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.979_983dup (p.Thr328_Ala329insTer)

Version: 1.0

CA2580098829

1996223 (ClinVar)

Gene: RUNX1 (HGNC:861)
Condition: hereditary thrombocytopenia and hematologic cancer predisposition syndrome (MONDO:0011071)
Inheritance Mode: Autosomal dominant inheritance
UUID: 81c58b3a-7ad1-4850-8369-dd1b60699b0f
Approved on: 2023/12/9
Published on: 2023/12/9

HGVS expressions

NM_001754.5:c.979_983dup
NM_001754.5(RUNX1):c.979_983dup (p.Thr328_Ala329insTer)
NC_000021.9:g.34792598_34792602dup
CM000683.2:g.34792598_34792602dup
NC_000021.8:g.36164895_36164899dup
CM000683.1:g.36164895_36164899dup
NC_000021.7:g.35086765_35086769dup
NG_011402.2:g.1197113_1197117dup
ENST00000675419.1:c.979_983dup
ENST00000300305.7:c.979_983dup
ENST00000344691.8:c.898_902dup
ENST00000399240.5:c.706_710dup
ENST00000437180.5:c.979_983dup
ENST00000482318.5:c.*569_*573dup
NM_001001890.2:c.898_902dup
NM_001754.4:c.979_983dup
NM_001001890.3:c.898_902dup

Likely Pathogenic

Met criteria codes 3

PVS1_Strong PM2_Supporting PM5_Supporting

Not Met criteria codes 23

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

Evidence Links 0

Expert Panel

Myeloid Malignancy VCEP

Criteria Specification Information

Criteria Specifications for this VCEP

Evidence submitted by expert panel

Myeloid Malignancy VCEP

This frameshift variant is in a region that is critical to protein function, and transcript is not expected to undergo NMD (PVS1_Strong). This variant is completely absent from all population databases with at least 20x coverage for RUNX1 (PM2_supporting). This frameshift variant is downstream c.98 (PM5_supporting). In summary, this variant meets criteria to be classified as likely pathogenic. ACMG/AMP criteria applied, as specified by the Myeloid Malignancy Variant Curation Expert Panel for RUNX1: PVS1_strong, PM2_supporting, PM5_supporting.

Met criteria codes

PVS1_Strong	✔	This frameshift variant is in a region that is critical to protein function, and transcript is not expected to undergo NMD (PVS1_Strong).
PM2_Supporting	✔	This variant is completely absent from all population databases with at least 20x coverage for RUNX1 (PM2_supporting).
PM5_Supporting	✔	This frameshift variant is downstream c.98 (PM5_supporting).

Not Met criteria codes

PS1	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PS2	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PS3	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PS4	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PP1	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PP2	✖	This rule is not applicable for MM-VCEP
PP3	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PP4	✖	This rule is not applicable for MM-VCEP
PM6	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

PM1		✖	This variant is not a missense variant.
PM3		✖	This rule is not applicable for MM-VCEP
PM4	ℹ	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BA1	ℹ	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BS2		✖	This rule is not applicable for MM-VCEP
BS1	ℹ	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BS4	ℹ	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BS3	ℹ	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BP5		✖	This rule is not applicable for MM-VCEP
BP7	ℹ	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BP4	ℹ	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BP3		✖	This rule is not applicable for MM-VCEP
BP1		✖	This rule is not applicable for MM-VCEP
BP2	ℹ	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

Curation History

Showing 1 to 1 of 1 rows

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