Case Report

Distinctive Phenotype in a Case of Ring Chromosome 22 with Features of 22q13.3 Deletion Syndrome

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Abstract

Chromosome 22q13.3 deletion syndrome (OMIM #606232) is a well defined clinical subtelomeric deletion syndrome characterised by severe expressive language delay, moderate mental retardation and somatic overgrowth without major internal organ anomalies and minimal cranio-facial dysmorphic features. We report a case of de novo ring chromosome 22 confirmed by FISH to have deletion 22q13.3, with typical features of 22q13.3 deletion syndrome; we emphasise the importance of cytogenetic analysis in children with severe speech delay, autism, hypotonia, developmental delay, accelerated growth and minimal cranio-facial dysmorphism.

Keyword: 22q13.3; Autism; Severe expressive language delay; Subtelomeric deletion syndrome