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Submicroscopic 11p13 deletion including the ELP4 gene in a girl wit

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SNP-array assay detected a heterozygous **11p13** microdeletion with a length of 518 kb in the proband, spanning two whole annotated genes, elongation factor protein 4 (**ELP4**), the paired box **gene** 6 ...

(PDF) 11p Microdeletion including WT1 but not PAX6 ...

https://www.researchgate.net/publication/24020807...

The WAGR syndrome is a multiple **congenital** anomaly–mental retardation syndrome caused by interstitial **deletion** of the distal portion of chromosome **11p13**. It is a contiguous **gene deletion** ...

Deletion and Duplication of 11p13-11p14: Reciprocal ...

https://www.researchgate.net/publication/51656327...

Aniridia can arise as part of the WAGR syndrome (Wilms tumour. aniridia, genitourinary anomalies, and mental retardation), due to a **deletion** or chromosomal region **11p13**. We **report** a **girl** with a ...

Narrowing of the Responsible Region for Severe ...

https://www.researchgate.net/publication/259395424...

Interstitial deletions of the **11p13** region are known to cause WAGR (Wilms tumor, aniridia, genitourinary malformation, and "mental retardation") syndrome, a contiguous **gene deletion** syndrome due ...

Planar Cell Polarity Gene Mutations in Autism Spectrum ...

https://www.sciencedirect.com/science/article/pii/B9780128001097000133

Jan 01, 2016 · For instance, a 2.28-Mb **deletion** encompassing the NF2 **gene** and a 1.61-Mb **deletion** containing the MN1 **gene** were found in a female with cleft palate, an open anterior fontanelle, and developmental delay. 293 Thus, orofacial clefting and IDs with later risks of developing tumors are commonly associated with the 22q12 region involving the NF2 **gene** ...

Cited by: 6 **Author:** Nathalie Sans, Nathalie Sans, Jérôme Ez...

Publish Year: 2016

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https://www.researchgate.net/publication/11074395...

8q12.1q12.3 de novo microdeletion involving the CHD7 **gene** in a patient without the major features of

Name of Journal: *World Journal of Clinical Cases*
Manuscript NO: 57995
Manuscript Type: CASE REPORT

Submicroscopic 11p13 deletion including the elongator acetyltransferase complex subunit 4 gene in a girl with language failure, intellectual disability and congenital malformations: A case report

Jaime Toral-Lopez, Luz María González Huerta, Olga Messina-Baas, Sergio A Cuevas-Covarrubias

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The ELP4 **gene** encodes a component of six-**subunit elongator complex**, a histone **acetyltransferase complex** that associates directly with RNA polymerase II during transcriptional elongation and this ...

Intellectual disability (Concept Id: C1843367)

https://www.ncbi.nlm.nih.gov/medgen/334384

Schinzel-Giedion syndrome is a highly recognizable syndrome characterized by severe mental

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Frontiers | Neurodevelopmental Genetic Diseases Associated ...

https://www.frontiersin.org/articles/10.3389/fgene.2018.00080