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Variant: NM_002185.5(IL7R):c.235G>T (p.Glu79Ter)

Version: 1.0

CA359427654

1323115 (ClinVar)

Gene: IL7R (HGNC:3575)
Condition: severe combined immunodeficiency, autosomal recessive, T cell-negative, B cell-positive, NK cell-positive (MONDO:0012163)
Inheritance Mode: Autosomal recessive inheritance
UUID: 1c8f4bc6-5f05-48ab-9c82-c81899baba0a
Approved on: 2024/1/30
Published on: 2024/1/30

HGVS expressions

NM_002185.5(IL7R):c.235G>T (p.Glu79Ter)
NM_002185.5:c.235G>T
NC_000005.10:g.35867319G>T
CM000667.2:g.35867319G>T
NC_000005.9:g.35867421G>T
CM000667.1:g.35867421G>T
NC_000005.8:g.35903178G>T
NG_009567.1:g.15431G>T
ENST00000303115.8:c.235G>T
ENST00000303115.7:c.235G>T
ENST00000506850.5:c.235G>T
ENST00000511031.1:n.369G>T
ENST00000511982.1:c.235G>T
ENST00000514217.5:c.235G>T
NM_002185.3:c.235G>T
NR_120485.1:n.338G>T
NM_002185.4:c.235G>T
NR_120485.2:n.364G>T
NR_120485.3:n.322G>T

Likely Pathogenic

Met criteria codes 2

PVS1 PM2_Supporting

Evidence Links 0

Expert Panel

Severe Combined Immunodeficiency Disease VCEP

Criteria Specification Information

Criteria Specification: *ClinGen Severe Combined Immunodeficiency Disease Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for IL7R Version 1.0.0*
Criteria Specification Approval History
Criteria Specifications for this VCEP

Evidence submitted by expert panel

Severe Combined Immunodeficiency Disease VCEP

The c.235G>T (p.Glu79Ter) (NM_002185.5) variant in IL7R is a nonsense variant predicted to cause a premature stop codon in biologically-relevant-exon 3/8 leading to nonsense-mediated decay in a gene in which loss-of-function is an established disease mechanism (PVS1 Met). The variant is absent in gnomAD v4 (PM2_supporting). There are no publications for this variant in the literature. As per the SCID VCEP specifications and the Bayesian interpretation of the ACMG/AMP combining rules,1 very strong and 1 supporting criteria results in a Likely Pathogenic classification. In summary, this variant meets the criteria to be classified as Likely Pathogenic variant for autosomal recessive severe combined immunodeficiency due to IL7R deficiency based on the ACMG/AMP criteria applied, as specified by the ClinGen SCID VCEP: PVS1_Met,PM2_supporting (VCEP specifications version 1).

Met criteria codes

PVS1	✔	The c.235G>T (p.Glu79Ter) (NM_002185.5) variant in IL7R is a nonsense variant predicted to cause a premature stop codon in biologically-relevant-exon 3/8 leading to nonsense-mediated decay in a gene in which loss-of-function is an established disease mechanism (PVS1 Met).
PM2_Supporting	✔	The variant is absent in gnomAD v4 (PM2_supporting).

Curation History

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