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Partial Monosomy 7q

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We report a case of partial monosomy 7q and partial trisomy 14q in a 4 year old male with microcephaly, prominent eyes, arched eyebrows, malformed ears and overlapping of toes. The unbalanced rearrangement resulted in monosomy of 7q33→qter and trisomy of 14q32.2→qter. The clinical phenotype was similar to the other cases of 7q deletion.

Key words: 7q monosomy, 14 q trisomy, Mental retardation, Translocation.

Reciprocal translocation carriers are at the risk of having a mentally and physically abnormal child because of “segmental aneusomy”. The imbalance is due to duplication or deletion of the chromosome segment involved in segregation. Partial autosomal monosomies and trisomies, although associated with congenital malformations, are known to be compatible with life.

7q deletions have been reported in more than 30 cases as either an isolated deletion or in combination with other chromosomal anomalies [1]. In most of the cases the associated clinical features are highly variable, and are found to share a few common features like microcephaly, broad nasal bridge, bulbous nasal tip, auricular malformations, micrognathia and genital anomalies, which delineate a distinct phenotype as ‘7q terminal deletion

CASE REPORTS

syndrome' [2]. Individuals with 7q monosomy are known to have malformations like holoprosencephaly and mid structural defects [2] and various grades of caudal deficiency sequence [3]. We herein describe a male patient with partial 7q monosomy and partial trisomy of 14q resulting from paternal $t(7;14)(q33;q32.3)$.

CASE REPORT

The patient presented to us as a 4-year-old male born to unrelated parents. The couple had history of two previous first trimester abortions. There was no history of antenatal intake of drugs or any systemic illness in the mother. The patient's birth history was uneventful and birth weight was reported as average. There was history of delayed milestones and at age of 4 years he was not able to stand with support or feed himself. There was no speech development and the patient was not able to obey commands. There was no history of seizures or any other problems. On examination, the patient had height of 91 cm (less than 5th centile), and head circumference of 46.5 cm (less than 5th centile). The dysmorphic features included microcephaly, malformed low set ears, depressed nasal bridge, bulbous nasal tip and overriding of fingers and toes (**Fig 1**).

His metabolic screening and MRI brain were normal. Abdominal ultrasound for renal anomalies and echocardiography was normal. Chromosomal analysis was performed on peripheral lymphocytes following standard procedure. The karyotype at 550 band level, showed an unbalanced chromosomal



FIG. 1 Facial features and overriding toes in proband.

rearrangement (46, XY del [7] (q33-qter). A total of 50 metaphases were analyzed and all showed abnormal karyotype, thus ruling out mosaicism. Parental cytogenetic analysis revealed a balanced reciprocal translocation between 7q and 14q in father (46, XY, $t(7;14)(q33;q32.3)$). Maternal karyotype was normal. Consequently, the proband's chromosomal imbalance was interpreted as partial monosomy 7q33-qter and partial trisomy 14q32.3-qter (46, XY, der (7), $t(7;14)(q33-q32.2)$ pat (**Fig. 2**).

DISCUSSION

Our patient did not have premaxillary agenesis single central incisor, holoprosencephaly, stigmata of caudal deficiency sequence, or congenital cardiovascular, renal/urinary or adrenal defect. Thus the 7q deletion in the index patient resulted in a relatively mild phenotype as compared to the literature [3].

Our patient did not show significant cerebral malformations, typical signs of holoprosencephaly,

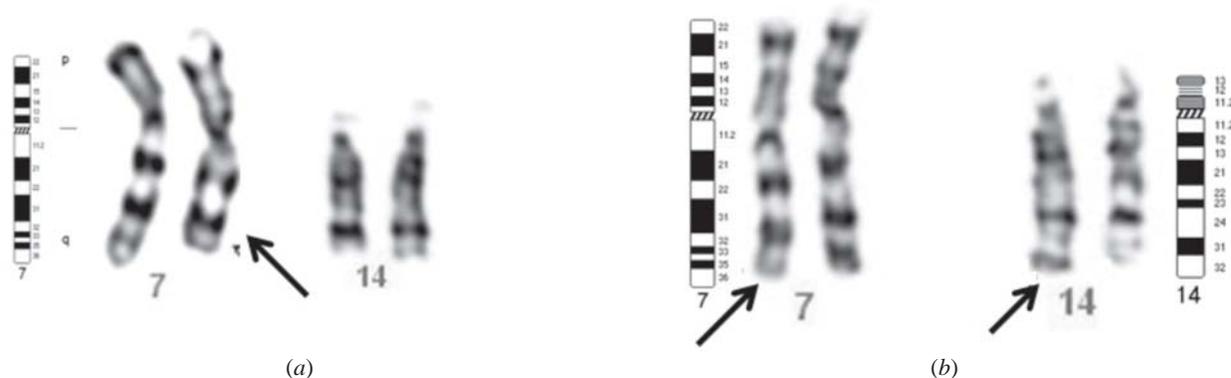


FIG. 2 (a) Partial karyotype of the boy with partial monosomy 7q33 showing normal chromosomes 7, 14 and a derivative chromosome 7 (arrow). (b) Partial karyotype of the father with balanced $t(7;14)(q33;q32.3)$ showing normal and derivative (arrows) chromosomes 7 and 14.

or relatively severe manifestations of the syndrome including , caudal deficiency sequence, congenital heart malformation as reported in most terminal 7q deletion cases [1-5,7,8]. The phenotype results from deletion of various genes distal to the breakpoint on 7q like Sonic Hedgehog (SHH), HLXB9 etc leading to haploinsufficiency of these genes. However our patient is similar to cases with 7q deletion with a mild phenotype [6,9,10]. This shows the wide phenotypic spectrum of cases with 7q deletion.

Patients with partial trisomy 14q show significant variability which may be due to the length of the 14q segment involved, or the nature of the other chromosome segment involved in the reciprocal translocation. The clinical features vary from severe to mild dysmorphic features associated or not with chromosome aberrations. The clinical features in our patient are consistent with those described in 7q deletion syndrome. Although the patient also had a small partial 14q trisomy, we cannot comment about the significance of this in relation to patient's phenotype. The typical partial 14q trisomy phenotype was not seen in the proband probably due to small size of partial trisomy or because this segment may contain genes that are not subject to dosage effect.

The identification of balanced translocation in the father helped us to counsel the family regarding risk of unbalanced chromosomal rearrangements in fetuses in subsequent pregnancies of the couple. The couple was counseled that there is risk of chromosomal imbalance in all future pregnancies. They were advised regarding availability of prenatal diagnosis and termination of pregnancy.

This case report further emphasizes the importance of doing karyotype in all cases of mental retardation, even if there are few dysmorphic features. The testing of proband will help both in counseling the parents regarding the cause of disease in their child as well as prevention of recurrence of

the abnormality in future pregnancies.

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