

Variant: *NM_005343.4(HRAS):c.175_176delinsCT* (*p.Ala59Leu*)

Version: 1.0

CA273607 [↗](#)

179260 (ClinVar) [↗](#)

Gene: HRAS ([HGNC:3265](#))

Condition: RASopathy ([MONDO:0021060](#))

Inheritance Mode: Autosomal dominant inheritance

UID: 96293b0b-7e4a-4ba8-b473-a9d3c5f4e8fe

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HGVS expressions

NM_005343.4:c.175_176delinsCT

NM_005343.4(HRAS):c.175_176delinsCT (*p.Ala59Leu*)

NC_000011.10:g.533880_533881delinsAG

CM000673.2:g.533880_533881delinsAG

NC_000011.9:g.533880_533881delinsAG

CM000673.1:g.533880_533881delinsAG

NC_000011.8:g.523880_523881delinsAG

NG_007666.1:g.6670_6671delinsCT

ENST00000397594.7:c.175_176delinsCT

ENST00000417302.7:c.175_176delinsCT

ENST00000417302.6:c.175_176delinsCT

ENST00000462734.2:c.175_176delinsCT

ENST00000311189.8:c.175_176delinsCT

ENST00000311189.7:c.175_176delinsCT

ENST00000397594.5:c.175_176delinsCT

ENST00000397596.6:c.175_176delinsCT

ENST00000417302.5:c.175_176delinsCT

ENST00000451590.5:c.175_176delinsCT

ENST00000468682.2:n.663_664delinsCT

ENST00000479482.1:n.96_97delinsCT

ENST00000493230.5:c.175_176delinsCT

NM_001130442.1:c.175_176delinsCT

NM_005343.2:c.175_176delinsCT

NM_176795.3:c.175_176delinsCT

NM_001130442.2:c.175_176delinsCT

NM_001318054.1:c.-145_-144delinsCT

NM_005343.3:c.175_176delinsCT

NM_176795.4:c.175_176delinsCT

NM_001318054.2:c.-145_-144delinsCT

NM_001130442.3:c.175_176delinsCT

NM_176795.5:c.175_176delinsCT

Likely Pathogenic

PP2 PM1 PM6 PM2 PS4_Supporting

RASopathy VCEP

Criteria Specification Information

Criteria Specifications for this VCEP

Evidence Links 0

Evidence submitted by expert panel

RASopathy VCEP

The c.175_176delinsCT (p.Ala59Leu) in HRAS was absent from gnomAD (PM2; PMID: 29493581). It has been identified as a de novo occurrence (parentage unconfirmed) in 1 individual with clinical features of a RASopathy (PM6, PS4_Supporting; SCV000205760.4, PMID: 26918529). Furthermore, the p.Ala59Leu variant is in a location that has been defined by the ClinGen RASopathy Expert Panel to be a mutational hotspot of HRAS (PM1; PMID 29493581). The variant is located in the HRAS gene, which has been defined by the ClinGen RASopathy Expert Panel as a gene with a low rate of benign missense variants and pathogenic missense variants are common (PP2; PMID: 29493581). In summary, this variant meets criteria to be classified as likely pathogenic for RASopathies in an autosomal dominant manner. RASopathy-specific ACMG/AMP criteria applied (PMID:29493581): PP2, PM1, PM2, PM6, PS4_Supporting.

Met criteria codes

Table with 3 columns: Criteria Code, Status (checkmark), and Description. Rows include PP2, PM1, PM6, PM2, and PS4_Supporting, all with checkmarks and descriptions referring to the summary above or general recommendations.

Curation History

Filter controls including a search box and two dropdown menus.

Showing 1 to 1 of 1 rows

Table header area for the curation history, currently empty.

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