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REFERENCES


Partial 9 trisomy by 3:1 segregation of balanced maternal translocation (7q+; 9q−)*

Summary. A male infant is presented with an extra derivative chromosome No. 9 resulting from a 3:1 meiotic segregation of a maternal balanced translocation involving the long arms of chromosomes No. 7 and 9. The patient is trisomic for the short arm and secondary constriction of the long arm of No. 9 and for the telomeric end of the long arm of No. 7. In addition to the features of the 9p trisomy syndrome he presents marked congenital myopia and extreme hypoplasia of the penis.

Trisomy for the short arm of chromosome No. 9 was first described by Rethoré et al (1970) and there are at present at least 11 reported cases in the literature (see review by Rethoré et al, 1973). Most cases were derived from parental balanced translocations and were trisomic for small segments of different chromosomes in addition to No. 9. The phenotypic spectrum of this syndrome includes microbrachicephaly, oblique antimongoloid palpebral fissures, ocular hypertelorism, malformed ears, palpebral hypoplasia, dermatoglyphic anomalies, and variable degrees of mental retardation.

In this report we present a male infant with trisomy for the short arm and secondary constriction region of the long arm of chromosome No. 9 and for the telomeric end of the long arm of chromosome No. 7 who, in addition to the phenotype of the 9p trisomy syndrome, has congenital myopia and genital hypoplasia.

Case report

The propositus is the first live born child of healthy and non-consanguineous parents. The mother was 25 years old and the father 26 years old at his birth. Three previous pregnancies ended in spontaneous abortions in the first trimester. Vaginal bleeding occurred during the second month of the propositus’ gestation. He was born at term, weight 3850 g, and required stimulation to initiate breathing; unilateral pneumothorax developed subsequently, but this resolved spontaneously. Early psychomotor development was delayed, while physical growth rate was normal. At 2 months of age weight was 6000 g, length 57 cm, and head circumference 40.5 cm. At 8 months of age weight was 10 100 g, length 71.9 cm, and head circumference 46 cm. At this time physical examination showed multiple anomalies (Figs. 1 and 2); acrocephaly, large anterior fontanelle, ocular hypertelorism, antimongoloid slant of the eyes, large low set ears with otherwise normal conformation, broad and bulbous nose, high arched palate, micrognathia, short and broad neck. Hands and fingers were short and broad, with bilateral clinodactyly of fifth fingers and hypoplastic nails. Cutaneous dimples were present in the external sides of both knees. The phallic was extremely small and the scrotum hypoplastic; testes were descended and small. Ocular examination revealed a severe myopia of 16°. Cardiovascular evaluation was normal. He had dysphonic voice and the psychomotor development was mildly retarded.

Radiology showed retarded bone age and hypoplastic

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Case reports

Dermatoglyphics

Right hand. Ulnar loops in the first, third, fourth, and fifth fingers; radial loop in the second finger. Simian line; axial triradius in r; terminates at 13°; atd angle: 43°. Digital triradii: a, medially displaced, terminates at 5'; b, hypoplastic, terminates at 7; c, forms a distal loop in the 3rd interdigital space and terminates at 9; d, medially displaced, terminates at 11. There are no thenar nor hypothenar patterns.

Left hand. Ulnar loops in all fingers. Simian line; axial triradius in r', terminates at 13°; atd angle: 45°. Digital triradii: a, medially displaced, terminates at 5'; b, hypoplastic, terminates at 7; c, forms a distal loop in 4th interdigital space and terminates at 7; d, medially displaced, terminates at 11. There is a whorl in the thenar area and no patterns in the hypothenar area.

Cytogenetics

Leucocyte culture from peripheral blood of the propositus showed a 47 chromosome count and XY constitution. Trypsin-Giemsa banding identified the extra chromosome as a deleted No. 9 with a break point at band 3 of region 1 of its long arm. The rest of the karyotype was normal (Fig. 3).

Chromosome analysis of the mother's blood with G-bandng (Fig. 4) revealed an apparently balanced translocation involving the long arms of chromosomes 7 and 9: t(7q+;9q-). The break points are located at the last distal band of the long arm of No. 7 (band 6 of region 3) and distal to the secondary constriction of the long arm of No. 9 (band 3 of region 1). The characteristics of the abnormal chromosome No. 9 suggest that the telomeric end of the long arm of No. 7 is reciprocally translocated; karyotype: 46,XX,t(7;9)(q36;q13). The karyotype of the propositus would therefore be as follows: 47,XY, + der(9),t(7;9)(q36;q13)mat, meaning that he is trisomic for the short arm and the secondary constriction region of the long arm of chromosome No. 9 as well as for band 6 of region 3 of the long arm of chromosome No. 7.

The father's karyotype was normal.

Discussion

There are at least 11 known cases with trisomy for the short arm of chromosome No. 9 (Rethoré et al,
1970; Cantu et al, 1971; Hoehn et al, 1971; Rott et al, 1971; Baccichetti and Tenconi, 1973; Rethoré et al, 1973). Four of these were in addition trisomics for variable segments of the long arm of chromosome No. 9 and for small segments of different other chromosomes (Rethoré et al, 1973).

The patient reported here is trisomic for the short arm and the secondary constriction region of the long arm of chromosome No. 9 and for the telomeric end of the long arm of chromosome No. 7, a combination not previously described. The anomaly resulted from a 3:1 segregation of a maternal reciprocal translocation involving the distal two thirds of the long arm of chromosome No. 9 and the telomeric end of the long arm of chromosome No. 7.

The phenotype of the propositus has many similarities with the recognized 9p trisomy, namely: antimongoloid slant of the eyes, ocular hypertelorism, large low set ears, broad and bulbous nose, hypoplastic phalanges, and delay in mental development. His dermatoglyphics are abnormal but he lacks the peculiar absence of fusion of digital tri-radii b and c, as reported in other cases. On the other hand he presents two previously unreported

**Fig. 3.** Karyotype of the propositus (trypsin-Giemsa banding) with the extra derivative chromosome No. 9 (arrow.)
findings: severe congenital myopia and hypoplastic genitalia. The observed phenotypic variation among reported cases of 9p trisomy may be due in part to the different additional trisomic segments other than the short arm of No. 9 chromosome, that were present in some patients.

The several possible kinds of meiotic segregation of the quadrivalent in the mother of our patient (adjacent-1, adjacent-2, and 3:1 disjunctions) may yield different types of unbalanced gametes with excess and/or deficiency of segments of chromosomes 7 and 9. The three previous spontaneous abortions in the sibship of the propositus suggest that the resulting embryos had nonviable aneuploid karyotypes.

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Pseudohermaphroditism with clinical features of trisomy 18 in an infant trisomic for parts of chromosomes 16 and 18: 47,XY,der(18),t(16;18)(p12;q11)mat

Summary. The case is presented of an infant who was diagnosed clinically as trisomy 18 with pseudohermaphroditism. Cytogenetic studies revealed an extra chromosome which represented a translocation chromosome derived from a balanced, reciprocal translocation between chromosomes 16 and 18: [der(18),t(16;18) (p12;q11)mat]. The infant's mother and a number of her relatives were found to be translocation carriers: [46,XX,t (16;18)(p12;q11)].

The trisomy 18 syndrome has a number of typical clinical features. Many organs are affected with a varying degree of frequency; the abnormalities have however been fairly constant in large series of cases (Warkany et al., 1966). A number of genital abnormalities have been described, but to our knowledge no case of pseudohermaphroditism has been previously described, nor can we find any previous reports of the trisomic chromosome being a translocation chromosome involving chromosomes 16 and 18.

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Case report

This was the first child of young parents, the mother being 26 years and the father 29 years. The mother had not had any previous miscarriages. The only drug taken during pregnancy was ferrous sulphate and she had not had any irradiation. Polyhydramnios was not recorded during the pregnancy. Oestriol levels were monitored from the 29th week of gestation because of small fetal size for period of gestation. The levels were below normal until the 34th week when the level was just within the normal range. Spontaneous premature labour occurred at 35 1/2 weeks. Delivery was uneventful and the baby's Apgar was 4 at 1 min and 8 at 4 min. Birth weight was 1700 g. The umbilical cord had one artery and one vein. The placenta weighed 370 g and was described as 'healthy'.

The baby had many of the characteristic features of trisomy 18 syndrome. The facial features were typical, including a prominent nose, depressed nasal bridge, and small palpebral fissures. There were no other ocular abnormalities. The mouth was small, the hard palate narrow and high-arched, and the soft palate cleft. The mandible was small and receding, and the alveoles low-set and malformed. The occiput was prominent and hair sparse. Other abnormalities included hypertonia with limited hip abduction, little subcutaneous tissue and loose folds of skin. The sternum was short and the nipples hypoplastic. The hands were clenched with the index overlying the third finger and the fifth finger overarching the fourth. The distal creases on the fifth fingers were absent; simian creases were not present. The feet showed the typical 'rockerbottom' deformity. Both the finger and toe nails were hypoplastic; the big toe short and dorsiflexed. The heart sounds were normal and there were no murmurs. Liver, spleen, and kidneys were not palpable abdominally. There were very large bilateral inguinal herniae which, when reduced, allowed palpation of a gonad in each hernial sac. The external genitalia appeared to be those of a female with clitoral hypertrophy, and only one orifice in the vestibule. No uterus could be palpated on rectal examination.

Under anaesthesia a more detailed examination of the genitalia was carried out. The perineal orifice was located between two poorly formed labia minora and behind a hypertrophied clitoris (Fig. 1a). A probe was inserted in the orifice and two tubular structures were entered close to the opening. Urethrocystoscopy showed the anterior structure to be a normal calibre urethra with no verumontanum. The posterior tubular structure, thought to represent a vagina, ended blindly, no cervix being visible. Contrast medium injected into the perineal orifice delineated the urethra and bladder, and demonstrated bilateral ureteric reflux; the ureters being moderately dilated. There was simultaneous filling of a blind cavity extending parallel and posterior to the urethra, to the base of the bladder (Fig. 1b). Bilateral inguinal herniorrhaphy and removal of the gonads was performed. A small midline abdominal incision was performed to examine the pelvic structures. Palpation failed to reveal uterus or ovaries. Histological examina-