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_001754.5(RUNX1):c.503del (p.Gly168fs)

Version: 1.0

CA2573157355

1363605 (ClinVar)

Gene: RUNX1 (HGNC:861)
Condition: hereditary thrombocytopenia and hematologic cancer predisposition syndrome (MONDO:0011071)
Inheritance Mode: Autosomal dominant inheritance
UUID: 7bdeb898-0ff9-4075-aed1-5a45e2a2fb71
Approved on: 2024/3/26
Published on: 2024/3/26

HGVS expressions

NM_001754.5(RUNX1):c.503del (p.Gly168fs)
NM_001754.5:c.503del
NC_000021.9:g.34880563del
CM000683.2:g.34880563del
NC_000021.8:g.36252860del
CM000683.1:g.36252860del
NC_000021.7:g.35174730del
NG_011402.2:g.1109150del
ENST00000675419.1:c.503del
ENST00000300305.7:c.503del
ENST00000344691.8:c.422del
ENST00000358356.9:c.422del
ENST00000399237.6:c.467del
ENST00000399240.5:c.422del
ENST00000437180.5:c.503del
ENST00000482318.5:c.*93del
NM_001001890.2:c.422del
NM_001122607.1:c.422del
NM_001754.4:c.503del
NM_001001890.3:c.422del
NM_001122607.2:c.422del

Pathogenic

Met criteria codes 3

PVS1 PM2_Supporting PM5_Supporting

Not Met criteria codes 23

BS2 BS1 BS4 BS3 BP4 BP3 BP1 BP2 BP5 BP7 PS1 PS2 PS3 PS4 PM1 PM3 PM4 BA1 PM6 PP1 PP2 PP3 PP4

Evidence Links 0

Expert Panel

Myeloid Malignancy VCEP

Criteria Specification Information

- **Criteria Specification:** *ClinGen Myeloid Malignancy Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines Version 2*
  - **PDF**
- **Criteria Specification Approval History**
- **Criteria Specifications for this VCEP**

Evidence submitted by expert panel

Myeloid Malignancy VCEP

In summary, this variant meets criteria to be classified as pathogenic. ACMG/AMP criteria applied, as specified by the Myeloid Malignancy Variant Curation Expert Panel for RUNX1: PVS1, PM5_supp, PM2_supp

Met criteria codes

PVS1	✔	This is a null variant predicted to undergo NMD, present in all biologically relevant transcripts (frameshift variant located between c.98-c.758)
PM2_Supporting	✔	This variant is completely absent from population databases with at least 20x coverage for RUNX1.
PM5_Supporting	✔	Frameshift variant that is downstream of c.98

Not Met criteria codes

BS2	✖	This rule is not applicable for the MM-VCEP
BS1	✖	Variant is not present in population databases (gnomAD v2.1 or v3.1.2 or v4.0.0)
BS4	✖	This variant has not been reported in affected family members.
BS3	✖	This variant has not been featured in in vitro or in vivo functional studies that show no damaging effect on protein function or splicing.
BP4	✖	REVEL data only returned for missense/single nucleotide variants
BP3	✖	This rule is not applicable for the MM-VCEP
BP1	✖	This rule is not applicable for the MM-VCEP
BP2	✖	This variant has not been detected in a homozygous state in an individual or in a population database (gnomAD).
BP5	✖	This rule is not applicable for the MM-VCEP
BP7	✖	This is not a synonymous or intronic variant
PS1	✖	This is not a missense variant, and there has not yet been an amino acid change determined to be pathogenic at this amino acid residue.

PS2		✖	This variant has not been found to co-segregate with the disease in the literature.
PS3	ⓘ	✖	This variant has not been featured in in vitro or in vivo functional studies showing a damaging effect on the gene or gene product.
PS4	ⓘ	✖	This variant has not been reported in probands.
PM1		✖	This is not a missense variant
PM3		✖	This rule is not applicable for the MM-VCEP
PM4	ⓘ	✖	This is not an inframe variant
BA1	ⓘ	✖	Variant is not present in population databases (gnomAD v2.1 or v3.1.2 or v4.0.0)
PM6	ⓘ	✖	This variant has not been reported in probands in the literature.
PP1	ⓘ	✖	This variant was not found to co-segregate with disease in multiple affected family members.
PP2		✖	This rule is not applicable for the MM-VCEP
PP3	ⓘ	✖	REVEL data only returned for missense/single nucleotide variants
PP4		✖	This rule is not applicable for the MM-VCEP

Curation History

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