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# 9p deletion syndrome

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A detailed description of a male patient with deletion of the short arm of chromosome 9 is presented. The most striking finding was that both the phenotype and the behaviour of the patient gave the impression of Down syndrome both after birth and at 3 years of age.

The delineation of the syndrome of 9p monosomy was first given by Alfi et al. (1). Afterwards, several cases were reported in the literature. To our best knowledge, their number now amounts to about 30 (2–8, 10–20). A further case will be reported below. The aim of this report is to point to the similarity between the phenotype of 9p monosomy and that of Down syndrome.

## Case Report

F. A., a male patient was born when his mother was 24 years old, the father 25 years old. This was the first pregnancy of the couple. The parents were unrelated and healthy. In the family history there was no event worth mentioning.

After uneventful pregnancy, delivery occurred without complication at the 36th week of gestation. The 1 min Apgar value was 5. The proband did not thrive, his hands and feet were swollen. Birth weight was 3000 g, body length, 51 cm, and the head circumference was 33 cm.

After birth the proband was registered as having Down syndrome. The diagnosis was based on the following clinical features: microcephaly with flattened occiput, ptosis of the upper eyelids, epicanthus, upward and outward slanting palpebral fissures, small nose with anteverted nostrils, small mouth, slight micrognathia, low-set and adherent ears, hypotonicity of the muscles, umbilical hernia and small pelvis (Fig. 1). The only symptom not corresponding to Down syndrome was the size of the hands and feet and the fingers (Fig. 2). In spite of lymphoedema, the hands did not seem broad and the fingers were thin and long. Besides, a protruding frontal bossing, long philtrum, short neck and wide-spaced nipples could be observed. He had also hypospadias.

At 8 months of age when the patient was examined, his weight was 7500 g (25 percentile), length was 70 cm (50 percentile), head circumference was 41 cm (under 3rd percentile). At 20 months of age the psychomotor retardation was expressed. At 3 years, his behaviour resembled that of Down patients, he was amiable and affectionate (Fig. 3).

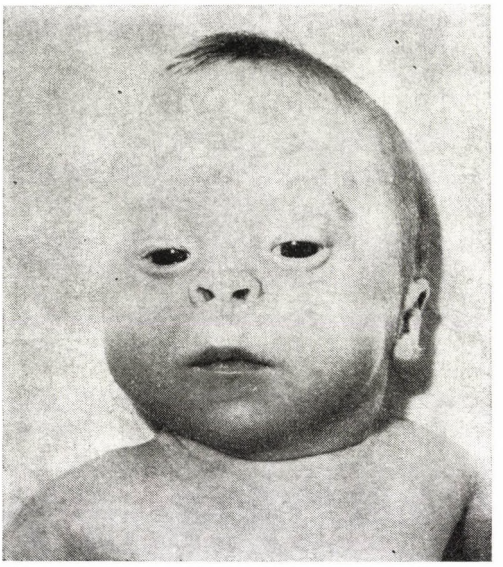

FIG. 1. The patient at 3 months of age

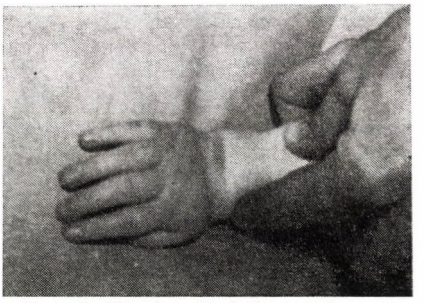

FIG. 2. The hand of the patient

*Cytogenetic findings*

In routinely cultivated blood cells after GAG staining deletion of the short arm of chromosome 9 was found. The breakpoint was identified as p22. The karyotype of the patient is thus 46, XY, del(9) (p22) (Fig. 4).

Both parents were normal cytogenetically.

## DISCUSSION

Partial monosomy of the short arm of chromosome 9 has mostly been observed as a de novo aberration. Parental translocations were found responsible in a few cases (2, 3, 15, 17). An analysis of the breakpoints in 24 reported cases (Fig. 5) showed that the band p22 has mostly been involved; in 18 cases the breakage occurred at this site.

The clinical characteristics of 9p deletion were the subject of several papers (1, 5, 9, 20). The characteristic cranio-facial dysmorphism has been described to consist of trigonocephaly with protruding forehead, mongoloid palpebral fissures, epicanthus, flat and wide bridge of nose, short nose

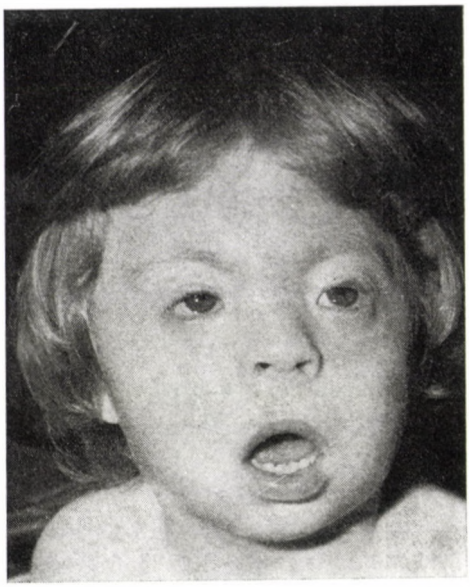

FIG. 3. The patient at 3 years of age

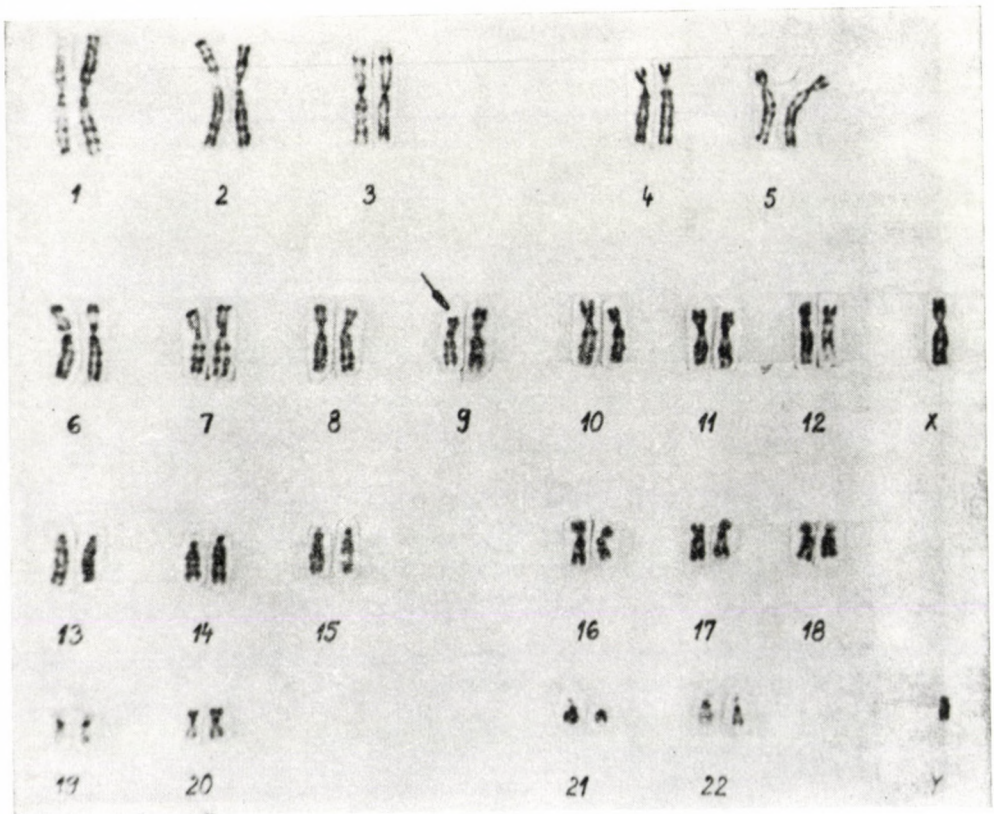


FIG. 4. The karyotype with the deletion of 9p

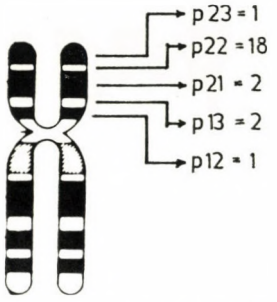


FIG. 5. Distribution of breakpoints in 24 cases of 9p— syndrome

with up-turned nostrils, small mouth, slight micrognathia, low-set and adherent ears. The neck is short, sometimes with more or less pronounced pterygium colli. The fingers and toes are long. The I. Q. is 30–60. Life expectancy does not seem to be diminished significantly.

Listing the main specific symptoms, the likeness between 9p— syndrome and Down syndrome is striking (Table I). There is no other chromosome

TABLE I

Comparison of the clinical symptoms in patients with 9p— and 21-trisomy syndromes

| Clinical symptoms | 9p— | 21-trisomy |
|---|---|---|
| Microcephaly | + | + |
| Trigonomicrocephaly | + | − |
| Flat occiput | + | + |
| Short neck | + | + |
| Protruding forehead | + | + |
| Upward and outward slanting palpebral fissures | + | + |
| Epicanthus | + | + |
| Flat nasal bridge | + | + |
| Short nose with anteverted nostrils | + | + |
| Small mouth | + | + |
| Protruding tongue | − | + |
| Small, adherent ears | + | + |
| Broad hands with short fingers | − | + |
| Long fingers | + | − |
| Hernia | + | + |
| Other malformations | − | + |
| Hypotonicity | + | + |
| Mental retardation | + | + |

aberration where the phenotype would remind of the appearance of patients with 21-trisomy to such a degree. Even our patient's behaviour resembled that of Down patients. In clinical differential diagnostics this has no practical importance, since 9p deletion is a much less frequent chromosome aberration than 21-trisomy. Studies of the molecular basis of the likeness of these two chromosomal syndromes would be more interesting.

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