Case Report

Smith-Magenis Syndrome in Chinese with Previously Unreported Craniofacial Features and Association with Joubert Phenotype

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Abstract

Smith-Magenis syndrome (SMS) is a clinically recognisable multiple congenital anomalies syndrome marked by distinctive craniofacial dysmorphism, behavioural and neurological abnormalities. Other additional abnormalities depend on the extent of contiguous genes involvement. We compared the clinical features of four SMS patients; and report new, consistent and previously unrecognised craniofacial features in the syndrome; and in addition, reporting the second case of SMS with Joubert phenotype in literature.

Keyword : Deletion 17p11.2; Joubert syndrome; RAI1; Smith-Magenis syndrome (SMS)