The ClinGen Evidence Repository is an **FDA-recognized human genetic variant database** containing expert-curated assertions regarding variants' pathogenicity and supporting evidence summaries. [Disclaimer]

Variant: NM_000203.5(IDUA):c.24C>A (p.Ala8_Ala9=)

Version: 1.3

CA145878

92638 (ClinVar)

Gene: IDUA (HGNC:3425)
Condition: mucopolysaccharidosis type 1 (MONDO:0001586)
Inheritance Mode: Autosomal recessive inheritance
UUID: e23cadf4-f755-4a73-aaae-370c5d6e2464
Approved on: 2025/1/2
Published on: 2025/1/2

HGVS expressions

NM_000203.5:c.24C>A
NM_000203.5(IDUA):c.24C>A (p.Ala8_Ala9=)
NC_000004.12:g.987108C>A
CM000666.2:g.987108C>A
NC_000004.11:g.980896C>A
CM000666.1:g.980896C>A
NC_000004.10:g.970896C>A
NG_008103.1:g.5112C>A
NG_033042.1:g.11329G>T
ENST00000247933.9:c.24C>A
ENST00000514224.2:c.24C>A
ENST00000247933.8:c.24C>A
ENST00000398520.6:c.576+4020G>T
ENST00000502910.5:c.24C>A
ENST00000504568.5:c.22C>A
ENST00000506561.5:n.33C>A
ENST00000508168.5:n.43C>A
ENST00000514698.5:n.65C>A
ENST00000622731.4:c.576+4020G>T
NM_000203.4:c.24C>A
NM_134425.2:c.576+4020G>T
NR_110313.1:n.112C>A
NM_134425.3:c.576+4020G>T
NM_134425.4:c.576+4020G>T

Benign

Met criteria codes 1

BA1

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 1.0.0*
Criteria Specification Approval History
Criteria Specifications for this VCEP

Evidence submitted by expert panel

Lysosomal Diseases VCEP

The NM_000203.5:c.24C>A variant in IDUA is a synonymous (silent) variant (p.Ala8=). The Grpmax Filtering AF (95% confidence) in gnomAD v4.1.0 is 0.5236 in the Admixed American population. This is higher than the ClinGen Lysosomal Diseases VCEP's threshold for BA1 (>0.005), and therefore meets this criterion (BA1). There is a ClinVar entry for this variant (Variation ID: 92638). In summary, this variant meets the criteria to be classified as benign for mucopolysaccharidosis type 1. IDUA-specific ACMG/AMP criteria applied, as specified by the ClinGen Lysosomal Diseases Variant Curation Expert Panel (Specifications Version 1.0.0): BA1. (Classification approved by the ClinGen Lysosomal Diseases Variant Curation Expert Panel on January 2, 2025)

Met criteria codes

BA1	✔	The Grpmax Filtering AF (95% confidence) in gnomAD v4.1.0 is 0.5236 in the Admixed American population. This is higher than the ClinGen Lysosomal Diseases VCEP's threshold for BA1 (>0.005), and therefore meets this criterion (BA1). Note: gnomAD v4.1.0. states "discrepant frequencies" between genomes and exomes. The Grpmax Filtering AF is 0.5498 in exomes, 0.4749 in genomes, and 0.5236 overall, all in the Admixed American population and all meeting the BA1 threshold.

Curation History

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