PES CAVUS AND LYMPHOEDEMA
An Unusual Familial Syndrome

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Pes cavus may occur sporadically and without apparent cause. In some patients it may be associated with other conditions. These may be acquired, such as poliomyelitis or peripheral nerve injury, or hereditary. Examples of the latter are Friedreich's ataxia and spina bifida. In this paper we record two patients both of whom had bilateral pes cavus and lymphoedema. We know of no previous report of such an association.

CASE REPORTS
Case 1—A girl aged seventeen, a pastry cook, presented with a history of constant swelling of the right foot and lower leg since birth. The swelling had increased considerably during the previous six months, during which time there had been pain and discomfort in the foot and leg. The left foot sometimes became swollen in the evenings, especially after exercise. The patient was born with idiopathic bilateral pes cavus and at the age of six months a corrective operation had been performed on the right foot at another hospital. Her general health was good. There was a family history of both pes cavus and lymphoedema. Figure 1 shows that the patient's mother, maternal grandfather and uncle had right-sided pes cavus and that an aunt and a cousin had a combination of bilateral pes cavus and lymphoedema.

![Family tree of Case 1.](image)

Physical examination confirmed bilateral pes cavus, the right foot being more severely affected, and pitting oedema of the right lower leg and ankle (Figs. 2 and 3).

Bilateral lymphography was performed by techniques that have been described elsewhere (Kinmonth, Taylor and Harper 1955; Gough, Guiney and Kinmonth 1963). There were no patent lymphatics on the dorsum of either foot. Patent blue was injected intradermally below the knee on each side. A lymphatic was then found through an incision at knee level and injected with ultrafluid lipiodol (Bengué), 7 millilitres on the right and 8 millilitres on the left. Only one trunk filled on either side up to about seven centimetres from the groin where only about five afferent lymphatics were seen, the lowest number that might be considered normal. From the groin upwards lymphography was normal.

This investigation confirmed the diagnosis of congenital lymphoedema with a deficient number of lymphatic vessels in both legs—the hypoplastic variety of lymphoedema (Figs. 4 and 5). There was no dermal backflow of dye in this patient. Conservative treatment was recommended.

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FIG. 2

Case 1. Figure 4—Lymphangiogram of right thigh. Normally five or more distinct trunks would be seen. Here a single large trunk breaks up into several small channels which later unite. A variety of "numerical hypoplasia". Figure 5—Lymphangiogram of left thigh. There is only one lymph trunk visualised up to the upper third of the thigh ("solitary hypoplasia"). Normally five or more would be seen.
Case 2—A married woman aged forty-three presented with a thirteen-year history of intermittent mild bilateral swelling of feet, ankles and lower legs. The swelling was worse after prolonged standing. Treatment with elastic stockings and diuretics had not improved her disability. She was born with idiopathic pes cavus and said that her sister also had bilateral pes cavus. There was no family history of lymphoedema. The patient was not a good historian, so that a family tree could not be constructed.

Physical examination confirmed bilateral pes cavus and mild swelling of both legs.

Bilateral lymphography showed dermal backflow of radiopaque fluid in the right inguinal region, indicating proximal lymphatic obstruction. A normal number of lymphatics were seen in the legs but there was abnormally slow passage of fluid through the pelvic lymphatics despite massage and exercise of the legs. The transit time from feet to thoracic duct was three times the normal. These findings confirmed the diagnosis of lymphoedema praecox. Conservative treatment was recommended.

DISCUSSION

There has been no previously reported association between pes cavus and lymphoedema, and the combination in these two patients might be considered fortuitous. The family history in Case 1, however, precludes this possibility. The same combination of pathology in three persons within two generations indicates a genetically linked association.

The high incidence of congenital anomalies (other than the lymphatic one) in patients with lymphoedema congenita and praecox has been discussed elsewhere. Kinmonth, Taylor, Tracy and Marsh (1957) reported an incidence of 14 per cent of other deformities in patients with primary lymphoedema. There were several examples of skeletal deformities in conjunction with lymphoedema but pes cavus did not occur in that series. The deformities were noted in patients with lymphoedema praecox as well as congenital lymphoedema. Of the two patients reported here, one had congenital lymphoedema and the other lymphoedema praecox. In both cases the lymphatic defect was almost certainly due to a hereditary inborn error, in the first patient causing oedema at birth but in the other a condition of milder degree not manifesting itself until later in life.

SUMMARY

1. Two patients with bilateral pes cavus and lymphoedema are reported. Lymphography confirmed the existence of lymphatic insufficiency in both cases. In both there was evidence of hereditary factors in the etiology.
2. This familial syndrome appears not to have been previously reported.

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REFERENCES