Partial Trisomy 16q−

Emilio Yunis*, José T. González, and Olga M. Torres de Caballero

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

Summary. A 5-month-old female was found with a 16q− trisomy; her mother was the carrier of a balanced translocation 46,XX,t(15p+;16q−)(15p12;16q11). This is the first report in the literature of a liveborn with this chromosome abnormality.

Chromosome 16 has frequently been found in a trisomic state among miscarriages. We now report the case of a 5-month-old female with a 16q− trisomy; her mother being the carrier of a balanced translocation 46,XX,t(15p+;16q−).

Case Report

The proposita is the product of the third pregnancy of a nonconsanguineous marriage; the father's age was 25 and the mother's 19. Previous pregnancies ended, the first in a miscarriage at the 4th month and the second in a male fetus who died 3 h after birth. The patient was born

Fig. 1A and B. Facies of the patient at 5 months of age

* To whom offprint requests should be sent
after a normal and full pregnancy with birth weight 2700 g and length 47 cm. Physical examination at 5 months of age showed length 50 cm, a round head with a wide fontanel and a head circumference of 34 cm, a flattened face showing a high and arched palate, maxillary prominence, depressed nasal bridge, wide nasal root, and anteverted nostrils. There were narrow palpebral fissures, and the intercanthic distance was of 20 mm (Fig. 1). The ears were rounded and down-set with hypoplastic tragus and hypoplastic lower portion of the helix. The outer crus of the antihelix was absent (Fig. 1). At the abdominal level was found a diastasis of the rectus abdominis. On the right hand there was agenesis of the thumb and camptodactyly of the second finger. Abduction of the hip was limited. Hypoplastic dermatoglyphics showed bilateral simian creases.

Fig. 2 A and B. Partial karyotype of the patient's mother (A) showing by G-banding method the translocation 15p+;16q− and (B) partial karyotypes of the proposita showing by C-banding technique the partial trisomy 16q−.

Fig. 3. C-banded karyotype of the patient's mother; single arrows indicate the chromosomes involved in the translocation 15p+;16q−; double arrow shows the pericentric inversion in chromosome 9.