Acta orthop. scand. 47, 525–533, 1976

POPLITEAL PTERYGIUM SYNDROME IN A 74-YEAR-OLD WOMAN

LARS INGVAR HANSSON, VIVECA HANSSON & KJELL JONSSON

Departments of Orthopaedic Surgery and Diagnostic Radiology, University Hospital, Lund, Sweden.

The case of a 74-year-old woman with the rare popliteal pterygium syndrome is presented. This syndrome is inherited as an autosomal dominant trait with incomplete penetrance and varying expression and consists of cleft lip and palate, lip pits, genital anomalies, popliteal pterygium, and malformations of the extremities. The various treatments our patient underwent over the years are reported. Treatment of popliteal pterygium involves special problems when removing the skin fold because the nerve and vascular cords lie immediately anterior to the posterior fibrous cord. In the present case there are widespread arthrotic changes, both in the extremity joints and in the spine. These patients are short in stature. This, together with the general arthropathy, suggests a hereditary metabolic disturbance in the cartilaginous tissue.

Key words: bone; bone diseases, developmental; cleft lip; cleft palate; clubfoot; growth disorders; hereditary diseases; pterygium; syndactylia

Accepted 12.vii.76

The popliteal pterygium syndrome consists of multiple anomalies, including cleft lip and palate, lip pits, genital anomalies, popliteal pterygium, and malformations of the extremities, but the malformations vary greatly in distribution (Hecht & Jarvinen 1967, Gorlin et al. 1968, Smith 1970, Sedano 1973). The syndrome is caused by gene mutation and is inherited as an autosomal dominant trait with incomplete penetrance and varying expression (Hecht & Jarvinen 1967, Gorlin et al. 1968, Sedano 1973). About 30 cases have been published (Smith 1970), the first cases being reported during the latter part of the nineteenth century (Trèlat 1869, Wolff 1889, Rydygier 1891, Basch 1891, 1892, Fischer 1893). The popliteal pterygium syndrome is diagnosed in infancy and early childhood (Champion & Cregan 1959).

The present case is interesting because of the many different malformations. The case history from birth to the present—the patient is 74 years of age—is well known and documented; moreover, the syndrome is interesting from the standpoint of the surgical treatment of the malformations of the extremities.

CASE HISTORY

Female, 74 years of age, height 142 cm, unmarried, intelligence normal (Figures 1, 2).

Heredity: She was the fifth born in a Swedish family of 10 siblings: 4 girls and 6 boys. One brother and one sister had conspicuously short

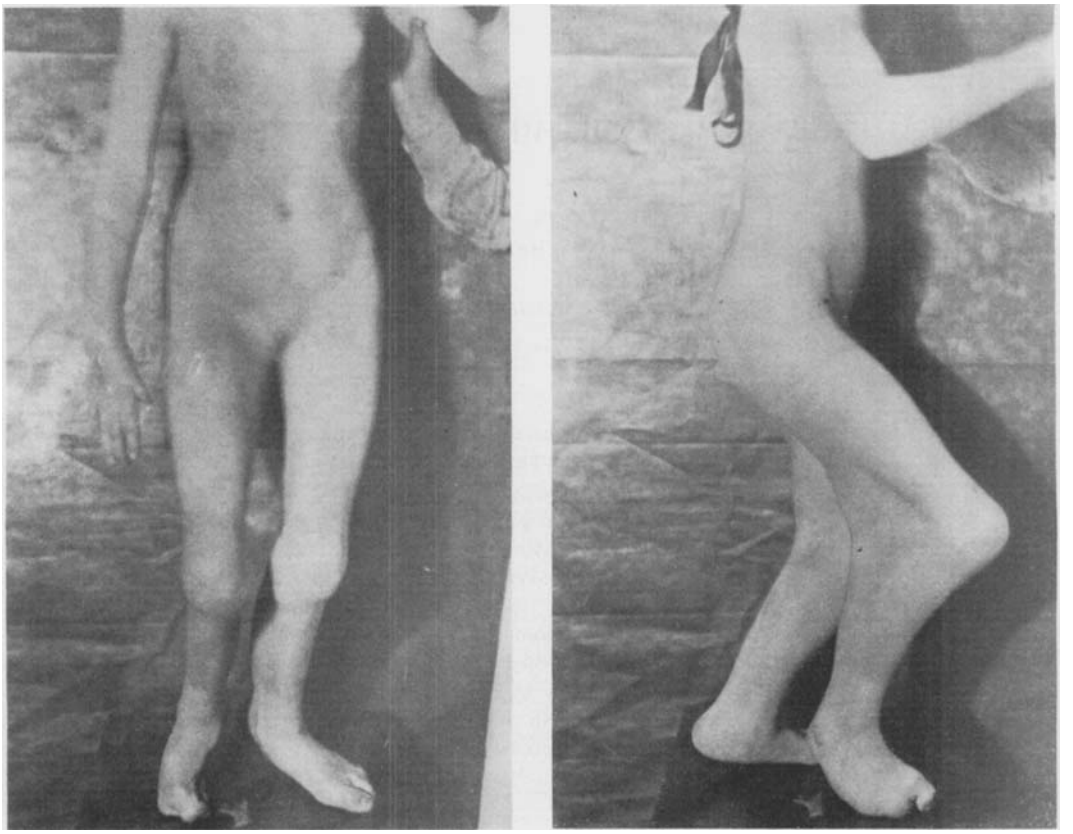


Figure 1. Photograph taken in 1914 at the age of 12 years. Left: Frontal view. Right: Side view.

hands and feet, and another sister had syndactyly of the fingers. The father also had short hands and feet; moreover, his shoulder, on one side, was smaller from birth. The patient's paternal grandmother and great grandmother had short hands and feet, and were noticeably short in stature. The great grandmother was actually called a dwarf. The mother's side of the family was normal. The patient's parents were related; her mother and father had the same ancestor, who lived from 1685 to 1762.

Birth: After a normal pregnancy, the patient was born in 1902 at home in Southern Sweden after a prolonged and difficult labour. She was judged to have a small chance of survival because of extensive anomalies.

Anomalies

A. Orofacial anomalies.

1. Fused eyelids (ankyloblepharon filiforme ad natum). At birth, a membrane between the eyelids was noted and cut bilaterally.

2. Cleft lip and palate (cheilo-gnatho-palatoschisis) (Figure 3). When an adult, the cleft was operated on to close the defects. She has now a posterior open cleft covered by an obturator which improves her speech.

3. Lower lip pits. At birth, bilateral pits were found in the lower lip, giving the impression of double lower lip. These pits were removed operatively at the age of 20.

4. Syngnathia. There were bands and membranes between the left upper lip and the nasal passage to the inside of the lower lip. These were cut at birth.

5. Oral webbing. Webbing between the upper and lower jaw, on both sides, was severed later in connection with operations in the oral cavity.

B. Extremity anomalies.

1. Popliteal pterygium (webbing). At birth, a fold of skin was found stretching from the gluteal region and tuber ossis ischii to the heels bilaterally with considerable extension defect in the knee joint and also equinus foot. She began to walk on her knees at 2 years of age and soon

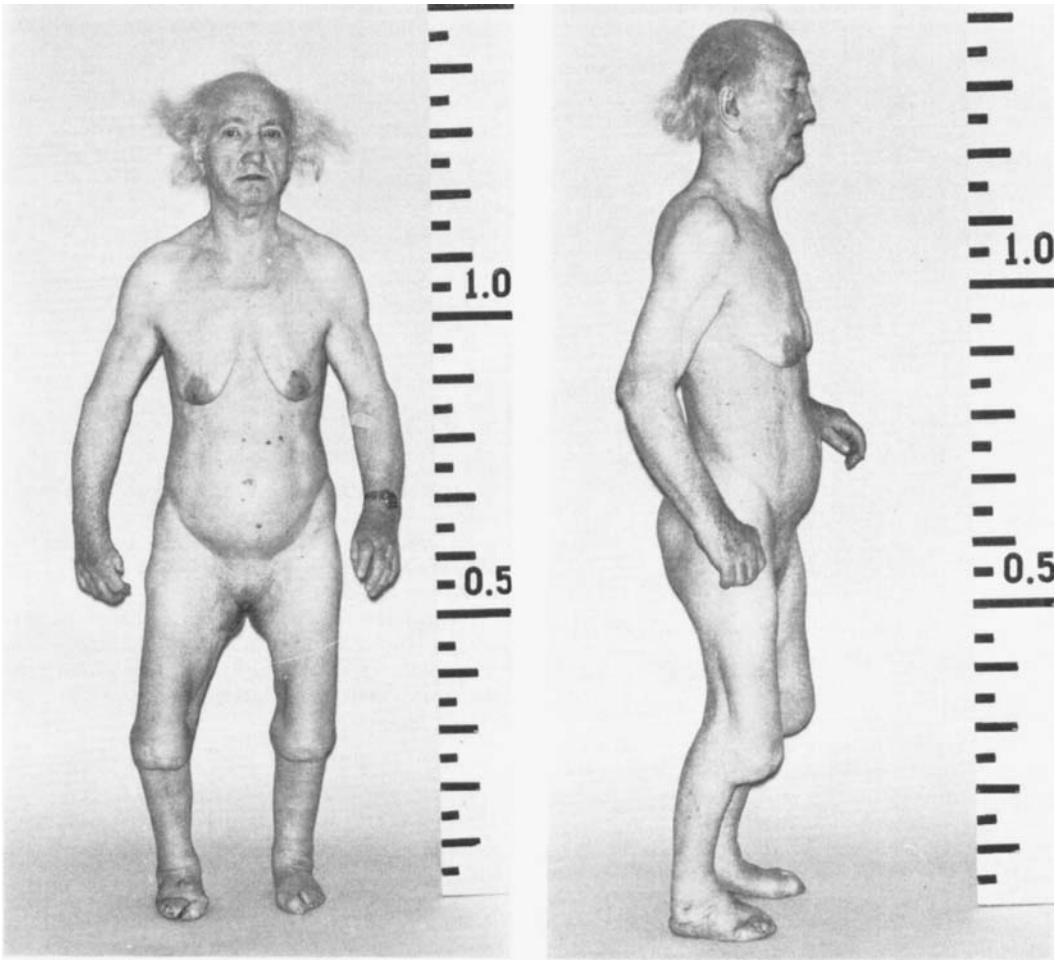


Figure 2. Photograph taken in 1973 at the age of 71 years. Left: Frontal view. Right: Side view.

she could walk and run, despite strongly bent knee joints. When she was 4, an attempt was made to extirpate a small piece of the skin fold on the left side, but the wound took 8 months to heal. When she was 12, the extension defects were about 90° bilaterally (Figure 1). At this age, bilateral exploration was carried out with the aid of longitudinal incisions laterally. This revealed a fibrous cord and the sciatic nerve and vessels furthest dorsally in the skin fold but no tight musculature; the operation areas were therefore closed. During the next 2 years, she was given correction treatment with plasters and splints. Since then, she has used lower limb braces, but has also been able to walk without braces. Bilaterally, she has a deformity in the knee joints with the femoral condyles dislocated ventrally to the tibial condyles, with considerable deformity of the femoral, tibial and patellar joint surfaces and an extension defect of 20–30° bilaterally (Figures 2, 4). In both lower limbs, there is also a varus deformity especially on the right side.

2. Foot anomalies. Primarily, a considerable equinus deformity existed on both sides (Figure 1); this was corrected (Figure 2). On the left side, there was also a valgus deformity and on the right side a varus deformity of the foot. The toes—except for the big toe—are connected by a syndactyly with nail dysplasia and aplasia. X-rays show a hypoplasia of the tarsal skeleton, and in the metatarsals dysplasia and aplasia; also osseous fusions between the metatarsal bones and between the first phalanges (Figure 5). In

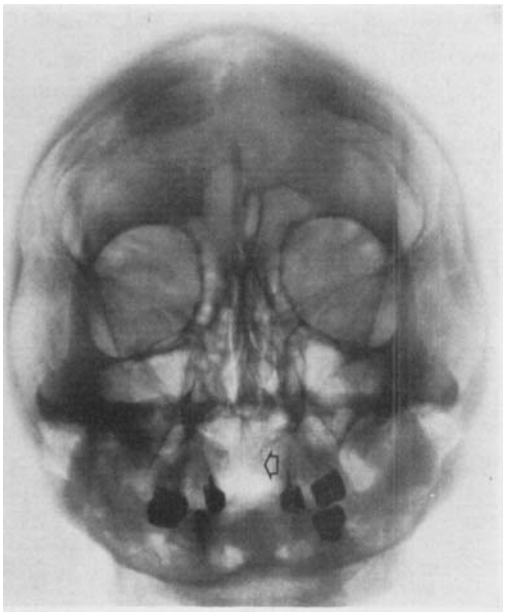


Figure 3. Skull. Roentgen examination. AP view. Note the cleft palate (arrow).

the foot region, no operations were carried out; instead, the deformities were corrected with plasters and splints. During adulthood, hallux valgus has developed bilaterally.

3. Hand anomalies. At birth, syndactyly was found in the hands between the middle and the ring fingers on both sides, and on the right side a hypoplasia of the thumb. The metacarpal skeleton and the phalanges are shorter than normal, and hypoplasia is found in the nail regions. At the age of 4 years, and again at 20, syndactyly operations were performed with good results. X-ray revealed that the right thumb is rudimentary, especially the base of the first metacarpal bone (Figure 6).

C. Genital and perineal anomalies.

External and internal genitalia correspond to female sex. Recent gynaecological examination showed virginal status. She has hypotrophic labia majora, vagina, and uterus; also hypertrophic clitoris. There is a tendency for intercrural webbing. Chromosome analysis of peripheral blood showed 46 chromosomes of normal female caryotype, 46/xx.

Menarche began at the age of 15 and menopause at 52. At 26 years of age she was operated for left-sided benign ovarian tumour.

D. Skin changes.

As mentioned, nail dysplasia and aplasia are found in the hands and feet.

On the trunk, the back of the hands, and lower arms, there are multiple pigmented naevi. The dermal ridge pattern, palmarly and plantarly, stretches somewhat more towards the dorsal parts of the hands and feet than is normal, but it is not found dorsally on the hands and feet.

In adulthood, she developed increasingly conspicuous male features as regards hair growth: baldness on the crown, beard on the face, and masculine hair in the axillae and pubis regions (Figure 2). Laboratory investigation did not show abnormal increase in metabolites, which would have indicated pathological androgen production.

Radiography

A recent extensive roentgenological investigation (1972–1975) showed three types of skeletal changes.

1. Congenital defects. Cleft palate (Figure 3), thumb-hypoplasia with a rudimentary base to metacarpal 1 dx (Figure 6); hypoplasia, aplasia and osseous fusion of the metatarsal bones and of the phalanges (Figure 5).

2. Secondary changes (i.e. secondary to congenital anomalies). Deformity of the knee skeleton because of extension defects and pronounced secondary arthrosis development (Figure 4).

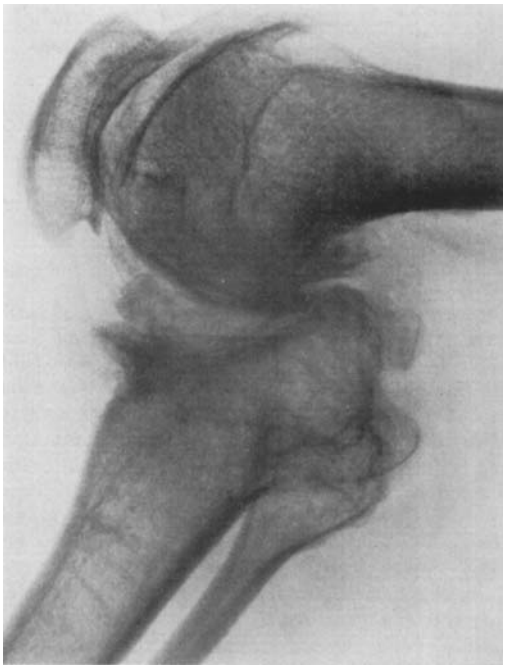
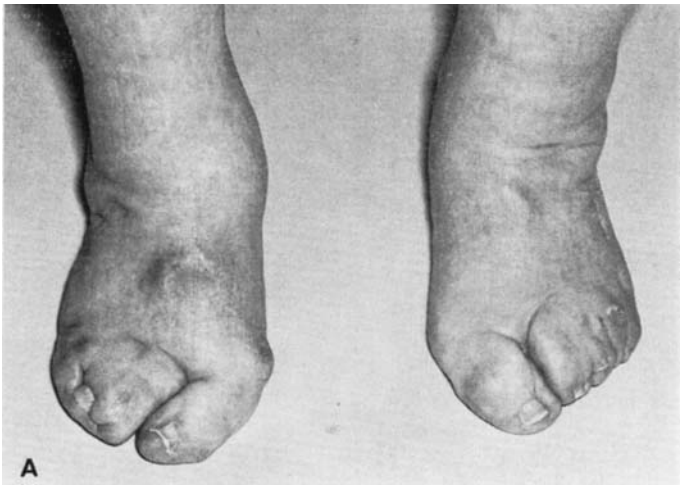
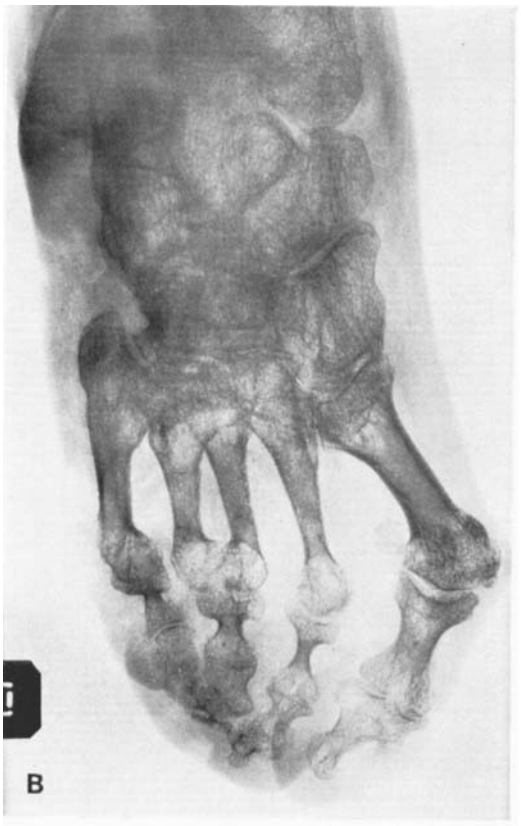


Figure 4. Left knee. Roentgen examination. Lateral view.





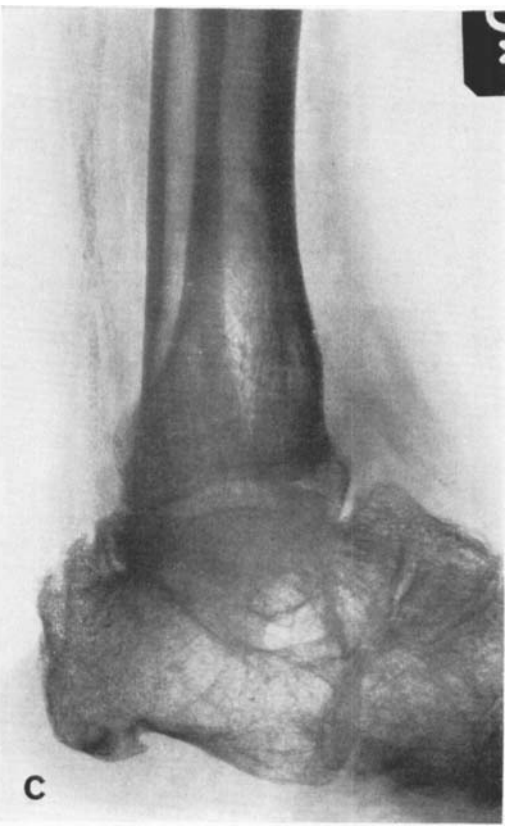


Figure 5. Foot and ankle.
A. Right and left foot. Photograph. Dorsal view.
B. Right foot. Roentgen examination. AP view.
C. Right ankle. Roentgen examination. Lateral view.

Deformity of the calcaneus and the foot skeleton because of an equinus position (Figure 5).

3. Other changes. Apart from earlier reported anomalies and deformities, there is general arthropathy with reduced articular cartilage, pronounced osteophytes and periarticular calcifications in many joints. Bilaterally, the hip joints show an increased CE angle (Wiberg 1939), which has become even more pronounced because of considerable osteophyte formation in

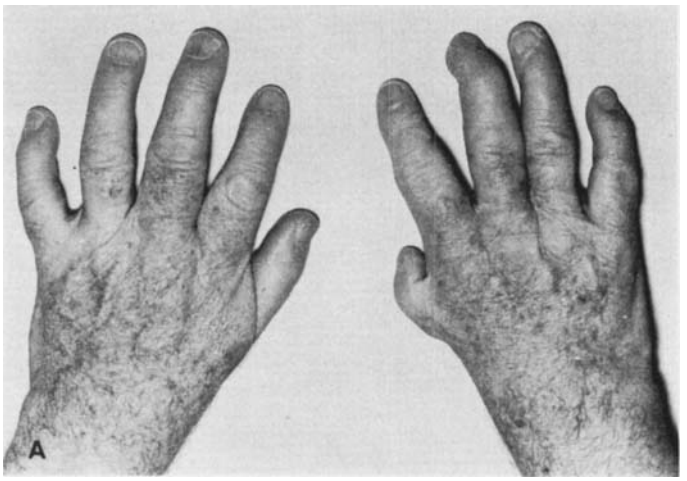
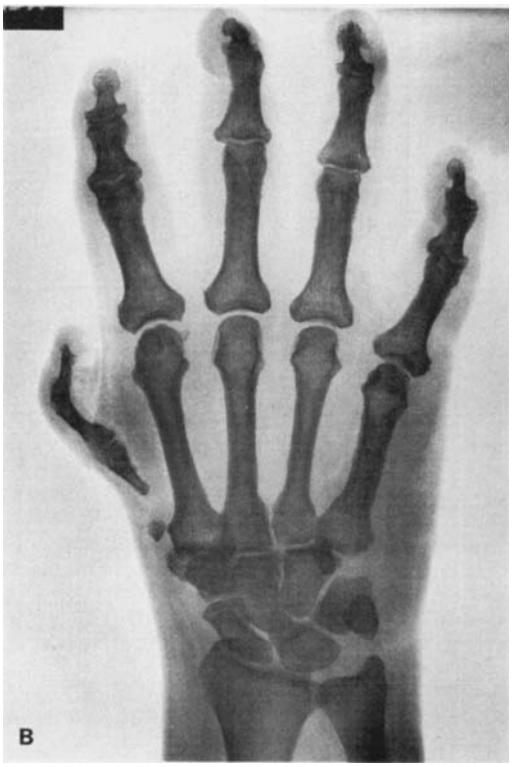


Figure 6. Hand.
A. Right and left hand. Photograph. Dorsal view.
B. Right hand. Roentgen examination. AP view.



the acetabulum. In the spine, there are generally severe spondylous changes.

Unrelated changes

In 1914 she had diphtheria. This was followed by bilateral otitis and mastoiditis. These were treated surgically on several occasions and resulted in severe reduction of her hearing bilaterally. In recent years, slight diabetes mellitus, nontoxic goiter, and cholelithiasis developed.

Social life

During childhood and adolescence, her schooling was satisfactory. Later, she had office training and worked in an office as a clerk for about 2 years. During the following years—up to the age of 50—she took care of her ageing parents. She was granted an orthopaedic disability pension when she was 30. Since 1964 she has lived alone in a modern flat on the first floor in a house with no elevator and has had the services of a home visitor for a few hours daily.

Up to the age of 50, she was able to walk both with, and without, braces for a distance of 2 kilometres. However, for the past 10 years she has used a stick and has been able to walk only about 300 metres.

Since the age of 20, she has experienced some temporary localized pain, but of low intensity, usually in the knees, the hips, or the shoulders. This has shown a tendency to increase during the past 10 years, but has not required analgesics.

DISCUSSION

The anomalies that exist in this case agree well with descriptions of popliteal pterygium syndrome and are considerably more extensive than the anomalies which, according to Sedano (1973), are required as minimum diagnostic criteria.

The most obvious anomaly—popliteal pterygium or webbing (Patagium, Flughautbildung)—has for a long time given the syndrome its name. The syndrome occurs with considerable variation, from insignificant anomalies, difficult to discover, to extremely extensive and serious anomalies. This has earlier been shown in families (Roselli & Gulienetti 1961, Klein 1962, Gorlin & Pindborg 1964, Hecht & Jarvinen 1967). Therefore an investigation should be made of the family where a member with isolated defects can point to popliteal pterygium syndrome. Thus it can be reasonably suspected that considerably more persons have this syndrome, complete or incomplete, than are reported in the literature, where the account, as a rule, refers only to cases with severe deformities or to families with members that have varying deformities. A larger proportion of the published cases come from Central Europe and from the USA (Gorlin et al. 1968), whereas no earlier case has been reported from the Scandinavian countries.

Naturally, the literature mostly describes children, as it is usually noted during the newborn period; therefore it is interesting to be able to report the present case from birth to old age. This has been possible because the various deformities and the treatment given have been carefully recorded both by the patient and the hospitals. Moreover, in this case, the family tree is well documented as far back as 300 years because of an interest in genealogy in the family.

There are several instances that can suggest milder forms of popliteal pterygium syndrome, in earlier generations and in the same generation, which agrees with the fact that the syndrome is inherited as autosomally dominant with incomplete penetration and varying expression (Hecht & Jarvinen 1967, Gorlin et al. 1968, Sedano 1973). Intermarriages are also earlier reported in the family.

The differential diagnostics in popliteal pterygium syndrome have been covered by Leiber & Olbrich (1966), Smith (1970), and Sedano (1973). The diagnosis in the severe cases does not seem to be difficult, whereas differential diagnostic difficulties exist where there are only a few anomalies or only orofacial anomalies, such as cleft lip and palate (Schönenberg 1955), because, according to Gorlin et al. (1968), there are about 20 different syndromes with this localized type of anomaly. This applies also to the other anomalies involved in popliteal pterygium syndrome although to a lesser degree, because the number of syndromes involved are fewer (Smith 1970). According to Sedano (1973), arthrogryposis multiplex congenita can be a differential diagnostic alternative for extremity anomalies with webbing, but in these cases, orofacial and peripheral extremity anomalies are lacking.

Congenital webbing is described in the literature in other localizations, such as the elbow, shoulder, and neck region, either associated with popliteal pterygium (Matolcsy 1936, Kopits 1937) or as an isolated deformity (Schramm 1940, Shun-Shin 1954, Smith 1970). Probably some of these are incomplete cases of popliteal pterygium syndrome, whereas others are of another genesis.

In the present case, some pigmented naevi were found on the trunk and the upper extremities, but these changes do not seem to differ in localization and size from what could normally be expected, although it is said that these changes are pathognomonic of popliteal pterygium syndrome (Leiber & Olbrich 1966).

According to the literature, the treatment of orofacial anomalies has usually been surgical in by far the majority of cases, as it has in the present case. Syndactyly in the hands has usually been operated on in infancy and childhood.

The treatment of popliteal pterygium and the flexion contracture of the knee

joint and the equinus foot has varied considerably. In some cases the only treatment was braces (Dahmen 1962). Marquardt (1938) employed conservative treatment with traction and plasters. Operations were made on soft tissues (Kopits 1937, Aberle-Horstenegg 1938, Schramm 1940, Lewis 1948, Champion & Cregan 1959, Fèvre & Languepin 1962, 1966) and bone in the form of osteotomy or articular resection in the knee region (Hackenbroch 1924, Matolcsy 1936). In the present case, at exploration in 1914, it was noted bilaterally that furthest dorsally in the skin fold from tuber ossis ischii to the heel there was a fibrous cord next to the nerve and vascular cords; therefore a radical operation was judged impossible. Later, a corresponding observation was made by Kopits (1937), Lewis (1948), Champion & Cregan (1959), and Fèvre & Languepin (1962, 1966). Champion & Cregan (1959) succeeded, in both their patients, in eliminating the extension defect without jeopardizing the vascular and nerve cords by Z-plastic and extirpation of the dorsal fibrous cord.

During infancy and childhood, our patient had a bilateral equinus deformity with varus on the right side and valgus on the left. The equinus deformity was later almost completely eliminated by correction with plasters and splints, but this was probably also affected by the contracture treatment in the knee joint, as the dorsal fibrous cord is attached to the heel part.

The arthrosis development seen in most joints in our patient is easily accounted for, for instance, in the knee joints where there is marked deformity with ventral dislocation of the femur in relation to the tibia. This was probably caused partly by the fibrous structures seen in the skin fold, and partly by the contracture treatment. However, there are also noticeably advanced arthrotic changes, for instance, in the elbow joints, shoulder joints, and hip joints, and also in the spine. Hip changes with an increased CE angle were earlier described by Hecht & Jarvinen (1967); our patient has a larger CE angle than normal, and it has increased further due to osteophyte formation. The widespread arthrotic changes suggest a general metabolic arthropathy. This is also supported by the fact that patients with popliteal pterygium syndrome are short in stature, probably due to a metabolic disturbance in the cartilaginous tissues, both in the growth plates and in the articular cartilage.

ACKNOWLEDGEMENTS

The authors express their sincere thanks to Miss Alfhild Runbäck for the material that she made available to them.

REFERENCES

Aberle-Horstenegg, W. (1938) Flughautbildung zwischen Ober- und Unterschenkel mit abnormer Muskelbildung. *Z. orthop.* **67**, 21–29.

Basch, K. (1891) Ueber sogenannte Flughautbildung am Menschen. *Prag. med. Wschr.* **16**, 572–573.

Basch, K. (1892) Ein weiterer Fall von sog. Flughautbildung. *Prag. med. Wschr.* **17**, 289–291.

Champion, R. & Cregan, J. C. F. (1959) Congenital popliteal webbing in siblings. *J. Bone Jt Surg.* **41**-B, 355–357.

Dahmen, G. (1962) Über die Vorsorgung einer doppelseitigen Kniestreckhemmung wegen Flugelfell mit einem provisorischen Knie-Ruhebein. *Z. Orthop.* **95**, 112–113.

Fèvre, M. & Languepin, A. (1962) Les brides cruro-jambières contenant le nerf sciatique. Le syndrome bride poplitée et malformations multiples. *Presse méd.* **70**, 615–618.

Fèvre, M. & Languepin, A. (1966) Brides poplitées simples et brides poplitées avec quadruple syndrome. *Ann. Chir. Plast.* **11**, 124–127.

Fischer, H. (1893) Ein Fall von sogenannter Flughautbildung zwischen Ober- und Unterschenkel. *Prag. med. Wschr.* **18**, 579–581.

Gorlin, R. J. & Pindborg, J. J. (1964) *Syndromes of the head and neck.* McGraw-Hill, New York, Toronto, London.

Gorlin, R. J., Sedano, H. O. & Cervenka, J. (1968) Popliteal pterygium syndrome. *Pediatrics* **41**, 503–509.

Hackenbroch, M. (1924) Ueber einen Fall von kongenitaler Kontraktur der Kniegelenke mit Flughautbildung. *Z. Orthop. Chir.* **43**, 508–524.

Hecht, F. & Jarvinen, J. M. (1967) Heritable dysmorphic syndrome with normal intelligence. *J. Pediat.* **70**, 927–935.

Klein, D. (1962) Cas observé. *J. Génét. Hum.* **11**, 65–71.

Kopits, E. (1937) Die als Flughaut bezeichneten Missbildungen und deren operative Behandlung. *Arch. orthop. Unfall-Chir.* **37**, 539–549.

Leiber, B. & Olbrich, G. (1966) *Die klinischen Syndrome.* Band 1. Urban und Schwarzenberg, München, Berlin, Wien.

Lewis, E. (1948) Congenital webbing of the lower limbs. *Proc. roy. Soc. Med.* **41**, 864.

Marquardt, W. (1938) Die angeborene Flughautbildung und ihre konservative Behandlung. *Z. Orthop.* **67**, 379–386.

Matolcsy, T. (196) Über die chirurgische Behandlung der angeborernen Flughaut. *Arch. klin. Chir.* **185**, 675–681.

Rosselli, D. & Gulienetti, R. (1961) Ectodermal dysplasia. *Brit. J. plast. Surg.* **14**, 190–204.

Rydygier, L. (1891) Demonstration von Abbildungen selterner Fälle von Missbildungen. *Arch. klin. Chir.* **42**, 769.

Schramm, G. (1940) Über die angeborene Flughautbildung. *Z. Orthop.* **70**, 189–195.

Schönenberg, H. (1955) Über die Kombination von Lippen-Kefer-Gaumen-Spalten mit Extremitätenmissbildung. *Z. Kinderheilk.* **76**, 79–90.

Sedano, H. O. (1973) Popliteal pterygium syndrome. In: *Birth defects.* Ed. Bergsma, D., p. 742. Williams and Wilkins, Baltimore.

Shun-Shin, M. (1954) Congenital web formation. *J. Bone Jt Surg.* **36**-B, 268–271.

Smith, D. W. (1970) *Recognizable patterns of human malformation.* Saunders, Philadelphia, London, Toronto.

Trélat, U. (1869) Sur un vice de conformation trés-rare de la lèvre inférieure. *J. Méd. chir. Prat.* **40**, 442–445.

Wiberg, G. (1939) Studies on dysplastic acetabula and congenital subluxation of the hip joint. *Acta chir. scand.*, Suppl. 58.

Wolff, J. (1889) Ueber einen Fall von angeborener Flughautbildung. *Arch. klin. Chir.* **38**, 66–73.

Correspondence to: Lars Ingvar Hansson, Department of Orthopaedic Surgery, University Hospital, S–22185 Lund, Sweden.