Syndrome of the month

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Popliteal pterygium syndrome

U G Froster-Iskenius

The popliteal pterygium syndrome is a highly characteristic congenital malformation syndrome affecting the face, limbs, and genitalia. Gorlin *et al*^{1 2} coined the term 'popliteal pterygium syndrome' on the basis of the most unusual anomaly, the popliteal web. In some publications the names of Fèvre and Languépin³ are used as an eponym. A more descriptive term suggested for the condition, on the basis of incomplete expression of the features of the syndrome, is 'faciogenitopopliteal syndrome'.⁴ However, the most widely used term for this disorder is 'popliteal pterygium syndrome'. Autosomal dominant inheritance with highly variable expressivity and incomplete penetrance is widely accepted. The term popliteal pterygium syndrome has also been used for two autosomal recessively inherited conditions,^{5 6} which are, however, clinically distinguishable.

Historical notes

The formation of tissue webs across the popliteal fossa is an uncommon event. First reports of this anomaly in medical publications date back to the second part of the 19th century. The first report is attributed to Trélat in 1869.⁷ Wolff⁸ reported a female patient with popliteal webs and further congenital anomalies, including a caudal appendage and reduction defects of the toes, to the Society of Surgeons in Berlin. He compared the very unusual webs with pterygia seen in some animals and stated that pterygia in the popliteal fossae had never been described in animals. Basch⁹ reported a 4 month old infant with popliteal pterygia, who died in infancy, and gave a very detailed necropsy report and drawing of the abnormal placement of tendons and muscle insertions within the pterygia. Kopits¹⁰ described four cases, three of

them belonging to the same family, and gave details of the operative techniques used.

To date a minimum of 81 patients with popliteal pterygium syndrome (PPS) can be found in published medical reports and at least 22 families with the disorder have been described.

The diagnosis has been made in various ethnic groups; beside Caucasians, Japanese¹¹ and Black families^{12 13} have been reported.

Incidence

The disorder is very rare and an incidence figure is difficult to calculate. Among 21 170 cases in an orthopaedic clinic, Kopits¹⁰ had observed four cases from two families. Assuming that both families had popliteal pterygium syndrome, an incidence of 0.9/10 000 among orthopaedic patients would be calculated. Hecht and Jarvinen,¹⁴ out of 594 cases with cleft lip or palate born between 1954 and 1963 in Oregon, described only one family with two affected sibs with popliteal pterygium syndrome. The incidence of cleft lip/palate in the general population varies between 10 and 18 per 10 000.¹⁵ Calculated from this, the incidence of the popliteal pterygium syndrome would be 1/300 000, which makes it an extremely rare condition.

Clinical features

The features of the syndrome are highly variable and show a wide range of expressivity even within families. Orofacial, cutaneous, musculoskeletal, and genital anomalies occur.

OROFACIAL ANOMALIES

Cleft palate with or without cleft lip has been found to be the most frequent anomaly in popliteal pterygium syndrome, being present in 91 to 97% of cases^{2 11} (table 1). All degrees of severity of clefting are described, ranging from bilateral cleft lip and palate to just a split uvula. Paramedian sinuses or pits of the

Department of Obstetrics and Gynaecology, Lübeck Medical University, Ratzeburger Allee 160, D-2400 Lübeck, Federal Republic of Germany.
U G Froster-Iskenius

Table 1 Frequency of anomalies in familial cases of popliteal pterygium syndrome.

	Reference																				No/total	%			
	12	13	14 a	14 b	17	19	20	21	22 a	22 b	23	24	25	26 a	26 b	27	28	29	30	31			32	33	
Affected family members	3	2	3	3	2	3	3	3	2	2	2	2	3	2	2	3	3	3	3	2	3	3			
Anomalies																									
Cleft palate	3	1	3	3	2	3	3	2	2	1	2	1	3	2	2	3	3	3	1	2	3	3	53/57	92.9	
Cleft lip	2	1	3	2	1	1	1	1	1	0	0	1	1	1	1	3	3	3	1	2	3	2	33/57	57.8	
Syngnathia	0	1	2	2	1	2	1	3	1	0	1	0	0	0	1	2	1	1	2	2	0	0	23/57	40.3	
Lip pits	0	0	2	3	1	1	0	1	1	0	1	2	1	2	0	3	3	1	1	1	0	2	26/57	45.6	
Ankyloblepharon	0	2	0	1	0	1	0	1	1	0	2	0	0	0	0	1	0	0	1	0	0	0	10/57	17.5	
Popliteal web	2	1	2	2	1	1	0	1	1	1	2	1	2	2	1	3	3	2	1	1	2	2	33/57	57.8	
Syndactyly	1	1	1	1	2	1	2	2	1	2	2	1	2	2	1	0	2	1	1	2	0	1	29/57	50.8	
Intercrural web	0	0	0	0	1	0	0	0	0	0	0	0	0	0	0	0	2	0	1	1	0	0	5/57	8.7	
Talipes equinovarus	0	0	1	0	0	0	0	1	0	0	0	1	2	1	1	0	0	0	1	0	0	0	8/57	14.0	
Digital reduction defects	1	0	1	0	0	0	0	0	0	0	0	0	0	1	0	0	0	1	0	3	0	0	7/57	12.2	
Genital anomalies	2	0	0	2	1	1	1	0	0	0	1	2	0	1	2	1	0	3	0	1	1	0	2	21/57	36.8
Nail anomalies	1	1	1	2	1	1	2	0	1	0	1	1	1	2	0	1	0	0	1	1	0	1	19/57	33.3

(a)

(b)

Figure 1 (a) Bilateral cleft lip and palate in a newborn boy with popliteal pterygium syndrome. Lip pits are visible in the lower lip. (b) Same patient at 3 years of age after surgical repair of the facial clefts.

lower lip occur in 45.6% of cases (fig 1). Intraoral tissue bands (syngnathia) were found in 42.6% of cases. They can seriously affect mouth opening and need to be removed surgically within the first year of life, or directly after birth if limitation of opening the mouth is so severe that feeding problems occur. Ankyloblepharon filiforme adnatum is found in approximately 20% of cases.

CUTANEOUS AND MUSCULOSKELETAL ANOMALIES

The most pertinent clinical feature is the popliteal pterygium (fig 2), reported to occur in 89.7 to 96% of cases.¹¹ This is probably an overestimation, considering that ascertainment of the syndrome usually occurs through patients with a popliteal web. Among familial cases this anomaly is found in 56.8% (table 1).

Figure 2 Popliteal pterygium: note the dense cord inhibiting full extension of the knee.

Figure 3 The peroneal nerve and vessels partly cleaned from the connective tissue of the popliteal web.

The popliteal web contains a palpable cord of connective tissue, and may contain the popliteal artery and the peroneal nerve. The cord usually extends from the heel to the ischial tuberosity. It may seriously limit extension, abduction, and rotation of the leg. In some subjects it was reported to be so tight that the heel almost touched the buttocks, while in other cases it could just be felt as a tight string without any severe limitation of the range of movements. Absence of muscles or abnormal muscle and tendon insertion may occur. On surgical intervention, care must be taken not to cut the vessels or nerves which supply the lower leg (fig 3). A thorough preparation of the tissue by plastic surgery with lengthening of the tendons and Z plasty of the skin is usually followed by a series of casting procedures (fig 4).^{16 17} Additional pterygia across the inguinal fossa occur in approximately 9% of cases. Webs across other joints are not

(a)

(b)

(c)

Figure 4 Situation after Z plasty and casting in a 5 year old patient with popliteal pterygium syndrome. (a) Full extension of the knee is possible. (b) Same patient at 10 years. (c) Note recurrence of contracture.

Table 2 Differential diagnoses in the popliteal pterygium syndrome.

Syndrome	Cleft lip/ palate	Lip pits	Syngnathia	Popliteal pterygium	Other	Inheritance	Reference
Cleft lip/ cleft palate syndromes	+	—	—	—	Various (see reference)	AD	34
van der Woude syndrome	+	+	—	—		AD	35
Cleft palate/ lateral synechia syndrome	—/+	—	+	—	Hypoplastic lower jaw and tongue, short upper lip	AD	36
Lethal popliteal pterygium syndrome	+	—	+	+	Microcephaly, corneal aplasia, ectropion, bony fusions, hypoplastic nose, absent thumbs	AR	5
Popliteal pterygium syndrome with ectodermal dysplasia	+	—	+	+	Woolly hair, brittle nails, ectodermal anomalies, fissure of sacral vertebrae	AR	6

part of the syndrome and suggest a different condition (table 2).

A very distinctive nail anomaly (fig 5) with a pyramidal skinfold extending from the base to the top of the nails has been described in up to 33·3% of cases. Other nail anomalies, such as hypoplastic nails, mostly involve the toes. Syndactyly of the toes or fingers occurs in up to 50·8%, usually affecting the second and third toe, but occasionally also toes 2 to 5. Reduction defects of the fingers or toes are rare, as are bifid first toes or hypoplastic or absent thumbs (fig 6).³⁷ Other skeletal anomalies include talipes equinovarus, spina bifida occulta, bifid ribs, and short sternum. Hyperpigmentation of the cord running across the popliteal fossa has been mentioned in two patients.

Figure 5 A pyramidal skinfold extending from the base to the tip of the nails in a patient with popliteal pterygium syndrome. Also note nail hypoplasia and syndactyly of the second and third toes, more prominent in the right than in the left foot.

Figure 6 Reduction defects of the thumb in a patient with popliteal pterygium syndrome.

GENITOURINARY ANOMALIES

Anomalies of the genitalia are reported in both female and male patients. In females, the most frequent finding is hypoplastic labia majora, but hypoplastic vagina and uterus have also been mentioned, as well as clitoral hypertrophy. In males, uni- or bilateral cryptorchidism, bifid or absent scrotum, but usually a normal sized penis are found (fig 7). In severe cases, genital anomalies may result in reduced reproductive fitness. Inguinal herniae are occasional but non-specific findings.

GROWTH AND MENTAL DEVELOPMENT

There is no recognisable growth problem. Birth weight and length, as well as head circumference at birth, are usually reported as normal. Intelligence is

Figure 7 Hypoplasia with cryptorchidism on the right in a patient with popliteal pterygium syndrome.

usually normal. Few cases with reduced intelligence have been reported. In the case described by Kopits,¹⁰ additional clinical anomalies, such as contractures of the upper limbs, epicanthic folds, a facial haemangioma, dysplastic ears, and spina bifida occulta, are suggestive of the multiple pterygium syndrome. In the female described by Wolff,⁸ additional unusual anomalies included a caudal appendage. The intellectual deficit, however, was attributed to a lack of schooling in this patient. The patient of Hackenbroch¹⁸ had, in addition, double rows of teeth, epicanthic folds, and spina bifida occulta. Diagnosis in these three cases is uncertain. In all other reported cases with confirmed popliteal pterygium syndrome intelligence was normal wherever explicitly mentioned.

Range of manifestation

While most of the sporadic cases and most index patients in the familial cases had popliteal pterygia, affected relatives frequently had only cleft palate or cleft lip/palate, or ankyloblepharon or lip sinuses or both (table 1). They probably would not have been identified as carriers of the popliteal pterygium syndrome if they had remained the only affected subject in the family.

The minimal diagnostic criteria for popliteal pterygium syndrome are any three of the following.²²

- (1) Cleft lip/palate.
- (2) Popliteal pterygium.
- (3) Paramedian lower lip sinuses.
- (4) Genital anomalies.
- (5) Toenail anomalies.

Differential diagnosis

Differential diagnosis is most important in the sporadic cases and in those with only mild expression. Two groups of disorders need to be considered: (1) syndromes with similar orofacial anomalies,³⁴⁻³⁶ and (2) disorders with similar limb defects^{5 6 38} (table 2). The first group includes cleft palate as an isolated inherited anomaly, cleft lip and palate syndromes,³⁴ van der Woude's syndrome,³⁵ which presents with paramedian lower lip pits and oral clefts and is inherited as an autosomal dominant trait, the cleft palate-synechia syndrome, which is also autosomal dominantly inherited, and the syngnathia congenita syndrome, which does not present with cleft palate and is also transmitted in an autosomal dominant fashion.³⁶ The combination of a hypoplastic thumb and cleft lip and palate can also be found in Juberg-Hayward syndrome,³⁹ which in addition presents with microcephaly. Syndactyly and hypoplastic genitalia can also be seen in Fraser's syndrome,⁴⁰ which, however, never presents with popliteal pterygia, but in the classical form has uni- or bilateral cryptophthalmos.

Among disorders presenting with similar limb defects the most important differential diagnosis is the disorder described by Bartsocas and Papas⁵ in a consanguineous family. This condition presents with severe popliteal webs, facial clefts, syngnathia, ankyloblepharon, thumb aplasia, severe syndactyly of the fingers and toes, hypoplastic labia majora, microcephaly, corneal aplasia, and hypoplastic nasal alae. In further cases of this apparently autosomal recessively inherited condition, ectropion and severe reduction of metacarpals and metatarsals in the completely syndactylous hands and feet have been described.^{37 41 42}

Another distinctive syndrome with popliteal pterygia⁶ displays, in addition, genitourinary tract malformations including absence of a kidney, fissures of the sacral vertebrae, epithelial lesions including dystrophic scalp with pityriasis scales, woolly, opaque hair, and dystrophic nails with subungual hyperkeratosis, scaling, and thinning of the lamina. Mental retardation, ectodermal dysplasia, and cicatricial atrophy of the scalp are the most important differentiating features of this autosomal recessively inherited syndrome.

The multiple pterygium syndrome⁴³ and lethal pterygium syndrome³⁸ also need to be considered in the differential diagnosis. However, they are usually clearly distinguished by pterygia formation across various other joints or additional, mainly vertebral, anomalies.

Natural history/management/treatment

The overall prognosis of the popliteal pterygium syndrome is good. Patients often undergo a series of plastic surgery operations, usually starting in the

newborn period and extending into puberty. In the newborn period the ankyloblepharon and oral synechia are excised to enable eye opening and proper feeding. Cleft lip and palate repair are done in consecutive sessions starting in the first year of life. An artificial palate can be placed temporarily if feeding is difficult. The cleft palate will usually be closed within the first year of life.

Speech and hearing problems may develop secondary to the clefting abnormality. If salivation from the lower lip pits is severe, these need to be surgically removed.

Early surgical intervention of the popliteal webs appears to be important with respect to long term results. Operations include excision of the fibrous band, mobilisation of nerves and vessels, Z plasty of the skin, and removal of tenotomy. During the operation special attention needs to be given to the vessels and nerves within the pterygium. Post-operatively, plaster casts and physiotherapy are used to maximise long term results.

Cryptorchidism may require surgical intervention within the first three years of life.

In a patient with popliteal pterygium syndrome, whom we were able to follow for 10 years,¹⁷ initial orthopaedic intervention for the popliteal pterygium was performed at 2 months of age. At 6 months the achilles tendons were lengthened and a bilateral capsulotomy was undertaken. Plaster casts were worn during the night and special casts during the day. At 18 months a vertical talus on the left foot had to be corrected. At 3½ years a recurrence of contractures in the right knee required a readjusting operation. A hearing problem occurred secondarily to frequent ear infections and ear tubes were inserted. He receives speech therapy because of the severe cleft of his palate. At 10 years he walks well, plays football, has almost complete mobility of his knees, and is of normal intelligence.

Inheritance

Affected subjects in consecutive generations and male to male transmission have been reported, making autosomal dominant inheritance likely.^{14 24 32 33} The male:female ratio is 1:1.¹⁴ Even though all affected subjects have the same disorder, they differ considerably in their pattern of expression of malformations. Monozygotic twins with different features of the popliteal pterygium syndrome have been observed.³¹ Variable expression occurs between generations, but sibs show a tendency towards similar phenotypic expression. Owing to incomplete expression, the syndrome is frequently only recognised if a family member with the fully expressed condition, and in particular a popliteal web, is born.

Most reported cases are sporadic; however, information on family history is very incomplete in

most sporadic cases. Since the primary gene defect is not known, a detailed examination of all first degree relatives of affected subjects is essential for genetic counselling. Special emphasis has to be given to minor anomalies or defects, such as syngnathia or ankyloblepharon, which might have been repaired long before examination.

Genital anomalies may cause infertility and thus affect the subject's reproductive fitness. This, and the high number of sporadic cases, suggest a high rate of new mutations. In seven sporadic cases where parental ages were given, the age of the mothers ranged between 22 and 46 years (average 32 years) and of the fathers 29 to 66 years (average 39 years). Thus advanced paternal age is found in a number of the sporadic cases, suggesting new mutations.

Prenatal diagnosis

Prenatal detection of cleft lip and palate by ultrasound is possible. In severe cases, the popliteal pterygium can also be disclosed by ultrasound because of abnormal movements and lack of ability to stretch the knee. However, in view of the good overall prognosis and the usually normal intelligence, ethical questions need to be taken into consideration if termination of pregnancy is requested by the parents.

Pathogenesis

The pathogenesis of the syndrome is only partly understood. Intraoral webs and ankyloblepharon filiforme adnatum suggest excessive proliferation of the epithelial layer of the blastoma of the lower jaw and eyelids, occurring in the early first trimester of pregnancy.^{44 45} In the popliteal pterygium, displacement of muscles and tendons occurs.¹⁶ The underlying mechanism for the popliteal pterygium syndrome is thus clearly different from the mechanism in multiple pterygium syndrome, where no displacement of muscles is found and the pterygia formation is attributed to limited intrauterine joint mobility.³⁸

The congenital sinuses of the lower lips are thought to be a remnant of the lateral sulci originating from a genetic defect.⁴ They have recently been found in a patient with a deletion of chromosome 1q,⁴⁶ who, in addition, had developmental delay, conductive hearing loss, microcephaly, and various dysmorphic facial anomalies, but no clefting of the palate or pterygia formation. However, chromosomal analyses of patients with popliteal pterygium syndrome have so far shown normal results, so that at present there is no suggestion for a possible gene location or the nature of the primary gene defect.

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