Partial Monosomy of the Long Arm of Chromosome 16 in a Malformed Newborn: Karyotype 46,XX,del(16)(q21)

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Summary. A polymalformed newborn with partial monosomy for the long arm of chromosome 16 is presented. Karyotype 46,XX,del(16)(q21).

Numerical abnormalities of chromosome 16 have never been reported in live-born infants, and only one case of partial trisomy for the long arm of chromosome 16 in a polymalformed newborn male child (karyotype 47,XY,+16q) has been published (Schmickel et al., 1975).

This report describes the first case of partial monosomy of the long arm of chromosome 16 in a live-born child. Karyotype: 46,XX,del(16)(q21).

Case Report

V.D.B. A. was the first child of healthy 23-year-old parents. Family history is negative with regard to congenital malformations, recurrent abortions, or mental retardation.

During the first two trimesters of pregnancy no problems were encountered. After 7.5 months, premature contractions occurred and the mother developed malleolar oedema. Delivery two weeks before term, was uneventful. Birthweight was 2550 g. Multiple congenital malformations were noted (Fig. 1). Craniofacial dysmorphism was present and characterized by a high forehead, a large anterior fontanelle, diastasis of all cranial sutures, a prominent metopic suture, and weakness of the cranial bones. The nasal bridge was broad and flat with marked hypertelorism (inner canthal distance: 2.6 cm; outer canthal distance: 6.4 cm). Other stigmata included slight mongoloid position of the eyes with small palpebral fissures and microphthalmia, severe micrognathism, low-set poorly lobulated small ears, and short neck. Abundant lanugo was present on the shoulders and in the neck. The thorax was narrow and small. A grade 3/6 holosystolic heart murmur was noted at the left sternal border. The anus was ectopic and situated in an abnormal anterior position, but otherwise completely separated from the vulva. The labia minora were hypoplastic. Bilateral simian lines were present on the hand palms. The fingers of both hands were in constant flexion. The child failed to thrive. At the age of 3 weeks staturoponderal measurements were: weight 2490 g, length 49 cm, and head circumference 34.3 cm. There was general hypotonia and oral feeding was not possible because of a feeble suck reflex. Moro’s reaction was incomplete. Muscle development was poor with diastasis of the abdominal muscles. A rockerbottom deformity was present on both feet and there was a
Fig. 1. Clinical photograph of the propositus at the age of 3 weeks

calcaneovarus deformity of the right foot with general malposition of all toes. The child suddenly died at the age of 1 month from aspiration pneumonia. A postmortem general X-ray survey was normal except that the proximal phalanges of the great toes were broad and stubby, and a marked diastasis of the cranial sutures was noted. At autopsy a complete malrotation of the intestinal tract was found. The presence of a large ventricle septum defect was confirmed. No other anomalies were found.

Cytogenetic Investigations

Using different banding techniques (G-, Q-, R-, C-banding), chromosome analysis of the propositus was performed on 93 peripheral blood lymphocytes and on 18 cells of a fibroblast culture of the right forearm. In all cells, except five in which there was random loss of one chromosome, 46 chromosomes were present. In all cells, a deletion of part of the long arm of one of the No. 16