Hum. Genet. 39, 113—116 (1977)

Human Genetics

Lymphedema – Distichiasis

A Rare Hereditary Syndrome

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Summary. The syndrome of familial lymphedema (type Meige) with distichiasis was observed in father and son. The association with uvula bifida and submucous cleft of the palate is described for the first time.

The association of congenital distichiasis with lymphostasis has been seldom observed. The first description was given by Campbell (1945). In connection with this syndrome, pterygium colli, ectropion of the lower eye-lid, spinal extradural cysts, and fusions of various thoracic vertebrae have been described. Uvula bifida and submucous palatoschisis in connection with this syndrome is reported here for the first time.

History

An 18-year-old male patient M. K., the only child of parents who are not related, showed distichisis of both upper eye-lids since his early childhood (Fig. 1). Because of nasal speech, his adenoids were removed. No serious diseases were noticed. Since the age of 16, edema of both legs began to develop (Fig. 2).

Clinical Findings

Height: 183 cm, weight: 89.2 kg; patient in good general condition. A further check-up showed myopia, anisometropia, uvula bifida, submucous palatoschisis, and an extensive edema of both legs with the first signs of a varicose ulcer on the medial part of the right ankle. We found congestive eczemas, single hyperpigmentation, and outstanding vein marks on both shanks. The finger joints bones could be superextended beyond average. All other organs were found normal.

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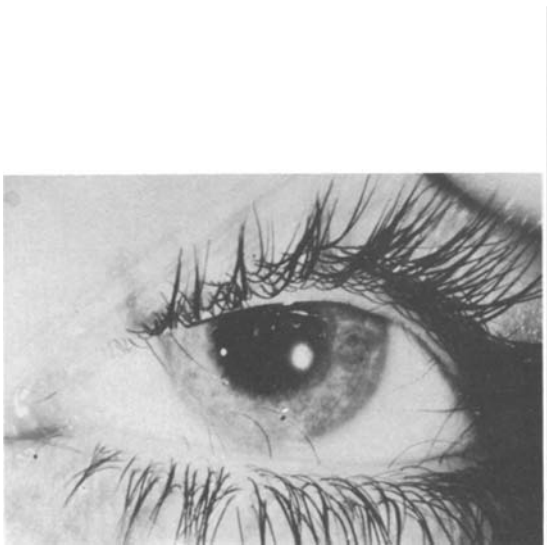

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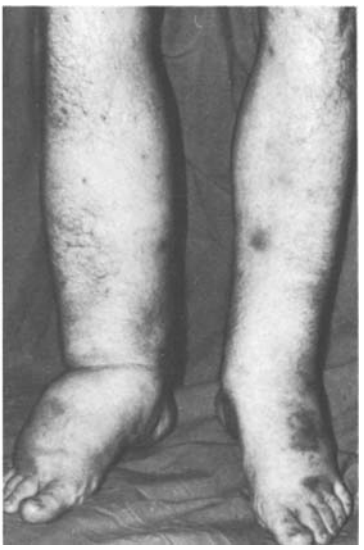

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Fig. 1. Patient M. K. with distichiasis of both upper eye-lids

Fig. 2. Patient M. K. with lymphedema of both legs

Laboratory Findings

Normal.

Lymphography (Fig. 3) shows spiral, ectatic lymphatic vessels with a slight collateral circulation on both shanks, and a reflow of contrast medium over the talocalcanean joint to the planta of the foot. The lymphatic vessels of the thighs and pelvis seem to be normal.

Phlebography (Fig. 3) shows insufficient venae perforantes with an outflow of contrast medium through the great saphenal vein. In addition to varicosis, phlebectasias on both calves can be seen. The vena poplitea and vena femoralis seem to be normal.

The examination of chromosomes gave normal results[1].

After intensive therapy with diuretics and Jobst-Massages (intermitting subdiastolic compressions with cuffs in time-intervals increasing) the edema of both legs disappeared.

The father of the patient, 53-year-old W. K., also shows distichiasis of both upper eye-lids, which was discovered in his early childhood too, and which had been treated with plastic surgery—using the mucous membrane of the lips—and epilation. In addition, he also has an uvula bifida. Submucous palatoschisis cannot be stated with certainty. We found an edema on both shanks, which was known since the age of 20. On the medial side of his left ankle there are remnants of repeated varicose ulcers. Furthermore, we noticed varicosis of both shanks and hyperpigmentation on the medial side of both shanks.

Repeated therapy of the varicose ulcers was not successful. The father of the patient was not willing to undergo diagnostic procedures.

Discussion

Campbell (1949) first described a family of which, in four generations, seven members suffered from primary edema, type Meige, and often from distichiasis as well.

[1] I wish to thank Dr. S. Flatz, Institute of Genetics, for examination of chromosomes

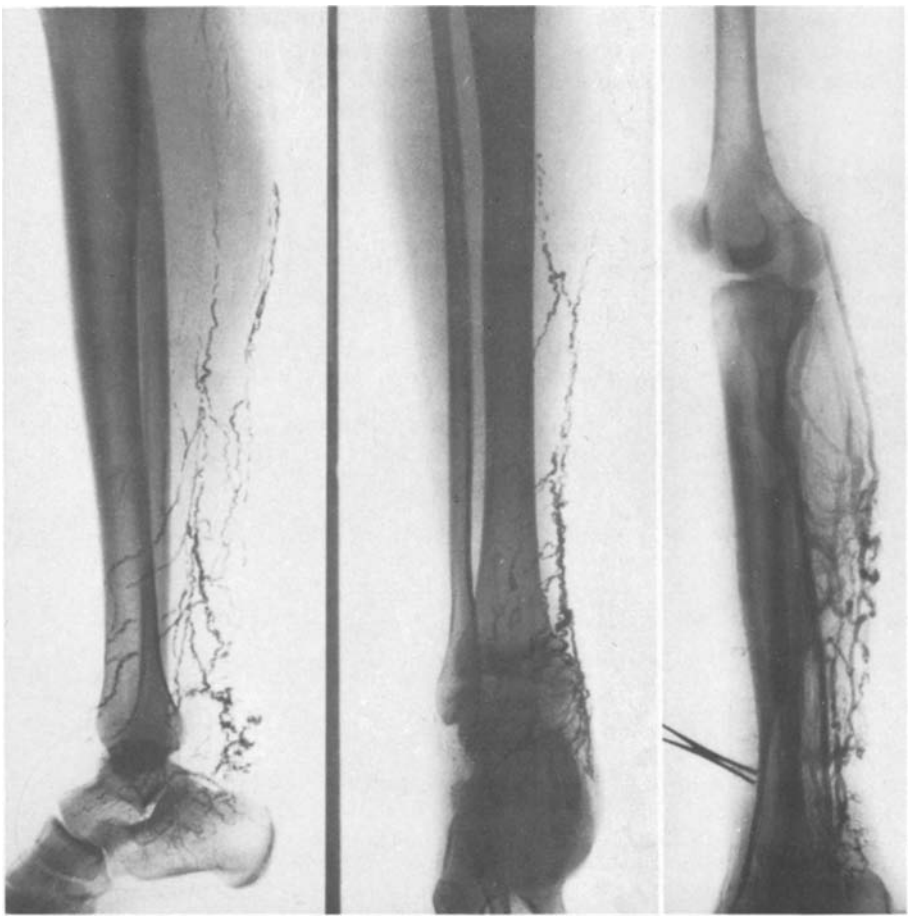

Fig. 3. Patient M. K., Lymphography of the leg with ectatic lymphatic vessels (*left, middle*). Phlebography of the same leg with varicosis, phlebectasias and insufficient venae perforantes (*right*)

Falls and Kertesz (1964) observed a family of which ten members in five generations had suffered from edema of the legs during puberty. Four out of five siblings, the father, and his brother showed distichiasis. In addition, some members of this family had pterygium colli and ectropion of the lower eye-lid.

Bergland (1968) described this syndrome in three members of another family, who, in addition, suffered from spinal extradural cysts.

Robinow et al. (1970) reported on a further deformation; apart from distichiasis and edema of the shanks a father and his son suffered from fusion of various thoracic vertebrae. A daughter had only distichiasis.

The few reports on this rare syndrome reveal that besides distichiasis and edema of the shanks (type Meige) other malformations can occur such as; pterygium colli, cuneiform vertebra, and uvula bifida in connection with submucous palatoschisis.

On the basis of our own and published observations, we suppose that this syndrome is due to a specific autosomal-dominant gene with variable manifestation of additional malformations.

References

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Received February 10, 1977 / June 27, 1977