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Chromosomal imbalance report

A novel microdeletion syndrome at 9q21.13 characterised by mental retardation, speech delay, epilepsy and characteristic facial features

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Abstract

The increased use of array-CGH and SNP-arrays for genetic diagnosis has led to the identification of new microdeletion/microduplication syndromes and enabled genotypeâ€“phenotype correlations to be made. In this study, nine patients with 9q21 deletions were investigated and compared with four previously Decipher reported patients.

Genotypeâ€“phenotype comparisons of 13 patients revealed several common major characteristics including significant developmental delay, epilepsy, neuro-behavioural

disorders and recognizable facial features including hypertelorism, feature-less philtrum, and a thin upper lip. The molecular investigation identified deletions with different breakpoints and of variable lengths, but the 750Â kb smallest overlapping deleted region includes four genes. Among these genes, *RORB* is a strong candidate for a neurological phenotype.

To our knowledge, this is the first published report of 9q21 microdeletions and our observations strongly suggest that these deletions are responsible for a new genetic syndrome characterised by mental retardation with speech delay, epilepsy, autistic behaviour and moderate facial dysmorphy.

Highlights

â–° First published report of a novel microdeletion syndrome at 9q21. â–° Syndrome characterized by developmental delay, epilepsy and mild facial dysmorphy. â–° The 750Â kb smallest overlapping region includes 4 genes: *RORB*, *TRPM6*, *NMRK1*, *OSTF1*. â–° *RORB* is a strong candidate for neurological phenotype, notably mental retardation.

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Keywords

9q21.13; Interstitial deletion; Mental retardation; Epilepsy; *RORB*; Array-CGH

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