Transposition of the Great Arteries – Part of phenotypic spectrum associat

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Phenotypic spectrum associated with de novo and inherited deletions and duplications at 16p11.2 in individuals ascertained for diagnosis of autism spectrum disorder. J Med Genet. ... 14: Laitenberger G, Donner B, Gebauer J, Hoehn T. D-transposition of the great arteries in a case of microduplication 22q11 .2. Pediatr ...

Phenotypic manifestations of copy number variation in ... - NCBI - NIH

https://www.ncbi.nlm.nih.gov › NCBI › Literature › PubMed Central (PMC) - 翻译此页

作者：SCS Nagamani - 2011 - 被引用次数：55 - 相关文章

2010年12月8日 - Recurrent deletions and reciprocal duplications in 16p13.11 have been ... Owing to the widespread spectrum associated with CNVs of the region, it is ... OLIGO (105K) array and patient 2 whose sample was run on V6.5OLIGO (44K) array. ... the other had transposition of great vessels with aortic coarctation.

Cytogenomic Evaluation of Subjects with Syndromic and ... - NCBI

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作者：KR de Souza - 2015 - 被引用次数：4 - 相关文章

2015年6月7日 - We classified the CNVs (gains/duplications and losses/deletions) into the .... In syndromes with CHD as part of the clinical spectrum, conotruncal heart ... Both 16p11.2 deletion and duplication are associated with ASD, whereas only ..... protein 1 (ISL1) is associated with d-transposition of the great arteries.

OMIM Entry - # 608363 - CHROMOSOME 22q11.2 DUPLICATION ...

https://www.omim.org/entry/608363 ▾ 翻译此页

Name of the Journal: *World Journal of Cardiology*

Manuscript NO: 34049

Manuscript Type: Case Report

Transposition of the great arteries – a phenotype associated with 16p11.2 duplications?

Zarmiga Karunanithi, Else Marie Vestergaard, Mette H Lauridsen

Abstract

Genetic analyses of patients with transposition of the great arteries have identified rare copy number variations, suggesting that they may be significant to the aetiology of the disease. This paper reports the identification of a 16p11.2 microduplication, a variation that has yet to be reported in association with transposition of the great arteries. The 16p11.2 microduplication is associated with autism spectrum disorder and developmental delay, but with highly variable phenotypic effects. Autism and attention deficit disorders are observed more frequently in children with congenital heart disease than in the general population. Neonatal surgery is proposed as a risk factor, but as yet unidentified genetic

Transposition of the great arteries – a phenotype associated with 16p11.:

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2: Mukaddes NM, Herguner S. Autistic disorder and 22q11.2 duplication. ... spectrum associated with de novo and inherited deletions and duplications at 16p11.2 in ... Hoehn T. D-transposition of the great arteries in a case of microduplication 22q11.2. ... are frequently inherited and are associated with variable phenotypes.

16p11.2 duplication - Genetics Home Reference

https://ghr.nlm.nih.gov/condition/16p112-duplication ▾ 翻译此页

3 天前 - 16p11.2 duplication is a chromosomal change in which a small amount ... a 16p11.2 duplication have problems related to speech or language.

缺少字词： ~~transposition great arteries~~

16p11.2 duplication | Genetic and Rare Diseases Information Center ...

https://rarediseases.info.nih.gov/diseases/12388/16p112-duplication ▾ 翻译此页

Susceptibility to Autism, 14B; AUTS14B; 16p11.2 duplication syndrome; ... The signs and symptoms present in individuals with a 16p11.2 duplication ... These resources provide more information about this condition or associated symptoms. ... and symptoms (phenotypes) of different diseases and discover common features.

缺少字词： ~~transposition great arteries~~

[PDF] 16p11.2 microduplications - Unique - Rare chromo

www.rarechromo.org/.../16p11.2%20microduplications%20FTNP.pdf ▾ 翻译此页

The information is believed to be the best available at the time of publication ... 16p11.2 duplication and summaries of relevant recent journal articles. ... Fernandez 2010: Phenotypic spectrum associated with de novo and inherited deletions and ... This is a channel between the two major

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PubMed Result - NCBI

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2: Mukaddes NM, Herguner S. Autistic disorder and 22q11.2 duplication. ... spectrum associated with de novo and inherited deletions and duplications at 16p11.2 in ... Hoehn T. D-transposition of the great arteries in a case of microduplication 22q11.2. ... are frequently inherited and are associated with variable phenotypes.

Genetics of Congenital Heart Disease - NCBI - NIH

https://www.ncbi.nlm.nih.gov › NCBI › Literature › PubMed Central (PMC) - 翻译此页

作者：AA Richards - 2010 - 被引用次数：100 - 相关文章

The phenotype of Noonan syndrome consists of cardiac defects, typically ... Table 2. Common Syndromes Associated with CHD Resulting from Single ... in a patient with d-transposition of the great vessels and mental retardation, ... More recently, a microdeletion or reciprocal microduplication of chromosome 16p11.2 was ...

16p11.2 duplication - Genetics Home Reference

https://ghr.nlm.nih.gov/condition/16p112-duplication ▾ 翻译此页

6 天前 - 16p11.2 duplication is a chromosomal change in which a small amount ... a 16p11.2 duplication have problems related to speech or language.

缺少字词： ~~transposition~~ ~~great~~ ~~arteries~~

Phenotypic manifestations of copy number variation in chromosome ...

https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3061988/ - 翻译此页

作者：SCS Nagamani - 2011 - 被引用次数：59 - 相关文章

2010年12月8日 - The phenotypes associated with CNVs of 16p13.11 are not consistent .... whereas the other had transposition of great vessels with aortic coarctation. ... Table 2. Clinical features of patients with duplication of 16p13.11 .... Discovery of a previously unrecognized microdeletion syndrome of