De novo Trisomy 9pter–q13

Nataline B. Kardon 1 *, Helen R. Salwen 2, Marian A. Krauss 1, Jessica G. Davis 1, and Edmund C. Jenkins 2

1 Department of Pediatrics, North Shore University Hospital, 300 Community Drive, Manhasset, New York 11030, U.S.A., and Department of Pediatrics, Cornell University Medical College, U.S.A.
2 Department of Genetics, New York State Institute for Basic Research in Mental Retardation, 1050 Forest Hill Road, Staten Island, New York 10314, U.S.A.

Summary. A case of de novo trisomy 9p was observed. Cytogenetic analysis of G-, R-, Q-, and C-banded preparations revealed a karyotypic description of 47,XY,+del(9)(pter–q13). In addition to the principal characteristics of the 9p trisomy syndrome, the child presented with skeletal and urogenital abnormalities. It appears that certain clinical abnormalities are due to trisomy of 9q1.

Trisomy 9p has been well established as a syndrome. Approximately 50 cases (Lurie et al., 1976) have been reported since Rethoré’s initial observation (Rethoré et al., 1970). The syndrome consists of mental retardation, cranio-facial abnormalities (i.e., microcephaly, deep-set eyes, anti-mongoloid slanting of the eyes, bulbous nose, low-set and protruding ears), bony hypoplasias of the extremities, hypoplastic nails, and specific dermatoglyphic findings (absence or fusion of the b and c interdigital triradii).

Most cases have been diagnosed as either familial or sporadic chromosomal translocations. We here report a case of de novo trisomy 9p with a small portion of 9q. Only three other similar cases of de novo trisomies have been identified (Hoehn et al., 1971; Turleau et al., 1975; Kaosaar et al., 1976). The two more recent reports are distinguished from the present case by means of the marker chromosomal breakpoints.

Case Report

The patient (Fig. 1), an American of Italian extraction, was born on June 15, 1975, to a gravida 2, 27-year-old mother and 29-year-old father. A normal male sibling was born in 1972. Family history was unremarkable and there was no occurrence of spontaneous abortion. The pregnancy was uncomplicated and the infant was delivered at term. Birthweight was 2888 g.

* To whom offprint requests should be sent
The propositus was first seen in our center at 4 months of age. Physical examination revealed that his head circumference, height, and weight were 41.5 cm, 58 cm, and 6908 g, respectively. All of these values were at the 50th percentile. Pertinent findings included the following: Deep-set eyes with hypertelorism, low-set, predominant and rotated ears, a bulbous nose, a high-arched palate, and the corners of the mouth were down-turned (Fig. 1). He had hypoplastic finger nails, bilateral clinodactyly (fifth digit), finger contractures, bilateral equinovarus, overlapping toes, and a first degree hypospadias.

At 7 months, the patient did not exhibit any developmental delays. At the age of 15 months his psychomotor development corresponded to that of an 8-month-old child. He was able to finger feed, hold two objects, make monosyllabic sounds, and sit without support.

A skeletal survey revealed hypoplastic digits. Intravenous pyelogram and chest X-ray were normal.

The patient had bilateral simian creases. The axial triradii were distally placed at the t' position. The atd angle was 50°. There was also bilateral fusion of the b and c triradii. Dermal ridge patterns consisted of 9 arches and 1 ulnar loop with a total finger ridge count of five.

Cytogenetics

A modal chromosome number of 47 was determined from an examination of 124 metaphases derived from short-term leucocyte cultures. The same modality was confirmed in 50 mitotic figures from cultured skin. Thirty-six karyotypes prepared from G- (Klinger, 1972), R- (Hsu, 1976), or C- (Salamanca and Armendares, 1974) banded preparations showed that the extra chromosome was a No. 9, which lacked most of its long arm (Fig. 2).

Evidence for the marker chromosome or a balanced reciprocal translocation was not seen in any of the 150 cells that were studied in short-term leucocyte cultures of the mother, father, and normal sib. Analyses of 28 G-banded karyotypes—several from each individual—were normal.

Using C-bandng, the lengths of the 9qh regions were studied in 30 cells derived from the mother, father and propositus. In a majority of cells, the maternal C-banded region was ≥ 1½ times the length of 21q (Muller et al., 1976), while the homologues did not differ from each other by ≥ 1¼ times (McKenzie and Lubs, 1975). The paternal qh regions were usually about equal to 21q while the homologous C-bands differed from each other by a factor of ≥ 1¼ times.

A majority of the marker chromosome qh regions were ≥ 1½ times the length of 21q. About half of the homologous qh regions differed by a factor of ≥ 1¼