Interstitial deletion of 2(q33q36) in a child with congenital abnormalities

Deletions of the long arm of chromosome 2 have been infrequently observed. Specifically, few cases of de novo 2q interstitial deletions involving 2(q33q36) have been reported.\(^1\) We present a previously unreported observation of deletion 2(q33q36) found in a patient with severe developmental delay and multiple congenital anomalies.

A 15 year old white female was referred to the C S Mott Children's Hospital for evaluation and treatment of recurrent aspiration pneumonia, multiple congenital anomalies, and severe developmental delay. Failure to respond to medical therapy in the past had led to tracheostomy, gastrostomy, oversewing of the vocal cords, and salivary gland excision. She was the only child of a healthy, unrelated 19 year old mother and a 21 year old father. Delivery was spontaneous, normal, and at term after a pregnancy complicated by pre-eclampsia requiring the mother to be confined to bed for the last two months of pregnancy. Birth weight was 1690 g. Dysmorphic facial features and musculoskeletal anomalies were noted at birth. All milestones were significantly delayed. She rolled over at 12 months, sat unsupported at 24 months, stood unaided at five years, and spoke single words at 10 years. Her IQ was estimated as 20. She had been treated for a generalised seizure disorder in infancy. At three years, an IVP following multiple urinary tract infections had shown left hydronephrosis.

When examined, the patient was unable to stand or speak and exhibited generalised writhing, dystonic movements. Her growth parameters were all well below the 3rd centile with a OFC of 48.5 cm, a weight of 25.5 kg, and length of 142 cm. Unusual facial features consisted of deeply set eyes, downward slanting palpebral fissures, low set ears, a prominent nasal bridge, and micrognathia (fig 1). A 43° thoracolumbar scoliosis involving T10 to L4, bilateral ulnarly deviated hands, coxa valga, hind foot valgus, and pes cavus with diffuse osteoporosis were shown radiographically.

Chromosomal analyses were performed on PHA stimulated peripheral lymphocytes using trypsin-Giemsa banding techniques. A total of 20 cells was analysed and all showed an interstitial deletion of the long arm of one chromosome 2 homologue. The karyotype was interpreted as 46,XX,del(2)(q33q36) (fig 2). The deleted material did not appear to be translocated to other chromosomes in the karyotype. The karyotype of the patient's mother was 46,XX. The father was unavailable for chromosome analysis.

Though no consistent phenotype emerges from a comparison of our patient and other previously reported cases of interstitial 2q deletions,\(^1\) several non-specific clinical findings are shared by the majority of patients, including low birth weight, failure to thrive, psychomotor retardation, microcephaly, and deformed, low set ears. Most striking,
however, is the apparent sparsity of clinical similarities among cases sharing relatively large portions of 2q monosomy, such as our present case and that of Narahara et al,\(^2\) which overlaps with our patient in being monosomic for 2(q33q35), and that of Warter et al,\(^3\) which overlaps with our patient in being monosomic for 2(q34q36). The case described by Narahara et al\(^2\) was unique in having strabismus; the case of Warter et al\(^3\) is set apart in having a small nose, macrostomia, and abnormal dentition; while the present case is distinctive in having antimongoloid slanted palpebral fissures, micrognathia, scoliosis, unlarly deviated extremities, and recurrent aspiration pneumonia. The clinical heterogeneity of these patients does not at present permit us to delineate interstitial 2q deletion syndrome in the absence of additional patients. The case reported here is the only one of which we are aware showing a deletion involving the region 2(q33q36).

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Interstitial deletion of 11q

A male patient with a interstitial deletion 11 (q13::q21) showed retarded growth and mental development, craniofacial abnormalities characterised by quadricephaly and coarse facial features, digital anomalies, and anomalies of the limbs. The clinical features of our patient were compared with other reported cases of interstitial deletion 11q.

The proband, a two month old infant, was the term product of the third pregnancy of a 28 year old mother and a 30 year old father. During the first trimester the mother received hormonal therapy and vitamin E. Delivery was uneventful, birth weight was 3000 g, and birth length 52 cm.

The first pregnancy ended in a first trimester spontaneous abortion and an older male sib was healthy. The parents were healthy and non-consanguineous and there was no family history of congenital defects.

The proband showed inadequate weight gain and poor suckling from birth. On admission to our Institute, the child weighed 3650 g (fig 1) and craniofacial dysmorphism was noted. The head was quadricephalic with frontal bossing and the head circumference was 39.5 cm. Downward slanting palpebral fissures, telecanthus, low set, malformed ears with attached lobules, micrognathia, and a high arched palate were noted. A grade 2-3/6 systolic murmur was audible, but ECG and x ray examination were normal. A very short sternum, low set umbilicus, and a hydrocele were also noted. Ortholany’s sign was positive and x ray studies showed dislocation of the hips. There were also malformations of the fifth toes, a contracture of the right elbow joint, and long, thin fingers. Neurological examination showed poor eye contact and the child was not interested in his surroundings. There was increased muscle tone in the limbs, poor head control, a positive Moro reflex, and slightly increased tendon reflexes. Audiopalpebral reflex and pupillary reactions to light were normal.

G banded chromosomes from blood lymphocytes showed a 46,XY,del(11)(pter→qter) karyotype in all metaphases analysed (fig 2). The parents had normal karyotypes.

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FIG 1 The proband at two months.
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