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., which overlaps with our patient in being monosomic for 2(q33q35), and that of Warter et al., which overlaps with our patient in being monosomic for 2(q33q36). The case described by Narahara et al. was unique in having strabismus; the case of Warter et al. 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).

JEROME L GORSKI, MI KYNE, WENDY UHLMANN, KATHY Loeffler, and THOMAS W GLOVER
Division of Pediatric Genetics, Departments of Pediatrics and Communicable Diseases, and Human Genetics, University of Michigan Medical Center, Ann Arbor, Michigan 48109, USA.

Interstitial deletion of 11q

A male patient with an interstitial deletion 11 (q13::q21) showed retarded growth and mental development, craniofacial abnormalities characterised by quadriicepsphaly 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 sucking from birth. On admission to our Institute, the child weighed 3650 g (fig 1) and craniofacial dysmorphism was noted. The head was quadriicepsphaly 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→q13::q21→qter) karyotype in all metaphases analysed (fig 2). The parents had normal karyotypes.

Correspondence to Dr Jerome L Gorski, Division of Pediatric Genetics, Departments of Pediatrics and Communicable Diseases, and Human Genetics, D1109 Medical Professional Building, Box 0718, Ann Arbor, Michigan 48109, USA.

References


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FIG 1 The proband at two months.
The 11q syndrome owing to terminal deletion of the long arm of chromosome 11 shows the following clinical manifestations: trigonocephaly with frequent microcephaly, hypoplastic nasal bridge, abnormal eye slant, carp shaped mouth, retrognathia, low set, malformed ears, digital anomalies, and cardiac anomalies. As far as we know, there are only six reported cases of interstitial deletion of the long arm of chromosome 11. All these patients showed less pronounced or absent signs of the 11q deletion syndrome.

From the clinical standpoint, our patient had retarded growth and mental development, quadriencephaly, micrognathia, high arched palate, low set, abnormal ears, digital anomalies, and anomalies of the limbs.

Our proband is a male although most of the reported cases of terminal or interstitial deletion of the long arm of chromosome 11 are female. Sirota et al. suggest that "the sex chromosome constitution determines the expression of an 11q deletion or contributes to a survival difference".

Establishing a definite phenotype for interstitial deletion of 11q will require more cases studied by high resolution banding techniques.

References

Correspondence to Dr M Guć-Šekić, Department of Paediatrics, Mother and Child Health Institute, 8 Radoja Dakića St, 11070 Novi Beograd, Yugoslavia.