Trisomy 20pter→q11 in a Malformed Boy from a t(13;20)(p11;q11) Translocation-Carrier Mother

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Summary. A 3½-year-old boy revealed moderate motor and mental retardation, normal growth, a congenital heart defect and multiple minor dysmorphic signs and anomalies including brachycephaly, orbital hypotelorism, upward slanting palpebral fissures, short and beaked nose, full cheeks, malformed auricles, hypoplastic external genitalia, rocker-bottom feet with prominent heels, and various minor radiologic anomalies of bones. An extra chromosome in his karyotype appeared to represent trisomy of the short arm of chromosome 20 due to a maternally inherited balanced t(13;20)(p11;q11) translocation.

Case Report

The propositus was the full-term product of the second pregnancy of a healthy 22-year-old mother. Her first pregnancy had resulted in spontaneous abortion during the first trimester, with a fetus being recognized but not further examined. Pregnancy was normal, and delivery at the 39½th week was spontaneous, vertex, and uncomplicated. Apgar was 10 after 1 min. Birth weight 2500 g, length 48 cm, and head circumference 33 cm. The initial postnatal course was normal, but when the infant was about 8 days old, he developed peripheral cyanosis, poor sucking, and tachypnea, which increased. Auscultation revealed a systolic murmur over the entire precordium and a split second heart sound. Cardiac catheterization at age 2 weeks disclosed a ventricular septal defect, atrial septal defect, and patent ductus arteriosus with a considerable left-to-right shunt. The next day, cardiac operation was performed including ligation of the ductus and banding of the pulmonary artery. Several dysmorphic signs were noted (Fig. 1 a and b): a prominent occiput (by palpation the occipital bone was overriding the parietal bones); a peculiar face with receding forehead and chin, beaked nose, and misshapen ears; and unusually long and narrow feet. Muscle tone was decreased, and muscle mass was sparse. Bilateral inguinal hernias (indirect on the left and direct on the right) were repaired when he was 6 weeks old. At operation it was noted that both testes were present in the scrotum. Moderately limited extension in both knees and hips was also noted. When he was 6 months old, several brief episodes of stiffness, paleness, and unconsciousness occurred. When he was 2½ years old a second catheterization was performed because of deterioration of his cardiovascular status with an increasing incidence of sweating, tachypnea, and spells of cyanosis. The child also showed clubbed fingers and spoon-shaped nails. The examination revealed that the pulmonary band had migrated to the bifurcation of the pulmonary artery and that it was thus no longer effective. At a second operation, 2 months later, the septal defects were closed, an infundibular pulmonic stenosis was resected, the band removed, and both branches of the pulmonary artery (which on operation appeared to be congenitally hypoplastic) were reconstructed.

Clinical Examination at 2½ Years of Age

Clinical examination at 2½ years of age (Figs. 1 c and d) revealed a hypotrophic, anxious little boy with multiple minor dysmorphic signs, severe hypotonia, and profound mental and motor retardation. Height was 92.2 cm, weight was 13.1 kg (both 25th percentile), and occipitofrontal head circumference was 48.5 cm (3rd–10th percentile). Sitting height 55.3 cm, lower segment 36.8 cm, and their ratio 1.5 (97th percentile). The skull was brachycephalic with prominence of the parietal regions; the anterior fontanel was closed. His face was round due to his full cheeks and receding forehead and mandible. There were redundant skin folds over the middle of the forehead, but the metopic suture was not palpably prominent. The eyes appeared deep-set and close together, and the palpebral fissures were short and slanted upward. Inner canthal distance was 2.6 cm (50th percentile), outer canthal distance 7.2 cm (25th percentile), and lid length 2.4 cm (normal for his age: 2.5 cm). No epicanthus or strabismus was present; there was a stellate iris pattern with small peripheral radial white, Brushfield-like spots; the fundi appeared normal. His nose was small (length 3.8 cm), narrow, and beaked; his upper lip (length 1.1 cm) and mouth were small; and—secondary to his full cheeks—the nasolabial folds were prominent. The philtrum was well developed. All deciduous teeth were present and well aligned; tongue and palate were normal, but the uvula was bifid. The mandible was slightly receded. His ears were large (length 5.4 cm, 75th percentile), deep-set, posteriorly rotated, and malformed: the antihelices were peculiarly shaped with prominent...
Fig. 1a–d. Propositus at ages 7 weeks (a and b) and 2 years and 9 months (c and d) showing upward slanting palpebral fissures, beaked nose, small mouth, full cheeks, small mandible, short neck, posteriorly rotated and poorly formed ears, prominent occiput in (a), and flat occiput in (d)