A Translocation Between
Chromosome 1 and 10 in a Boy
with Mental Retardation and
Dysmorphic Features

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This report describes a boy with delayed physical and mental milestones, dysmorphic features, congenital anomalies of the feet and convulsions. He was found to have translocation between long arms of 1st and 10th chromosome. Such a 1:10 translocation with phenotypic consequences has not been reported in Indian literature so far.

Case Report

A two-year-old proband was a product of Grade I consanguinous marriage between healthy young first cousins. The antenatal period was uneventful and the child was delivered normally. Abnormalities of both feet were noted at the time of birth. The child was referred to us for convulsions; generalised tonic and clonic, off and on for one year. History revealed that mental and physical milestones were delayed even before the onset of convulsions.

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Clinical examination revealed a microcephalic child with length and weight less than 10th centile for the age. Anterior forntanel was 1 cm × 0.5 cm in size. A beak shaped nose along with micrognathia was present. Dentition was normal. Both feet had congenital talipes equinovarus deformity. Partial simian crease was present unilaterally while both palms were flat with ill demarcated flexor creases pattern. The mental and physical milestones achieved were of 6 to 7 months of age.

Cytogenetic Studies: Peripheral blood lymphocytes were cultured following the standard method of Moorhead et al. and chromosomes were analysed by trypsin Leishman banding(1). Analysis of blood revealed a deletion of a major portion of the q arm of chromosome No. 10, translocated on to the q arm of chromosome No. 1 in all cells. The Y chromosome too was abnormally large (Fig. 1). The karyotype was determined as 46, XY, t(1q: 10q) (q4400: q2100) (Fig. 2)

Fig. 1. Karyotype showing translocation between 1st and 10th chromosome.

Fig. 2. Partial karyotype showing translocation between long arms of 1st and 10th chromosome.

Discussion

Prior to the advent of banding techniques, it was thought that several chromosomes are spared from getting aberrant. But now it has been shown that each and every chromosome can be implicated in such exchanges. We herewith describe a tandem translocation involving chromosome 1 and 10. The terminal segments of long arm of chromosome 10 have got dislodged and attached to terminal segment of long arm of 1 without a demonstrable loss of material from chromosome 1 (Figs. 1 & 2). This is possibly one of the few reports in the world literature involving long arms of chromosome 1 and 10, but only one of them was symptomatic(2-4).

Ours is likely to be the first report where the translocation is likely to have resulted in somatic aberration in the form of dysmorphic features, talipes equinovarus, mental and motor retardation. Bonfante has described a familial translocation and
in one case in his article, described a neo-
ama. Dying after 26 days and autopsy

revealing cardiac anomalies.

Boue et al. described a translocation
causing partial monosomy of chromosome
10 and partial trisomy of chromosome 1 in
a spontaneous abortus. In the present case
as parents refused to give blood for karyo-
typing, it may be difficult to comment on
the inheritance of the translocation and
whether it is a balanced one or not. Appar-
ently, the dysmorphic features, mental and
motor delay, talipes equinovarus could be
related to the tandem translocation but
definite correlation may not be possible.
Though this is so, it is important from the
point of view of studying position effect
phenomenon in individuals with apparently
balanced translocation with phenotypic
consequences.

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Cerebral Gigantism
(Sotos Syndrome)

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Sotos and colleagues in 1964 described
a new syndrome characterised by exces-
sively rapid growth with acromegalic fea-
tures and a non-progressive neurological
disorder in 5 patients(1). This disorder is
now known as cerebral gigantism (Sotos
syndrome) and is well-recognized by the
presence of salient features such as ad-
vanced height, weight and bone age with
distinctive facies characterised by large
dolicocephalic head, hypertelorism, anti-
mongoloid slant of the palpebral fissures,
high arched palate, long arm span and
large hands and feet(2). Most children are
mentally retarded and clumsy with no gross
neurological abnormalities. Radiological
studies demonstrate ventricular enlarge-
ment in most of them. Over 150 patients
have been reported in world literature
since the original publication. On a survey
of Indian literature, we could come across
only two case reports of this condition.

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