Clinical Case Reports

"de novo" Trisomy 1q32 → 1qter and Monosomy 3p25 → 3pter

Emilio Yunis*, Hernán Egel, Ruth Zúñiga, Efraín Ramirez, Olga María Torres de Caballero, and Myriam Leibovici

Sección de Genética Humana, Departamento de Morfología, Facultad de Medicina, Universidad Nacional de Colombia, Bogotá, Colombia, SA

Summary. Minor abnormalities are described in an 11-month-old female in which a "de novo" trisomy 1q32 → 1qter and a monosomy 3p25 → 3pter has been produced. The amount of the exceeding material in this case is less than that found in previous reports of partial trisomy 1q and in cases of parental 1q balanced translocations which has originated recurrent abortions.

Chromosome rearrangements of the long arm of chromosome 1 have not been reported very often. Except for the reports concerning 1r chromosome, only 7 cases have been recorded, 3 of them corresponding to cases of parental balanced translocations originating extensive 1q trisomies (van den Berghe et al., 1973; Neu and Gardner, 1973; Norwood and Hoehn, 1974), and 4 to cases of parental balanced translocations as the cause of recurrent abortions (Friedrich and Nielsen, 1974; Rożynkowa et al., 1975). As a general feature, trisomy of the 1q segments involved has been deleterious or has been the cause of a severe malformation producing perinatal or neonatal death.

The following observation of a live-born infant represents the first example of partial trisomy 1q and terminal monosomy 3p with a picture of minor malformations. As the parental karyotypes were normal, this is also the first report of a "de novo" partial trisomy 1q and partial monosomy 3p.

Case Report

The proband is a one-year-old female born from a nonconsanguineous marriage; father 32-years-old and mother 24-years-old. Mother's first pregnancy ended with the birth of a healthy boy now 3½-years-old. Weight at birth was 2500g and size 48 cm. The pregnancy was full-term, threatened at 3 months by abortion which was controlled with rest.

At the time of the physical examination (11 months of age) it was found: size, 72 cm; weight, 7100 g; C. P. 44 cm; T. P. 42 cm; intercanthic distance, 34 mm.

* To whom offprint requests should be sent
Fig. 1. Physical appearance of the propositus

The skull showed hyperbrachycephaly, a wide fontanel, wide and prominent forehead and depressed temples (Fig. 1).

The face is flattened, round and wide with a prominent zygomatic bone; hypertelorism, epicanthic folds, blepharophimosis, depressed nasal bridge, broad nasal root, low-set ears, and carp-mouth are present. The neck is short with skin redundancy; there is umbilical hernia and diastasis of "rectum dominis." The fingers are long (Fig. 1), with clinodactyly of the 5th fingers. There is enlarged separation between the 1st and 2nd toes.

Dermatoglyphics are hypoplastic, showing bilateral simian creases and 7 whorls and 3 cubital loops in the fingertips.

X-ray analysis of skull, long bones, hands and feet are within normal limits. Bone age was reported as normal.

Cytogenetic Studies

Chromosome analysis of the proband and her parents were done by conventional as well as Q-, G-, and T-banding methods for identification of chromosome rearrangements suspected by the presence of an abnormal long chromosome 3 in the proband's Giemsa karyotype. The chromosome aberration, very well defined by G-banding (Fig. 2), consists of an abnormal extra segment identified as band 1q32→qter translocated to a 3p chromosome which is monosomic for 3p25→pter. Parent's karyotypes were normal.