SHORT REPORTS

Atypical case of Aarskog syndrome

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At present, around 100 males with Aarskog syndrome from different ethnic groups have been reported.[1-3] The main clinical features of this syndrome are peculiar facies, saddle bag (or 'shawl') scrotum, and abnormalities of the hands and feet. It is presumed to be an X linked recessive disorder, although autosomal dominant inheritance has also been suggested.[2] The present report describes the first Estonian case.

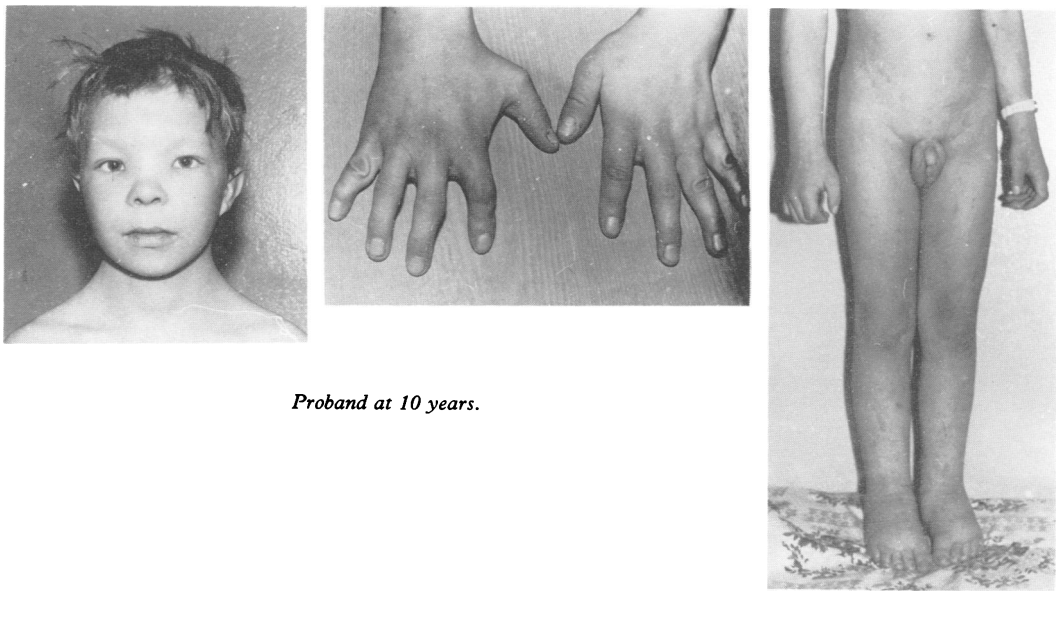
The proband, a boy, was the result of the first pregnancy of unrelated, healthy parents; the mother was 25 and the father 28 years old at the time of his birth. Pregnancy and delivery (two weeks after term) were uncomplicated, birth weight was 2950 g, and length 45 cm.

Examination at the age of 10 years (figure, table) showed a boy with short stature: height 125 cm, weight 51 kg, and head circumference 51 cm. His psychomotor and mental development were normal. He had hypertelorism, a flat nasal bridge, upward slanting palpebral fissures, epicanthus, and a short neck. The fingers were short with camptoclinodactyly. He had a thoracic deformity and kyphoscoliosis. The toes were short and turned medially. He had painless lymphoedema of his feet extending up to his knees. There were no heart or vascular abnormalities. The genitalia were enlarged and the scrotum encircled the base of the penis ('shawl scrotum').

Radiographs showed shortened fifth fingers, the third to fifth fingers deviated towards the second finger, lumbokyphoscoliosis, and pes planovalgus. Serum levels of total protein, thyroxin, TSH, and growth hormone were normal, but the prolactin level was slightly increased (455 nmol/l). Chromosome analysis showed a normal male karyotype and the mother's karyotype was also normal.

The face of our proband showed dysmorphic features described in both Aarskog syndrome and Noonan syndrome. However, short stature, normal intellect, 'shawl scrotum', camptodactyly, and the absence of heart pathology are more typical of Aarskog syndrome and only lymphoedema is characteristic of Noonan syndrome.

Investigation of the proband's family showed that his mother and father were healthy and normal. The mother's height was 162 cm with no signs of Aarskog syndrome or lymphoedema. Neither the proband nor his mother has sibs. Investigation (together with photographs) of the other members of the family showed the same atypical variant of Aarskog syndrome in the maternal grandfather who had normal intellect, hypertelorism, camptodactyly, and lymphoedema of the feet. His mother, father, and two brothers were healthy and had normal intellect.



Proband at 10 years.

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Frequency of symptoms (%) in Aarskog syndrome.

	Grier *et al*[2]	Our proband
Short stature	88	+
Hypertelorism	87	+
Upward slanting palpebral fissures	69	–
Broad, flat nasal bridge	87	+
Anteverted nostrils	87	+
Long philtrum	84	–
Low set, malformed ears	60	–
High arched palate	+	–
Short neck	+	+
Thoracic abnormalities (pectus excavatum)	57	+
Limb abnormalities		
Short fingers and toes	82	+
Syndactyly	63	+
Clinodactyly	68	+
Simian creases	70	–
Shawl scrotum	81	+
Normal intellect	88	+
Heart defect		–
Lymphoedema		+

There are some reports on the association of Aarskog syndrome with vascular pathology,[3] but no case of Aarskog syndrome with lymphatic vessel pathology has been reported. In our case, family investigation showed that the association of Aarskog syndrome and lymphoedema is unlikely to be coincidental but that they are linked through a common genetic basis on the X chromosome. It is also possible that we could be dealing with a new (X linked recessive) syndrome.

1 Aarskog D. A familial syndrome of short stature associated with facial dysplasia and genital anomalies. *J Pediatr* 1970;77:856–61.
2 Grier RE, Farrington FH, Kendig R, Mamunes P. Autosomal dominant inheritance of the Aarskog syndrome. *Am J Med Genet* 1983;15:39–46.
3 van den Bergh P, Fryns JP, Wilms G, Piot R, Dralands G, van den Bergh R. Anomalous cerebral venous drainage in Aarskog syndrome. *Clin Genet* 1984;25:288–94.