liac disease. J. Pediatr 1974, 85: 527-529.
4. Visakorpi JK. Diabetes and celiac disease. Lancet 1969, ii: 1192.
5. Komrower GM. Celiac disease in a diabetic child. Lancet 1969, i: 1215.
6. Stokes PL, Asquith P, Holmes GKT, Mackintorh P, Cooke WT. Histocompatibility antigens associated with adult celiac disease. Lancet 1972, ii: 612.
7. Cudowth AG, Woodrow JG. HLA antigens and diabetes mellitus. Lancet 1974, ii: 1153.
8. Mauriac P. Gros ventre, hepatomegalie, troubles de la croissance chez les enfants diabetiques: Traites depcus plusiers annes par l'insuline. Gaz Hebe Sci Med, Bordeaux 1930, 26: 402-410.
9. Hanson P. Enlargement of the liver in diabetes mellitus. JAMA 1936, 106: 914-916.

# Lethal Multiple Pterygeum Syndrome

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Lethal multiple pterygeum syndrome (LMPS) manifests in the newborn with multiple contractures and have been subclassified based on associated features(1). They are often referred to the geneticists as cases of arthrogryposis multiplex congenita (AMC). Identification of this group with pterygeum is important for management and counselling. We present here four cases because of the rarity of this condition and importance of recognition as a separate entity, as less than 50 cases have been reported so far in the world literature(2).

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## Case Reports

Four cases being reported were referred to the Genetics Department in 1990, as cases of AMC for evaluation and counselling. The details of the four cases are given in *Table I.*

## Discussion

All the above four cases were diagnosed as multiple pterygeum syndrome. All were male. The male:female ratio as per literature is 1:1. Two of the four couples were consanguinous and one was advanced in age. There was no recurrence or family history of similar defect. All the four presented as extended breech, probably due to reduced motility of the fetus due to physical defect. Intrinsic neuromuscular abnormalities of the fetus may prevent the fetus from undergoing normal version, hence the association between breech presentation and fetal abnormalities(3). Only one child is alive and under follow-up.

The abnormalities reported in literature are–popliteal web, toe nail hypoplasia, cleft lip with or without cleft palate and lower lip pits(2). The occasional findings reported are: oral frenula, cutaneous webs between eyelids, intercrural pterygeum, syndactyly, hypoplasia of digits, bifid or absent patellae, scoliosis, cryptorchidism, ambiguous external genitalia and inguinal

**TABLE I**—*Summary of Clinical Features of LMPS*

| Clinical features | Patient number | | | |
|---|---|---|---|---|
| | 1 | 2 | 3 | 4 |
| Age | 7 mo | 30 days | 30 days | 2 mo |
| Sex | M | M | M | M |
| Birth order | 1 | 2 | 1 | 2 |
| Mother's age (yrs) | 19 | 32 | 21 | 29 |
| Father's age (yrs) | 23 | 45 | 29 | 31 |
| Consanguinity | + First cousins | – | + First cousins | – |
| Family history | – | Father's side 3 infant deaths | – | – |
| Antenatal history | – | Attempted abortion with allopathic and native medicine | Treated for urinary tract infection in 2nd month | Treated for fever and respiratory infection (2nd month) |
| Gestation | 30 weeks | Term | Term | Term |
| Presentation | Breech | Breech | Breech | Breech |
| Delivery | Normal vaginal | Assisted vaginal | LSCS | Assisted vaginal |
| Birth weight | AFD | AFD | SFD | SFD |
| Development | Delayed | – | – | – |
| *Physical features* | | | | |
| Webbing | | | | |
| Neck | + | + | – | + (*Fig. 1*) |
| Cubital fossa | + | + | + | – |
| Popliteal | + (*Fig. 3*) | + | + | – |
| Intercrural | – | + | – | + |
| CTEV | – | Bilateral | Bilateral | Bilateral |
| Dimple over the knees | – | + | – | + |
| Dolicocephaly | + | – | – | + |
| Face | Asymmetric | – | – | Asymmetric |
| Ankyloglossia | – | + | – | – |
| Retrognathia | – | – | – | + |
| Nose | – | – | – | Bulbous with wide bridge |
| Low set ears | + | – | – | – |
| Chest | – | – | – | Flat-widely spaced nipples |
| Abdomen | – | Right inguinal hernia | – | Bulging (*Fig. 2*) weak abdominal muscles |
| Phallus | Normal | Small | Normal | Small |
| Testis | Normal | Undescended | Normal | Undescended |

| Clinical features | Patients number | | | |
|---|---|---|---|---|
| | 1 | 2 | 3 | 4 |
| CVS | – | – | – | VSD |
| Respiratory | Infection | Infection | Pulmonary hypoplasia | Infection |
| Spine | Scoliosis | Scoliosis | Scoliosis | Hemivertebrae lower thoracic scoliosis |
| Hip | – | Hypoplasia of ilea | – | Hypoplastic ilea |
| Limbs | Short and thin tibiae | Femora and tibiae small and thin | – | Small femora + Tibiae– Fracture shaft left femur |
| Karyotype | Not done | 46 XY | Not done | 46 XY |
| Management | Orthopedic | Neonatal care-Respiratory infection | Medical management | Respiratory and orthopedic management, cardiology follow-up |
| Outcome | Died at 9 months | Died within a few days | Died on 40th day | On follow -up |

hernia. Our cases had the above features except cleft lip, lip pits and web between eyelids.

This phenotype is not specific but represents an etiologically heterogeneous deformation sequence that can result from a variety of congenital neuropathies and myopathies.

LMPS presents in the neonate with multiple congenital contractures. The existence of LMPS and Lethal Popliteal Pterygeum Syndrome are accepted(4,5). Association with hydranencephaly(6) and progressive nemaline myopathy(7) have been reported. This condition is autosomal recessive in genetic inheritance, with intrafamilial variability and prenatal diagnosis by ultrasound is warranted.

Pena and Shokir described sibs with camptodactyly, club feet, ankylosis of the hips and knees and unusual facies and pulmonary hypoplasia from an uncle-niece marriage(4).

This phenotype has ben listed among fetal 'akinesia sequence' by Graham(8) who has reported a case with multiple pterygea and cystic hygroma which was diagnosed prenatally by ultrasound. The muscle pathology in this case was neurogenic atrophy. The consequence of fetal akinesia may include multiple joint contractures, micrognathia, polyhydramnios, costovertebral anomalies with scoliosis, pulmonary hypoplasia, growth retardation and short umbilical cord(9). The clinical features often include breech presentation with deformities such as micro-retrognathia, facial asymmetry, CTEV, calcaneovalgus and hip dislocation. This fetal akinesia sequence may result from maternal myasthenia, maternal myotonic dystro-

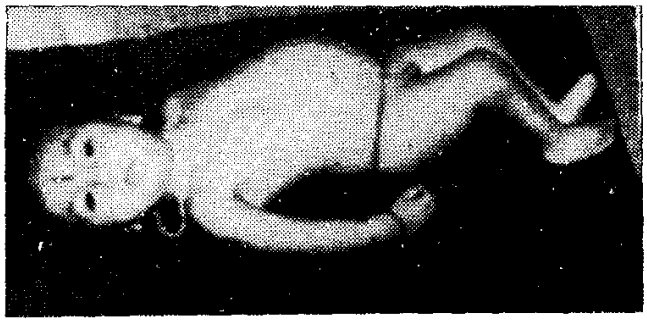

*Fig. 1 Asymmetric face with webbing neck, bulging abdomen intercrural and popliteal webs in Case 4.*

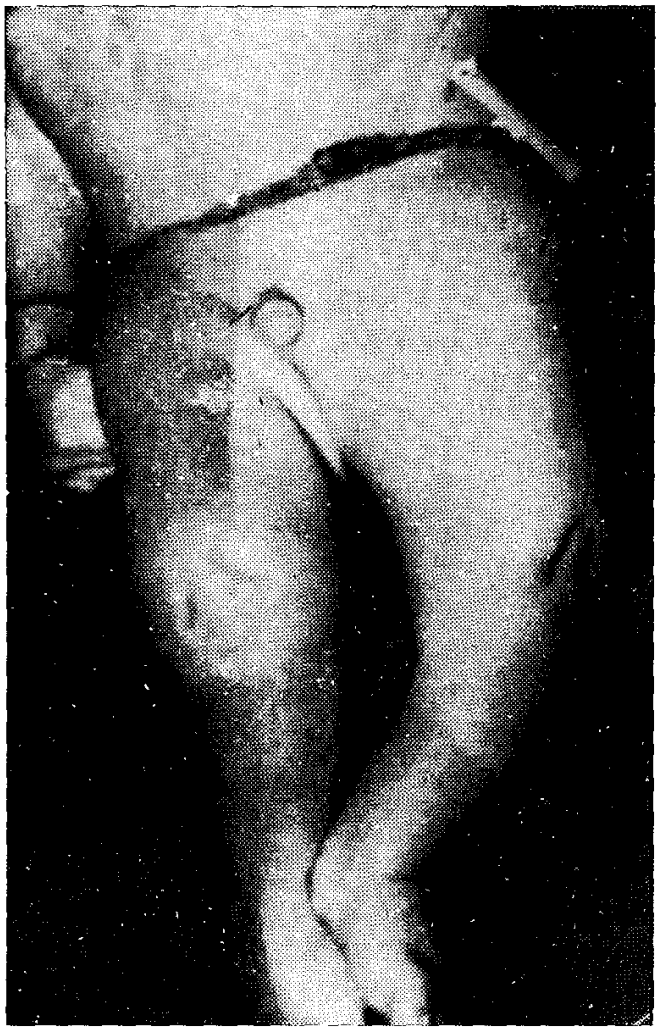

*Fig. 2. Intercrural web, absent scrotal sacs-small phallus, dimple over the knees and left inguinal hernia.*

phy or following tubocurarine therapy in early pregnancy.

The etiologic heterogeneity of this phenotype calls for a thorough history and pedigree recording. Since most infants are nonviable, a complete examination of the neonate is to be made to enable proper genetic counselling. Recurrence risk relates to the basic problem and depends on its mode of inheritance or environmental factor. Prenatal diagnosis is possible with ultrasonography.

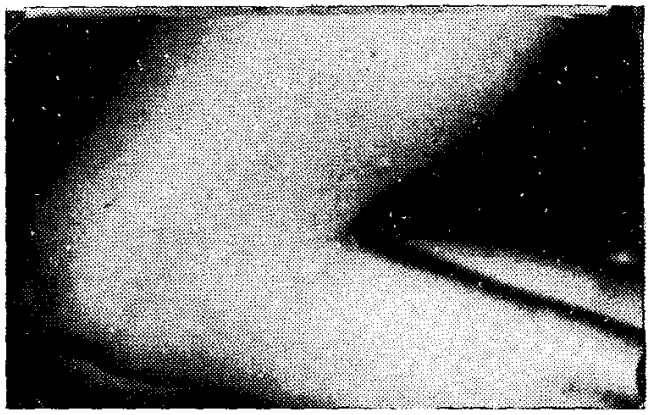

*Fig. 3. Popliteal web in Case 1.*

In the neonate, attempts at oxygenation may be unsuccessful if there is pulmonary hypoplasia. Vigorous orthopedic management with physiotherapy is warranted. Careful releasing of the webs must precede physiotherapy. Hence the importance of recognition of web syndrome from other forms of AMC.

## REFERENCES

1. Hall JG, Reed SD, Rosenbaum KN. Limb Pterygeum syndrome—A review and report of 11 cases. Med Genet 1982, 12: 377-409.
2. Smith DW. Recognizable Pattern of Human Malformation, 2nd edn. Philadelphia, W.B. Saunders 1976, pp 140-141.
3. Braun FRT, Jones KL, Smith DW. Breech presentation as an indication of fetal abnormality. J Pediatr 1975, 86: 419-420.
4. Barstacos CS, Papas CV. Popliteal pterygeum syndrome J Med Genet 1972, 9: 222-226.
5. Papadia F, Zimbalath F, Gactile BC. The

Barstacos papes syndrome. Autosomal recessive form of popliteal pterygeum syndrome in a male infant. Am J Med Genet 1984, 17: 841-847.

6. Mbakop A, Cox JN, Stormann C, Delozier–Blanchet CD. Lethal multiple pterygene syndrome. Report of a new case with hydranencephaly. Am J Med Genet 1986, 25: 575-579.
7. Papadia F, Longo N, Serlengel–Porzio G. Progressive form of multiple pterygeum syndrome in association with nemaline myopathy. Report of a female followed for 12 years. Am J Med Genet 1987, 26: 73-83.
8. Graham J. Fetal akinesia. Sequence. *In:* Recognizable Pattern of Human Deformation, 2nd edn. Ed Smith DW. Phipadelphia, WB Saunders, 1988, pp 108-111.
9. Punnett HH, Kistemmacher M, Valdes, Dpeng M. Syndrome of ankylosis and facial anomalies and pulmonary hypoplasia–A lethal condition. Pediatrics 1974, 85: 375-376.

# Myositis Ossificans Progressiva

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Myositis ossificans progressiva is a rare disease characterized by skeletal abnormalities particularly of toes and fingers and ectopic ossification in connective tissues of the muscles, tendons, fascia and aponeurosis. Since the 17th century, 550 cases have been reported in world literature(1), the largest series being reviewed by Lutwak in 1964(2). In all 12 cases have been reported in the Indian Literature(3-5). We report one such case due to its rarity.

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## Case Report

An eight-year-old girl was referred with the presenting complaints of swellings over preauricular region and neck for two years and inability to move right upper limb for two months. Over a period of two years, similar swellings appeared in the sternocleidomastoid and upper cervical regions, left scapular, right scapular, occipital, paravertebral and right shoulder regions, associated with limitations of movements of various joints. There was no history of trauma, arthritis or any medication. Her perinatal events and subsequent milestones were normal. No other family members were affected.

General examination revealed a short child with a height of 107 cm and weight of 17.5 kg. Her vital signs were normal. There was clinodactyly, hallux valgus and short great toes, the latter having been noticed by the mother since birth. Stony hard swelling of variable sizes (0.5 to 2.5 cm) were observed in all the areas described above. Skin over the swellings was normal. There were marked restrictions of the movements of neck, flexion of hip and opening of mouth. However, there were no feeding difficulties.

Laboratory investigations revealed a normal hemogram and normal serum levels of calcium, phosphorus, alkaline phosphatase and creatinine phosphokinase. Her skeletal survey showed ectopic ossification in paravertebral, subscapular and neck regions (*Fig.*). Muscle biopsy