Familial Occurrence of 18q—

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Summary. An unusual segregation of the partial long arm deletion of a chromosome 18 is reported. This aberration was found in the feeble-minded mother and in her 4 daughters. The fifth child has XXY-Klinefelter’s syndrome. The carriers of 18q— in this family reveal small stature, microcephaly, and mental deficiency in the range from feeble-minded to severe imbecility. Other characteristic features commonly found in patients with 18q— syndrome, as mid-face retraction, downward slanting mouth, heart defect, and atretic ear canals were not observed.

In 1964, De Grouchy described the first case of a partial deletion of the long arm of the chromosome 18. At present time about 25 cases are known and this condition is considered a chromosomal syndrome. It is characterized by mental retardation, microcephaly and short stature. Very often hypertelorism, hypoplasia of the nasomaxillary region, downward slanting mouth, atresia of the ear canals, prominent helix and anti-helix, nystagmus or strabismus and excess of whorls on finger-tips are observed. The birth-weight tends to be low at term.

In this report we present a family in which the occurrence of 18q— in the mother and her 4 daughters was found and the only son had the XXY-Klinefelter’s syndrome.

Familial Data

As the pedigree (Fig. 1) shows, chromosomal and phenotypical abnormalities were found in the mother and in all 5 living children.

The mother’s (JN150333) mental level is of the feeble-minded degree. She is of short stature and her head circumference is 52 cm (Fig. 2). She does not reveal other features commonly found in the patients with 18q— syndrome.

III/1. The first child was a boy and he was the only one of all siblings who attended normal school. Unfortunately, he was victim of an accident, when he was at the age of 18.

III/2. The second pregnancy resulted in a delivery of a dead, mature fetus, probably due to a strangulation by an umbilical cord.

III/3. The result of the third conception is a boy (VN280954), who is now 16 years old. The cytogenetical studies performed throughout the family discovered the karyotype 47, XXY, and the Klinefelter’s syndrome has been later confirmed also by clinical examinations. He is 174 cm high and his mental level is in the degree of feeble minded. He attended the school for mentally subnormal children, but he was able to absolve only 5 of 9 classes.

III/4. The second living child (JN061156), who is the product of the fourth pregnancy is a girl of 13½. She was born at home and thus her birth-weight is only approximative—3000 g. She is 147 cm of height and her head circumference (48 cm) shows strong tendency to microcephaly. Her palate is high arched. She attends the school for mentally subnormal children.
The third of the living children is a girl of 11½ (MN 191 058). She lives in an asylum. Her birth-weight was 2200 g. Her height is below the average value for her age (130 cm) and her head circumference shows also tendency to microcephaly (50 cm). In the age of 1 year she suffered of meningoencephalitis purulenta. In the sacral region she has a naevus pilosus 2 × 2 cm large.

The two youngest girls are twins and they are living in an asylum. They are in the age of 7 years (Fig. 3).