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A novel de novo mutation in the ASXL3 gene in a Chinese boy with

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Novel de novo frameshift variant in the ASXL3 gene in a ...

https://www.ncbi.nlm.nih.gov/pubmed/31180560

De novo sequence variants, including truncating and splicing variants, in the additional sex-combs like 3 **gene** (**ASXL3**) have been described as the cause of **Bainbridge-Ropers syndrome** (BRS). This pathology is characterized by delayed psychomotor development, severe intellectual disability, growth delay, hypotonia and facial dimorphism.

Cited by: 4 **Author:** Marketa Wayhelova, Jan Oppelt, Jan Sm...
Publish Year: 2019

Novel splicing mutation in the ASXL3 gene causing ...

https://onlinelibrary.wiley.com/doi/abs/10.1002/ajmg.a.37653

Apr 13, 2016 · **Novel** splicing **mutation in the ASXL3 gene** causing Bainbridge ... feeding problems, short stature, autism, and sleep disturbance with a heterozygous **de novo** splicing **mutation in the ASXL3 gene**. ... Global developmental delay and postnatal **microcephaly**: **Bainbridge-Ropers syndrome** with a new **mutation** in **ASXL3**, Neurología (English Edition), 10 ...

Cited by: 16 **Author:** Ikumi Hori, Fuyuki Miya, Kei Ohashi, Yut...
Publish Year: 2016

Novel compound heterozygous ASXL3 mutation causing ...

https://link.springer.com/article/10.1186/s13633-017-0047-9

Aug 04, 2017 · Background. **De novo** truncating and splicing mutations in the additional sex combs-like 3 (**ASXL3**) **gene** have been implicated in the development of **Bainbridge-Ropers syndrome** (BRPS) characterised by severe developmental delay, feeding problems, ...

Cited by: 3 **Author:** Dinesh Giri, Daniel Rigden, Mohammed D...
Publish Year: 2017

Novel Splicing Mutation in the ASXL3 Gene Causing ...

https://www.researchgate.net/publication/301300703...

Request PDF | **Novel** Splicing **Mutation in the ASXL3 Gene** Causing **Bainbridge-Ropers Syndrome** | **Bainbridge-Ropers syndrome** (BRPS) is characterized by ...

Novel compound heterozygous ASXL3 mutation causing ...

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Novel de novo frameshift variant in the ASXL3 gene in a ...

https://www.spandidos-publications.com/10.3892/mmr.2019.10303

A recently described **case** of a male child with atypical BRS also revealed the occurrence of a **novel** heterozygous de novo variant, p.P1010Lfs*14, **in the ASXL3 gene** . This variant leads to an identically truncated **ASXL3** protein as that seen in the proband of the present study, but the phenotypic features between the cases are different, namely ...

Cited by: 4 **Author:** Marketa Wayhelova, Jan Oppelt, Jan Sm...
Publish Year: 2019

Novel Splicing Mutation in the ASXL3 Gene Causing ...

https://www.researchgate.net/publication/301300703...

To date, almost all **ASXL3 gene** variants reported in the literature are nonsense **mutations** and frameshift **mutations**, except for one splice site **mutation** (c.3039+1G>A) [1]. We **report** a **novel** ...

De novo truncating mutations in ASXL3 are associated with ...

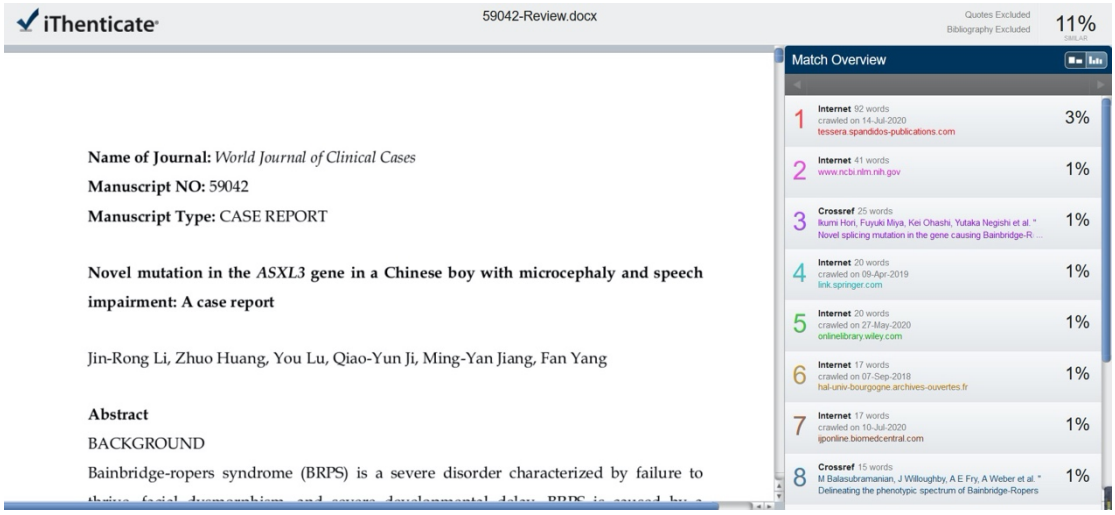
https://genomemedicine.biomedcentral.com/articles/10.1186/gm415

Feb 05, 2013 · Molecular diagnostics can resolve locus heterogeneity underlying clinical phenotypes that may otherwise be co-assigned as a specific syndrome based on shared clinical features, and can associate phenotypically diverse diseases to a single locus through allelic affinity. Here we describe an apparently **novel** syndrome, likely caused by de novo truncating **mutations** in **ASXL3**, which shares ...

Cited by: 128 **Author:** Matthew N Bainbridge, Hao Hu, Donna M...
Publish Year: 2013

A De Novo Mutation in DYRK1A Causes Syndromic ...

https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6877748



Name of Journal: *World Journal of Clinical Cases*
Manuscript NO: 59042
Manuscript Type: CASE REPORT

Novel mutation in the *ASXL3* gene in a Chinese boy with microcephaly and speech impairment: A case report

Jin-Rong Li, Zhuo Huang, You Lu, Qiao-Yun Ji, Ming-Yan Jiang, Fan Yang

Abstract

BACKGROUND

Bainbridge-ropers syndrome (BRPS) is a severe disorder characterized by failure to thrive, facial dysmorphism, and severe developmental delay. BRPS is caused by a

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Novel Splicing Mutation in the ASXL3 Gene Causing ...

https://www.researchgate.net/publication/301300703_Novel_Splicing_Mutation_in_the...

To date, almost all **ASXL3 gene** variants reported in the literature are **nonsense mutations** and **frameshift mutations**, except for one **splice site mutation** (**c**.3039+1G>A) [1]. We report a **novel** ...

Novel de novo frameshift variant in the ASXL3 gene in a ...

https://www.spandidos-publications.com/10.3892/mmr.2019.10303

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Cited by: 4 **Author:** Marketa Wayhelova, Jan Oppelt, Jan S...
Publish Year: 2019

(PDF) Novel compound heterozygous ASXL3 mutation causing ...

https://www.researchgate.net/publication/318915781_Novel_compound_heterozygous_ASXL3...

Results By obtaining genotype:phenotype data, we have been able to demonstrate a second **mutation** cluster region within **ASXL3**. This **report** expands the phenotype of older patients with BRPS; common ...

A De Novo Mutation in DYRK1A Causes Syndromic Intellectual ...

https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6877748

In conclusion, we identified a **novel** nonsense **mutation** in DYRK1A **in a Chinese** family, expanding the **mutation** spectrum of MRD7. Meanwhile, we provide a review of the formerly reported cases to summarize variations in DYRK1A **gene**, which can provide convenience for clinical application. And also, we emphasize the value of WES for genetic diagnosis ...