Short Report

Aarskog syndrome associated with hypermetropia and toe anomaly

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Aarskog syndrome is characterised by a disproportionately short stature and facial, skeletal and urogenital anomalies ('shawl' scrotum and cryptorchidism). Ophthalmic findings include a slight downward slant to the palpebral fissures, hypertelorism, blepharoptosis, strabismus, ophthalmoplegia, hypermetropic astigmatism and a large cornea. Findings on the extremities include joint hyperextensibility, short and broad hands, interdigital webbing, a short fifth finger, clinodactyly and broad feet with bulbous toes. We report on a 7½-year-old boy with typical findings of Aarskog syndrome, hypermetropia and bilateral proximal implantation of the fifth toes. These associated abnormalities have hitherto never been described, to our knowledge.

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Aarskog syndrome (AS), also known as faciogenital dysplasia, is characterised by disproportionately short stature, and facial, skeletal and urogenital anomalies.[1-4] It is an X-linked developmental disorder and molecular genetic analyses mapped the disease locus to chromosome Xp11.21.[2,5] Pasteris *et al.*[2] demonstrated that an isolated cDNA, FGD1, was disrupted by the breakpoint, and that FGD1 mutations characterise the disease. In addition, they suggested that FGD1 is a Rho/RacGEF involved in mammalian development and that FGD1 was responsible for faciogenital dysplasia. The incidence of the syndrome is unknown. However, it is probably underestimated because of its mild phenotypical appearance.[6,7] We encountered a boy with the syndrome, who had associated hypermetropia and proximal implantation of the fifth toes. To our knowledge, these associated abnormalities have hitherto never been described.

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Case report

A 7½-year-old boy was admitted to our hospital with undescended testes. Orchiopexy had been performed when he was 3 years old. In addition, he had used phenobarbital for epilepsy between the ages of 3 and 6 years. His family history was unremarkable.

Physical examination revealed that his weight was 21 kg (25th - 50th percentile) and his height 114 cm (3rd - 10th percentile). His face was overly round. He had a broad philtrum, and a crease below the lower lip, his lower anterior teeth were irregular and the auricular lobules were fleshy. On ophthalmological examination, bilateral hypermetropia (+6.0 dioptre) and hypertelorism with an intercanthal distance of 32 mm and interpupillary distance of 55 mm were diagnosed (Fig. 1). Examinations of the fundus and the anterior segment of the eyes were normal and he had no strabismus. He had mild pectus excavatum, and the umbilicus was a flat configuration. On examination of the genito-urinary system, a scar on the scrotum from a previous orchiopexy and 'shawl' scrotum were noted, but the testes were in the scrotum (Fig. 2). On examination of

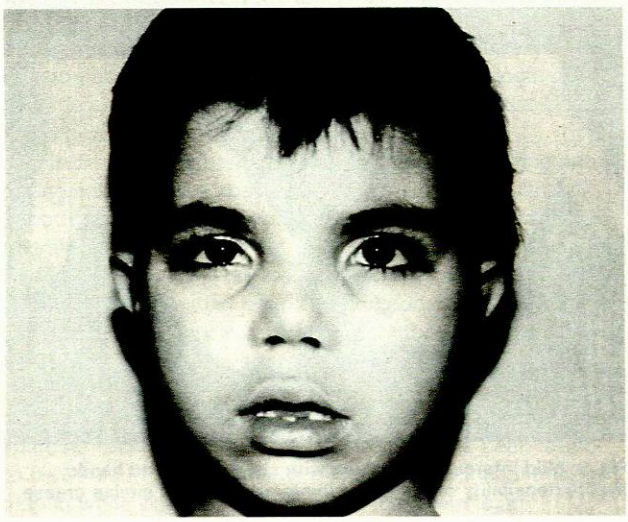

Fig. 1. Rounded face, hypertelorism and broad philtrum shown.

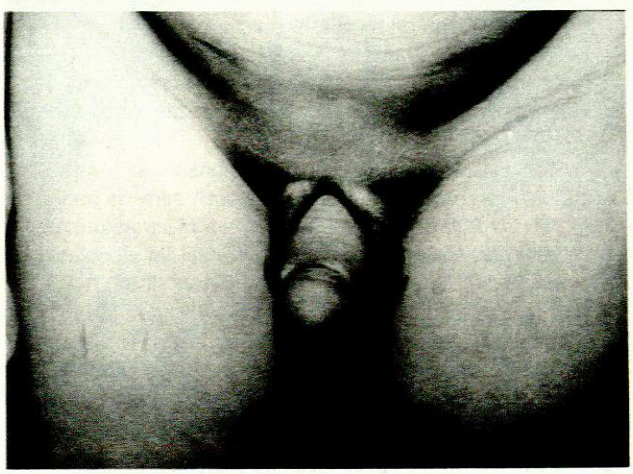

Fig. 2. 'Shawl' scrotum shown in our patient.

the extremities mild interdigital webbing with short and broad hands, hyperextensibility of the finger joints, bilateral simian crease, broad feet with bulbous toes, and bilateral proximal implantation of the fifth toes were seen (Figs 3 and 4). Laboratory investigations, including routine urine and blood analyses, and electrocardiographic and echocardiographic examinations were normal. Chromosome analysis revealed 46, XY.

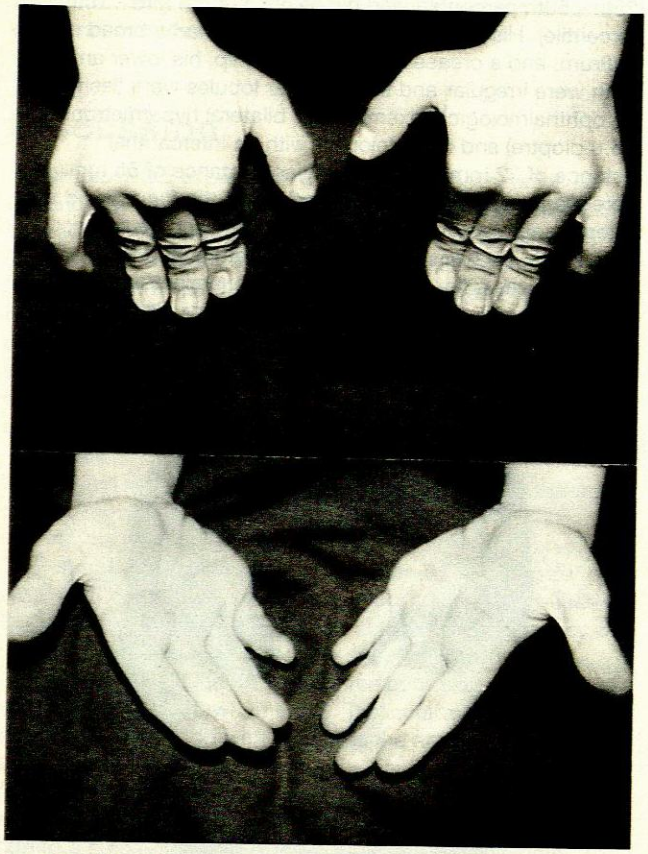

Fig. 3. Mild interdigital webbing with short and broad hands, hyperextensibility of the finger joints and bilateral simian crease.

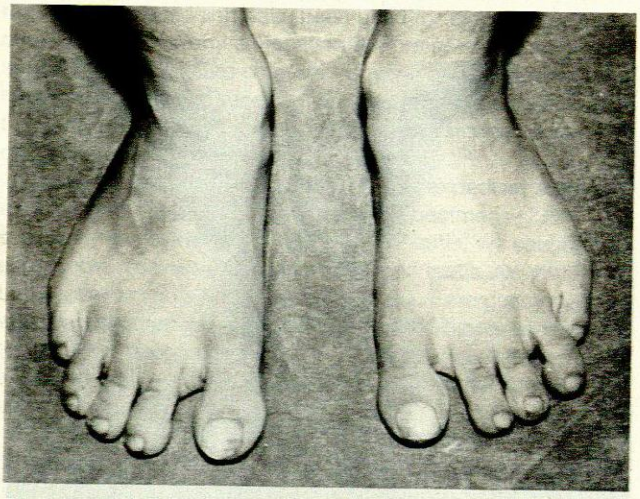

Fig. 4. Broad feet with bulbous toes and bilateral proximal implantation of the fifth toes.

Discussion

Teebi *et al.*[8] reported five individuals with AS in one family. Based on analysis of this family and others from the literature they derived primary and secondary diagnostic criteria for AS. Primary criteria include short stature, hypertelorism, short nose with anteverted nares, maxillary hypoplasia, a crease below the lower lip, mild interdigital webbing with short and broad hands, a short fifth finger with clinodactyly and shawl scrotum. Secondary criteria include abnormal auricles with fleshy lobules, posteriorly angulated ears, widow's peak, ptosis, a downward slant to the palpebral fissures, joint hyperextensibility, broad feet with bulbous toes, cryptorchidism, inguinal hernia and prominent umbilicus. Other anomalies associated with the syndrome include a rounded face, small and/or broad nose, long and/or broad philtrum, low-set auricle, hypodontia, retarded dental eruption, orthodontic problems, prominent metopic suture, pectus excavatum, cervical vertebral anomalies, scoliosis, cubitus valgus, metatarsus adductus, cleft scrotum, phimosis and simian crease.[4,7] Our patient had almost all of these manifestations and fitted the criteria. Aside from these findings, he had bilateral proximal implantation of the fifth toes, which has not previously been reported in association with AS.

Ophthalmic findings in AS include a slight downward slant to palpebral fissures, hypertelorism, blepharoptosis, strabismus, and ophthalmoplegia.[3,4,7] In addition, bilateral retinal vessel tortuosity, Brown's syndrome, hypermetropic astigmatism and a large cornea were reported in association with the syndrome.[3,9,10] Our patient had hypertelorism and bilateral hypermetropia; the latter anomaly's association with AS has not previously been published.

In conclusion, we want to stress that hypermetropia and bilateral proximal implantation of a fifth toe could be associated with AS, and that all patients with AS should be investigated for these anomalies.

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