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Longitudinal Study in a Patient with Trisomy 8 Mosaicism: Cytogenetic and Molecular-Genetic Investigations over a Period of Eleven Years

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ABSTRACT We report on the cytogenetic and molecular-genetic investigations of a child with mosaic trisomy 8, analysed over a period of eleven years. The female patient showed clinical features and facial dysmorphisms characteristic of the syndrome as well as mentally impairment. The mosaic trisomy 8 was diagnosed prenatally in amniotic fluid cells (38%) and fetal lymphocytes (78%) and was confirmed postnatally in the umbilical cord (52%), placental biopsy (40%), lymphocytes (between 55% and 70%) and buccal mucosa cells (between 30% and 42%), demonstrating the overall prevalence of the trisomy 8 cell line in this patient was quiet high. We found no evidence of an appreciable increase or decrease in the frequency of the trisomic cell line over a period of eleven years. Molecular genetic investigation demonstrated the maternal origin of the additional chromosome 8.

Home Back