

# ClinVar

Genomic variation as it relates to human health

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## NM\_001032221.6(STXBP1):c.37+2dup

Cite this record

Interpretation:

Pathogenic/Likely pathogenic

Review status:

★ ★ ☆ ☆ criteria provided, multiple submitters, no conflicts

Submissions:

2

First in ClinVar:

May 4, 2020

Most recent Submission:

Sep 27, 2020

Last evaluated:

Aug 6, 2020

Accession:

VCV000870412.2

Variation ID:

870412

Description:

1bp duplication



### Variant details

#### NM\_001032221.6(STXBP1):c.37+2dup

Allele ID:

858591

Variant type:

Variant length:

1 bp

Cytogenetic location:

9q34.11

Genomic location:

9: 127612441-127612442 (GRCh38) [GRCh38](#) [UCSC](#)  
9: 130374720-130374721 (GRCh37) [GRCh37](#) [UCSC](#)

HGVS:

Nucleotide	Protein	Molecular consequence
<a href="#">NM_001032221.6:c.37+2dup</a> <a href="#">MANE SELECT</a>		splice donor
<a href="#">NM_001374306.2:c.37+2dup</a>		splice donor
<a href="#">NM_001374307.2:c.-101+2dup</a>		splice donor

... [more HGVS](#)

Protein change:

-

Other names:

-

Canonical SPDI:

NC\_000009.12:127612441:T:TT

Functional consequence:

sequence variant affecting splice donor [Sequence Ontology [SO:1000072](#)]

Global minor allele frequency (GMAF):

-

Allele frequency:

-

Links:

[dbSNP: rs1838431452](#)  
[VarSome](#)

Conditions

Aggregate interpretations per condition


Interpreted condition	Interpretation	Number of submissions	Review status	Last evaluated	Variation/condition record
<a href="#">Developmental and epileptic encephalopathy, 4</a>	Pathogenic/Likely pathogenic	2	criteria provided, multiple submitters, no conflicts	Aug 6, 2020	<a href="#">RCV001089985.2</a>

Clinical features observed in individuals with this variant





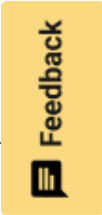
Epileptic encephalopathy  
Global developmental delay  
Dystonia

Gene(s)

Gene	OMIM	ClinGen Gene Dosage Sensitivity Curation		Variation viewer	Related variants	
		HI score	TS score		Within gene	All
STXBP1		Sufficient evidence for dosage pathogenicity	No evidence available	GRCh38 GRCh37	783	840

Submitted interpretations and evidence

Interpretation (Last evaluated)	Review status (Assertion criteria)	Condition (Inheritance)	Submitter	More information	
Likely pathogenic (Jun 09, 2018)	criteria provided, single submitter (ACMG Guidelines, 2015) Method: clinical testing	Developmental and epileptic encephalopathy, 4 Affected status: yes Allele origin: de novo	Victorian Clinical Genetics Services,Murdoch Childrens Research Institute Accession: SCV001244977.1 First in ClinVar: May 04, 2020 Last updated: May 04, 2020	Comment: A heterozygous duplication variant, NM_003165.3(STXBP1):c.37+2dupT, has been identified in intron 1 of 19 of the STXBP1 gene. This variant alters the conserved splice donor recognition ... (more)	
Pathogenic (Aug 06, 2020)	criteria provided, single submitter (ACMG Guidelines, 2015) Method: clinical testing	Developmental and epileptic encephalopathy, 4 (Autosomal dominant inheritance) Affected status: yes Allele origin: de novo	Institute of Molecular Medicine and Oncology,Chongqing Medical University Accession: SCV001431535.1 First in ClinVar: Sep 27, 2020 Last updated: Sep 27, 2020	Comment: We observed a young female patient with a de novo variant NM_003165.6 c.37+2dup of STXBP1.Her clinical symptoms overlapped with symptoms of STXBP1-E, including seizures, intellectual ... (more)	



Functional evidence

Functional consequence	Method	Result	Submitter	Supporting information (See all)
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Functional consequence	Method	Result	Submitter	Supporting information (See all)
sequence variant affecting splice donor (SO:1000072)			Institute of Molecular Medicine and Oncology,Chongqing Medical University Accession: SCV001431535.1 Submitted: (Sep 15, 2020)	Evidence details

Citations for this variant

There are no citations in ClinVar for this variation. If you know of citations for this variation, please consider submitting that information to ClinVar.

Text-mined citations for rs1838431452 none

These citations are identified by LitVar using the rs number, so they may include citations for more than one variant at this location. Please review the LitVar results carefully for your variant of interest.

Record last updated Apr 24, 2022 ⓘ

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