TRISOMY 8 MOSAIC SYNDROME IN TWO CHILDREN FROM BULGARIA

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ABSTRACT

Trisomy 8 is defined as the presence of full copies of chromosome 8 in all human cells. Mosaic trisomy 8 describes the situation that occurs when only a portion of these cells contains trisomy 8, while others contain the usual two copies of that chromosome. The condition is also referred to as trisomy 8 mosaicism (T8mS) or mosaic Warkany syndrome. Full trisomy 8 occurs in about 0.1% of spontaneous miscarriages. It is estimated to occur in about 0.1% of all recognized pregnancies. When seen at birth it is always due to mosaic trisomy 8, as opposed to full trisomy 8. The study of Nielson et al. (1991) found one child with T8mS among 34 910 newborns. To present cases known about 75 cases.

INTRODUCTION

There was identified trisomy 8 mosaic syndrome in 2 dysmorphic boys. The chromosomal analysis in peripheral blood showed trisomy 8p47, XY+8{3}/46,XY{53} and 47,XY{3}/34,XY{53}. They had typical clinical features: moderate mental retardation, dysmorphic faces, low-set malformed ears, strabismus and ptosis, bone and tissue abnormalities, sole creases- furrowed appearance, hydrocephaly, agenesis of the corpus callosum and cryptorchidism. Although the low percent 12% of mosaic trisomy 8 in our cases, the clinical picture was expressive. This confirmed the opinion that the percentage of cells with trisomy 8 does not appear to correlate with the types of symptoms in the affected persons.

RESULTS

1. Case I: S.T.G (date of birth: 18.08.98) is a 9 year's old boy with a Gipsy origin , born from third normal pregnancy, after which the mother had 7 spontaneous miscarriages. The birth weight was 2500gr and length-47cm. He had dysmorphic face: microcephaly, micrognathia, a lot of freckles on the tongue and alveolar ridges, for which he was operated. He had thalassemia deformity, camptodactily, and contractures of the fingers, kyphosis. The metabolic screening was normal. The EEG was normal. The CAT showed internal hydrocephaly and agenesis of corpus callosum. The chromosomal analysis in peripheral venous blood showed 47,XY+8{3}/46,XY{53}. The investigation of the parents proceeds.

2. Case II: D.A.D. (date of birth: 20.11.98) is a 9 year's old boy from Bulgarian-Turkish origin, born from first pregnancy with vacuum extraction. During his childhood he showed food allergy, recurrent bronchitis and asthma. He had mental, speech retardation and vitiligo at 7 year's old. The general condition of the boy was good with weight-24 kg, height-128 cm; head circumference 53 cm. He was dysmorphic with strabismus, ptosis of left eye lid, prominent nose, thinned and downturned lower lip, alveolar anomalies, hypotelorism, hypoplasia of the rears, and external hydrocephaly. His IQ= 42 /Wechsler/-moderate mental retardation with aggressive and autoaggressive behaviour. The CAT showed internal hydrocephaly. The metabolic screening was normal. The chromosomal analysis showed trisomy 8p47, XY+8{56}/46,XY{20}. The IQ of D.A.D. (date of birth: 10.11.98) is a 9 year's old boy from Bulgarian-Turkish origin, born from first pregnancy with vacuum extraction. During his childhood he showed food allergy, recurrent bronchitis and asthma. He had mental, speech retardation and vitiligo at 7 year's old. The general condition of the boy was good with weight-24 kg, height-128 cm; head circumference 53 cm. He was dysmorphic with strabismus, ptosis of left eye lid, prominent nose, thinned and downturned lower lip, alveolar anomalies, hypotelorism, hypoplasia of the rears, and external hydrocephaly. His IQ= 42 /Wechsler/-moderate mental retardation with aggressive and autoaggressive behaviour. The CAT showed internal hydrocephaly. The metabolic screening was normal. The chromosomal analysis showed trisomy 8p47, XY+8{56}/46,XY{20}.

CONCLUSION

We identified Trisomy 8 mosaic syndrome in two dysmorphic boys. The diagnosis was confirmed in peripheral blood cells. The clinical examination and the two treatment procedures.

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DISCUSSION

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