsense variant predicted to cause a premature stop codon in biologically-relevant-exon 4 out of 14, leading to nonsense mediated decay in a gene in which loss-of-function is an established disease mechanism (PVS1).

Curation History

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