

Variant: *NM_000419.4(ITGA2B):c.2127G>A (p.Lys709=)*

Version: 1.0

CA8602814 [↗](#)

323547 (ClinVar) [↗](#)

Gene: ITGA2B ([HGNC:3674](#))

Condition: Glanzmann's thrombasthenia ([MONDO:0010119](#))

Inheritance Mode: Autosomal recessive inheritance

UUID: 974b3842-4997-48f5-8619-5a26d4705f51

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HGVS expressions

NM_000419.4:c.2127G>A

NM_000419.4(ITGA2B):c.2127G>A (p.Lys709=)

NC_000017.11:g.44377758C>T

CM000679.2:g.44377758C>T

NC_000017.10:g.42455126C>T

CM000679.1:g.42455126C>T

NC_000017.9:g.39810652C>T

NG_008331.1:g.16748G>A

ENST00000262407.6:c.2127G>A

ENST00000648408.1:c.1558G>A

ENST00000262407.5:c.2127G>A

ENST00000592462.5:n.922G>A

NM_000419.3:c.2127G>A

NM_000419.5:c.2127G>A

Benign

Met criteria codes **1**

BA1

Not Met criteria codes **2**

BP7

PP4

Evidence Links **0**

Expert Panel

[Platelet Disorders VCEP](#) [↗](#)

Criteria Specification Information **!**

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

Evidence submitted by expert panel

Platelet Disorders VCEP

The Lys709= synonymous variant occurs at an allele frequency in gnomAD of 0.001729 with a MAF of 0.01699 (424/24,960 alleles) in the African population. This is above the >0.0024 threshold. This variant meets criteria to be classified as Benign by the ClinGen Platelet Disorders VCEP. GT-specific criteria met: BA1.

Met criteria codes

BA1			The overall allele frequency in gnomAD is 0.001729 with a MAF of 0.01699 (424/24,960 alleles) in the African population. This is above the >0.0024 threshold.
Not Met criteria codes			
BP7			HSF identifies activation of an exonic cryptic acceptor site, with potential alteration of splicing. However this is not identified by other splice site predictors.
PP4			This variant has been identified by an inherited thrombocytopenia panel in the heterozygous state in one patient who does not have a phenotype highly specific to Glanzmann thrombasthenia. The patient has thrombocytopenia with no history of bleeding.

Curation History 	
	
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