The Aarskog Syndrome

J. P. Fryns, J. Macken*, L. Vinken**, L. Igodt-Ameye*, and H. van den Berghe***

Division of Human Genetics, Department of Human Biology, Minderbroedersstraat, 12, B-3000 Leuven, Belgium

Summary. In this report a description is given of the Aarskog syndrome in six males belonging to three different families. Partial expression of the syndrome was confirmed in two of the three examined obligate female heterozygotes, who had short stature, small hands and feet, short neck, and a round face with widow's peak and, in one of them, ptosis of the eyelids.

Introduction

After the original descriptions of the facial-digital-genital syndrome by Aarskog (1970) and Scott (1971), subsequent reports by others have clearly delineated this malformative syndrome (Berman et al., 1974; Furukawa et al., 1972; Melnick et al., 1976; Scott, 1971; Sugarman et al., 1973). In this report six males are described who belong to three different families and exhibit the peculiar stigmata of the syndrome. The partial expression of the syndrome in female relatives is further documented.

Case Reports

Family S

S. D. was the first child of unrelated parents. At birth the father was 24 years and the mother 19 years old. Pregnancy and delivery, at term, were normal. Birth weight was 2400 g (third percentile: 2630 g), length and head circumference were not noted. No major difficulties occurred within the first two months of life. During the next few months, until the age of 14 months, several different infections necessitated almost continuous hospitalization. Failure to thrive was striking, psychomotor development was retarded, and severe hyperkinetism with episodes of aggressiveness became evident. At the age of four years a left-side cryptorchidy was surgically...
corrected. Because of manifest growth retardation and permanent character problems, a general medicopsychological evaluation was made at the age of 5 years. Statureponderal measurements were: length 96.5 cm (third percentile: 111.5 cm), weight 15.35 kg (third percentile: 19.4 kg) and head circumference 51 cm.

Biochemical screening was negative, including amino-acid chromatography and thyroid function. Chromosomal analysis showed a normal male karyotype. The bone age, on an X-ray of the left wrist, was slightly retarded, and hypoplasia of the third and fourth cervical vertebral bodies was noted. Ophthalmologic examination was normal. On psychological evaluation the IQ was 64. Now, at the age of 10 years, psychorelational problems have persisted. Length was 123.5 cm (2.5 percentile: 127.4 cm), weight 22.5 kg (third percentile), and head circumference 53 cm (50th percentile). Craniofacial dysmorphism was obvious (Fig. 1): round face with large forehead, hypertelorism with broad base of a small, somewhat upturned nose, short upper lip, and low-set ears. The neck was short and triangular, and the posterior hairline was low-implanted. Hands and feet were short, with bilateral cutaneous syndactyly between fingers II and III, and with hyperextensibility of the proximal interphalangeal joints and broad-ending terminal phalanges. A large gap was present between the first and second toes. The penis was normal; the right testis was of normal size, whereas the left was atrophic. The left part of the scrotum was hypoplastic and the scrotal folds extended ventrally surrounding the penis (Fig. 2).

The mother of the propositus (Fig. 3) had a short stature (151 cm) and presented a number of distinct craniofacial stigmata: bilateral ptosis of the eyelids, broad forehead with widow's peak, hypertelorism, upturned somewhat stubby nose, and short neck. Hands and feet were small with discrete interphalangeal webbing. She was born after a 34-week pregnancy. Birth weight was 1200 g. Mental development and performance were normal. Her two brothers were pheno-