NM_000249.3(MLH1):c.191A>G (p.Asn64Ser)

Variation ID:	89947
Review status:	reviewed by expert panel

Interpretation

Go to:

Clinical significance: Uncertain significance
Last evaluated: Nov 3, 2016
Number of submission(s): 5
Condition(s):

- Lynch syndrome I [MedGen - OMIM]
- Lynch syndrome II [MedGen - OMIM]
- Lynch syndrome [MedGen - Orphanet - OMIM]
- Hereditary cancer-predisposing syndrome [MedGen]

See supporting ClinVar records

Allele(s)

Go to:

NM_000249.3(MLH1):c.191A>G (p.Asn64Ser)

Allele ID: 95421
Variant type: single nucleotide variant
Cytogenetic location: 3p22.2
Genomic location:

- Chr3: 36996693 (on Assembly GRCh38)
- Chr3: 37038184 (on Assembly GRCh37)

Other names:

- p.N64S:AAT>AGT

Protein change: N64S
HGVS:

- NG_007109.2:g.8344A>G
- NM_000249.3:c.191A>G
- NM_001167617.1:c.-99A>G
- NM_001258273.1:c.-517+3030A>G

...more

Links:

- UniProtKB: P40692#VAR_004438
- dbSNP: 63750952

NCBI 1000 Genomes Browser: rs63750952

Molecular consequence:

- NM_000249.3:c.191A>G: missense variant SO:0001583
- NM_001167617.1:c.-99A>G: 5 prime UTR variant SO:0001623
- NM_001258273.1:c.-517+3030A>G: intron variant SO:0001627

Allele frequency:

- GO-ESP 0.00008 (G)
- ExAC 0.00005 (G)

Assertion and evidence details

Go to:

1 Affected gene

mutL homolog 1 (MLH1) [Gene - OMIM - Variation Viewer]

Haploinsufficiency - *Sufficient evidence for dosage pathogenicity* (Nov 16, 2015)

Triplosensitivity - *No evidence available* (Nov 16, 2015)

Search ClinVar for variants within MLH1

Search ClinVar for variants including MLH1

Variant frequency in dbGaP

No dbGaP data has been submitted for this variant.

Browser views

RefSeqGene

Variation Viewer [GRCh38 - GRCh37]

UCSC [GRCh38/hg38 - GRCh37/hg19]

Related information

dbSNP

Functional Class

Gene

MedGen

OMIM

PMC

PubMed

Related genes (specific)

Clinical assertions | Summary evidence | Supporting observations

Germline

Filter:

Clinical significance (Last evaluated)	Review status (Assertion method)	Collection method	Condition(s) (Mode of inheritance)	Origin	Citations	Submitter - Study name	Subı acce
Uncertain significance (Nov 3, 2016)	reviewed by expert panel • Guidelines v1.9	research	Lynch syndrome I [MedGen | OMIM]	germline	• Other citation	InSiGHT	SCV
Uncertain significance (Oct 4, 2016)	criteria provided, single submitter • GeneDx Variant Classification (06012015)	clinical testing	not specified [MedGen]	germline		GeneDx	SCV
Uncertain significance (Jan 5, 2017)	criteria provided, single submitter • Invitae Variant Classification Sherloc (09022015)	clinical testing	Lynch syndrome [MedGen | Orphanet | OMIM]	germline		Invitae,	SCV
Uncertain significance (Nov 23, 2016)	criteria provided, single submitter • Ambry Autosomal Dominant and X-Linked criteria (10/2015)	clinical testing	Hereditary cancer-predisposing syndrome [MedGen]	germline		Ambry Genetics	SCV
Likely pathogenic (Sep 1, 2016)	criteria provided, single submitter • Counsyl Autosomal Dominant Disease Classification criteria (2015)	clinical testing	Lynch syndrome II [MedGen | OMIM]	unknown	• PubMed (7) [See all records that cite these PMIDs]	Counsyl	SCV

Last Updated: Oct 7, 2017