

ea0039 P1-163

CASE REPORT-Chromosome 9p Trisomy with insulin dependent diabetes.

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Introduction

Trisomy 9p is one of the most frequent autosomal anomalies compatible with long survival rate. It is the fourth most frequent chromosome anomaly in live-borns after 21, 18 and 13¹. Most often these children present with developmental delay, craniofacial malformation and growth deficiency. We present a case of insulin dependent diabetes in a 5 year old boy known to have a diagnosis of Chromosome 9p trisomy.

Case

Born at 38+3, by normal vaginal delivery with birth weight of 2980g (25th cent.), birth head circumference 33cm (25th cent.). Mum – G2Po, TIOP 9/40 for personal reasons, age 29yrs; well during pregnancy. He is the first born of non-consanguineous parents.

Noted to have soft dysmorphic features at birth- left correctable talipes calcaneovalgus, apparent short penis, small cup like ears, marked tongue tie, no cleft palate, hypoplastic toe nails, widely placed nipples. Rest of the physical exam was normal.

Chromosomal analysis revealed unbalanced male karyotype with two additional abnormal chromosomes. The larger of the additional chromosome appears to comprise of short arm and proximal long arm of chromosome 9- therefore trisomic for this region. No evidence of mosaicism. Parental studies showed that the abnormal chromosomes detected, is of de novo occurrence.

He was delayed in his motor, speech and language development. His cardiac ECHO was normal.

Aged 4, he presented with h/o polyuria, polydipsia and weight loss in Diabetic Keto acidosis. He was commenced on CSII once his acidosis improved. He continues to remain on CSII with insulin requirement of around 0.5 U/kd/day.

Conclusion

There have been no reported cases of insulin dependent diabetes in this syndrome. Early management with CSII should be considered based on age and underlying degree of developmental impairment.

¹SA Temtamy et al. Phenotypic and cytogenetic spectrum of 9p Trisomy. Genetic Counseling Vol.18,No1,2007,pp29-48.