117. An 18p- Case with Severe lower Abdominal Deformities

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Since the deletion of the short arm of chromosome no. 18 was described by de Grouchy *et al.* (1963), more than 80 cases have been referred to in the literature as the 18p- syndrome, and the *de novo* deletion at varying portions of the short arm of no. 18 was most common in them (de Grouchy 1969; Schinzel *et al.* 1974; Aksu *et al.* 1976; Rethoré 1977). The 18p- syndrome has been known to be associated with varying phenotypes (de Grouchy and Turleau 1977).

In the following is reported a female infant who is remarkable

Fig. 1. The proband on 3 days after birth, showing abdominal deformities. 1, umbilical cord. 2, omphalocele. 3, diastases of musculi recti abdomini. 4, urachal cyst. 5, anovestibular fistula with meconium.

Fig. 2. An X-ray photograph showing no shadow of left kidney. 1, right kidney. 2, urinary bladder.

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by clinical deformities such as diastases of muscoli recti abdomini, patent urachus, umbilical hernia, atresia ani, and defect of the left kidney, as well as by the chromosome abnormality showing the partial deletion of the short arm of no. 18.

Case reports. The propositus was a well-developed new female, born to a 30-year-old mother and a 30-year-old father as a third child. There was no history of spontaneous abortion, still birth, congenital malformation, exposure to atomic bomb, and consanguinity in this couple. Clinical examinations revealed that her parents and two elder sisters were phenotypically normal without evidence for mental deficiency. The birth weight of the propositus was 3400 g, with circumference of 35 cm in her head and 33 cm in her breast at 42 weeks of gestation.

Clinically, diastases of muscoli recti abdomini, patent urachus, umbilical hernia, anovestibular fistula combined with atresia ani (Fig. 1), and defects of the left kidney (Fig. 2) were especially evident in this child. Other abnormalities observed involved horizontal palpebral fissures, short upper lip, low-set ears, short neck, widespread nipples, and broad and short hands.

Cytogenetical findings. Chromosome slides were prepared on the basis of leucocyte cultures from the propositus, her two elder sisters, her mother and her father. G-banding differential staining

Fig. 3. G-banding karyotype of the proband. Arrow indicates chromosome 18 with partial deletion of the short arm.

(Seabright 1971) was applied to air-dried chromosome slides. Chromosome counts were made on 50 well-delineated metaphases. Karyotype analyses were made on 20 cells each from conventional and G-banding slides prepared from the propositus, two elder sisters, her mother and father.

The karyotype of the proband based on the conventional Giemsa specimen contained 46 chromosome which included a chromosome no. 18 with an unusually short arm. On the basis of differential staining, the conclusion became possible that a chromosome 18 with an abnormal short arm resulted from the partial deletion involving about one half of the short arm: 46,XX,del(18) (p1105) (Fig. 3).

Her two elder sisters and parents showed no sign of chromosome change, each having an apparently normal karyotype (Fig. 4).

Remarks and conclusion. It is interest to see that the correlation seems to be present between the variation in the break point of the short arm of chromosome 18 and the clinical signs. It is likely that one might assume a rather severe effect on phenotype due to the

Fig. 4. Partial karyotypes of the proband and her family members. 1, proband. 2, father. 3, mother. 4, eldest sister. 5, elder sister.

complete absence of the short arm of chromosome 18. But, considerably severe clinical features have been observed in patients having a partial deletion of the short arm of no. 18 (Faust *et al.* 1976). The situation is suggestive of that the etiologically important portion may be limited to a small terminal segment of the short arm of no. 18 (Funderburk *et al.* 1977). The literature pointed to the fact that the patients with the 18p- chromosome constitution showed abnormal phenotypes of varying degrees. These variable phenotypes might be responsible for the pseudodominance effect of certain recessive mutant genes located in the hemizygous state on the short arm of chromosome 18 (Uchida *et al.* 1965), and/or accounted for by the hypothesis of gene-dose compensation (Faust *et al.* 1976).

In reference to the above pictures, the present 18p- case is interesting in showing severe lower abdominal deformities together with a few other minor abnormalities. But, there was in this patient no sign for short stature, microcephaly, round and flat face, broad-based nose, hypertelorism, epicanthus, ptosis of eyelids and strabismus, carp mouth, cleft palate, microretrognathia, auricular deformities, webbed neck, low hair line, muscular hypotonia, clinodactyly of the fifth digit, and toes and feet deformity, though they were commonly found in the 18p- syndrome.

Summary. A female newborn infant having severe lower abdominal deformities is described. She has a 46,XX,del(18)(p1105) karyotype based on G-banding analysis. Her parents and two sisters are chromosomally and clinically normal.

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