

# Congenital Coxa Vara, Patella Aplasia and Tarsal Synostosis

## *A New Inherited Syndrome*

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### SUMMARY

A kindred is described with a probably new inherited syndrome, involving bilateral congenital hip dysplasia with coxa vara, patella aplasia and tarsal synostosis. This association is probably inherited as an incomplete dominant trait, apparently as a result of a new mutation.

In this case a single pleiotropic gene might be held responsible for the multiple skeletal defects.

From a bibliographical review, it may be concluded that the nosological association of these multiple defects has never been published so far.

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The subject of this report is a complex association of a congenital pelvic and hip dysplasia, with patella aplasia and tarsal synostosis with oligodactyly of the feet.

This association is probably inherited as an incomplete dominant trait which apparently occurred by new mutation.

In the account that follows, the clinical characteristics of one affected individual are presented. An outline of the radiological symptoms of other family members is also given. Brief mention is also made of some other syndromes associated to hip dysplasia, patella aplasia or tarsal synostosis.

### Personal Observations

We have studied a family (cf. Fig. 1) including:

- 1) One female with a bizarre bilateral pelvic and hip dysplasia, bilateral patella aplasia, and bilateral tarsal synostosis with oligodactyly;
- 2) One female with the same bizarre bilateral pelvic and hip dysplasia, and bilateral patella aplasia;
- 3) One child with bilateral patella aplasia.

STUDY OF THE PROBAND

F. M., married woman, 44 years old, born in Wernrode (Nordhausen, Germany), in 1924. Husband, D. L. G., living in Overboelare (Belgium).

Congenital bilateral hip anomaly. Normal menarche. A spontaneous abortion at 25 years. At 42, pyelonephritis, gastritis and irregular menses were noted. At 43, she was treated for a suspected hypothyroidism. At 44, colitic complaints appeared.

Clinical examination on admission (March, 1968) does not reveal any peculiarity. Nor-

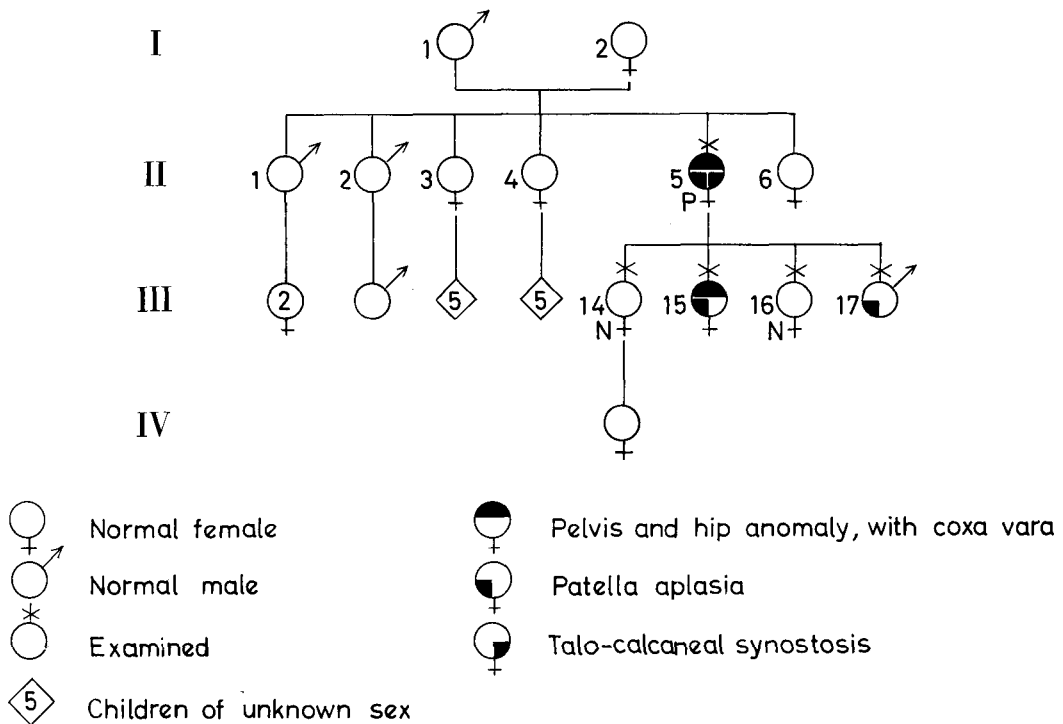


Fig. 1. Familial congenital coxa vara, patella aplasia and tarsal synostosis

mal blood pressure. The motility of the joints is normal. No nail dystrophy. Normal tendinous reflexes. The peripheral arteries are palpable. No peripheral trophic lesions. No signs suggestive of hypothyroidism.

Bizarre dysmorphic habitus. Short stature. Bilaterally, the femora seem to be shortened. Absence of patellae. Flat feet and bilateral hallux valgus.

There are only four toes on the left foot. Five toes on the right foot. She walks with difficulty. The shoulders and the upper extremities are strictly normal. Vitiligo on the skin. A lipoma in the abdominal wall. Somatic features: height 144 cm, weight 66 kg, span 170 cm; vertex to pubis 92 cm; pubis to ground 52 cm.

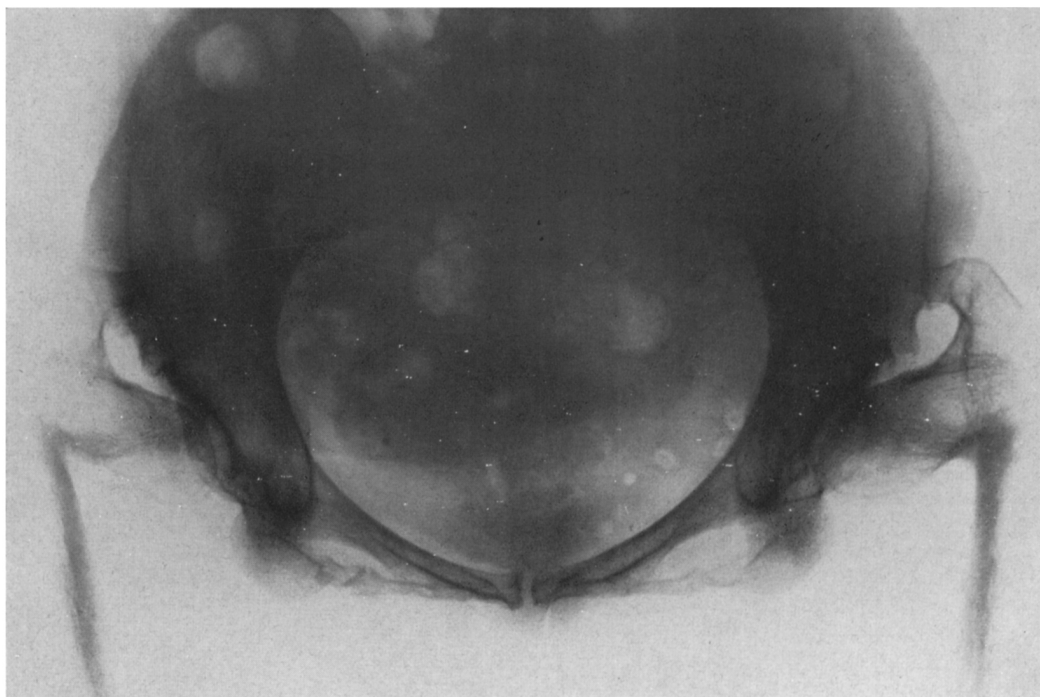


Fig. 2. Bilateral congenital coxa vara («*Hirtenstabform*»). Bilateral hypoplasia of the descending parts of the pubic arches

*Radiographic examinations*<sup>1</sup>

Skull: no abnormalities. Calcification of the falx cerebri.

Thorax: normal.

Lumbar spine: slight scoliosis convex to the right. Slight spondylarthrosis deformans.

Pelvis: bizarre configuration. Bilateral hypoplasia of the descending parts of the pubic arches. Bilaterally, severe atypical dyschondroplastic anomalies of the hips. Extreme coxa vara bilaterally. However, there is no real luxation or subluxation of the coxo-femoral articulation (cf. Fig. 2).

Femora: Moderate bilateral hypoplasia. Femur length, only 32 cm (normal femur length, for a woman of 144 cm height: 39 cm). (cf. Figs. 3 and 4).

Knees: bilateral aplasia of the patellae. Slight osteoporosis (cf. Fig. 5).

Feet: synostosis between the talus and calcaneum, bilaterally. Synostosis between the cuneiformia. Bilaterally, only four metatarsal bones, with bifidity of the fourth metatarsal on the right foot (cf. Figs. 6, 7 and 8).

Hands: normal.

Abdomen: I. V. Pyelography: Cholecystography: no abnormalities.

Gastro-intestinal tract: dolicho-transversum. Severe colitis. No other abnormalities.

<sup>1</sup> By Prof. Van de Velde.

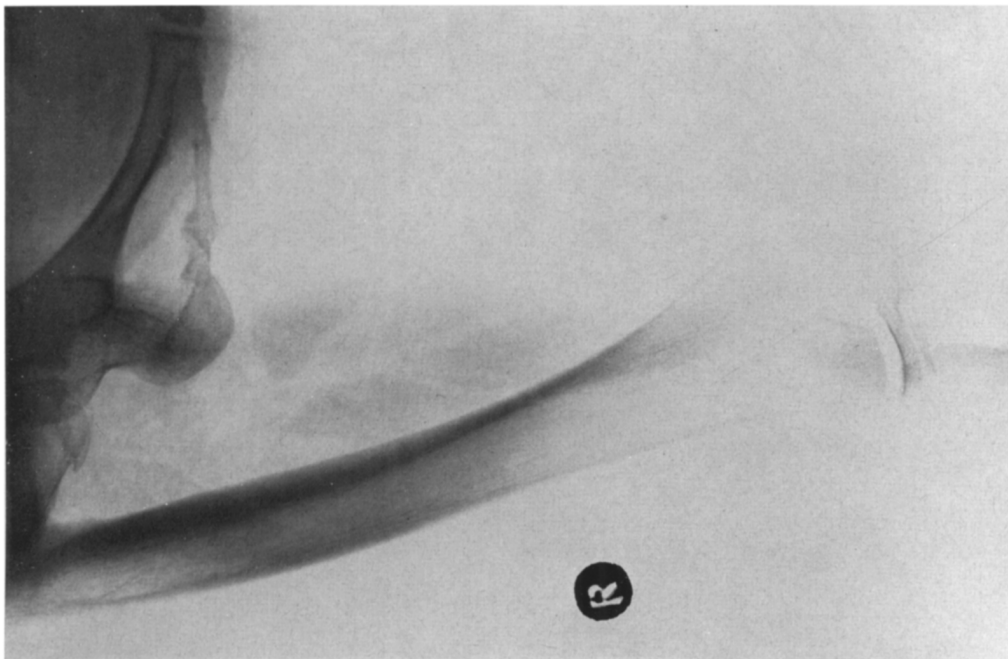


Fig. 4. Slight hypoplasia of right femur



Fig. 3. Slight hypoplasia of left femur



Fig. 5. Bilateral aplasia of the patellae

*Other examinations*

ECG: Supraventricular extrasystoles. Otherwise normal.

Ophthalmological examination: normal vision. Normal eye ground.

Basal metabolic rate: + 6%.

Normal status genitalis.

Dermatoglyphic examination: Normal palms; high total finger ridge count: 229 (113 + 116).

Karyotype: normal.

*Laboratory examinations*

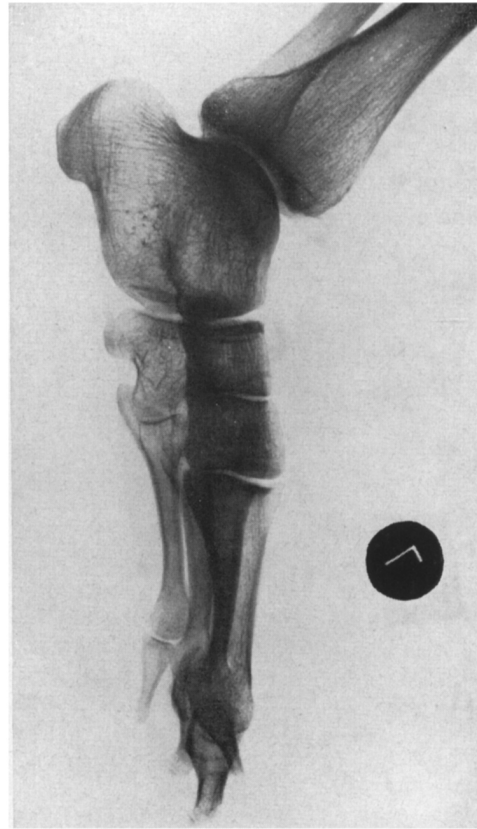
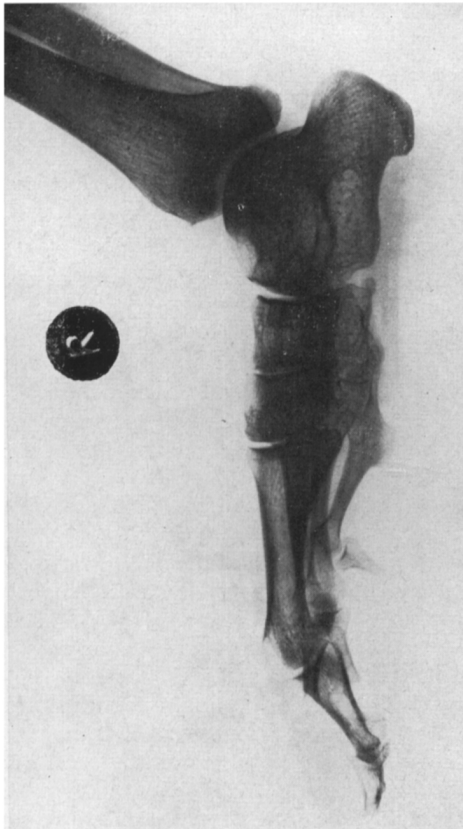
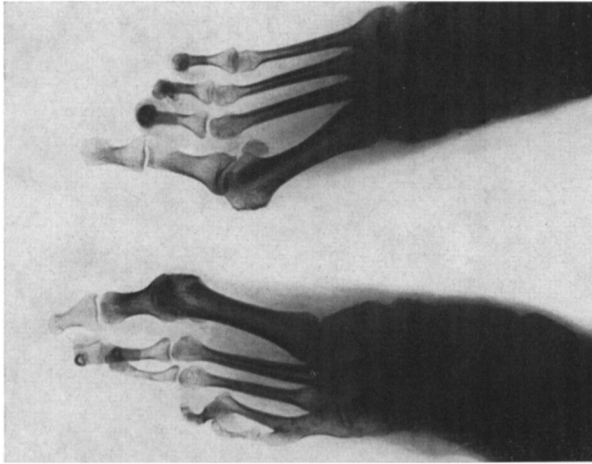
*Urine.* Protein: traces; glucose: negative; urinary sediment: some leucocytes, erythrocytes and epithelial cells. *Proteus mirabilis* (140 000/ml urine). Cultures for BK: negative. Calciuria: 56 mg/24h.

*Blood.* Peripheral blood: normal; sedimentation rate: 6/25 mm.

Glycemia, blood urea, uricemia, kreatininemia: normal. Cholesterolemia: 275 mg/100 ml. Bilirubine, flocculation tests, transaminases and alkaline phosphatases: normal. Total protein and electrophoretic pattern: normal. Calcemia: 9.6 mg/100 ml. Phosphate-mia: 2.8 mg/100 ml.

Phenol red test: 25% excretion after 15 minutes, 65% total excretion after 70 minutes.





Figs. 6, 7 and 8.  
Talo-calcaneal synostosis. Oligometatarsy

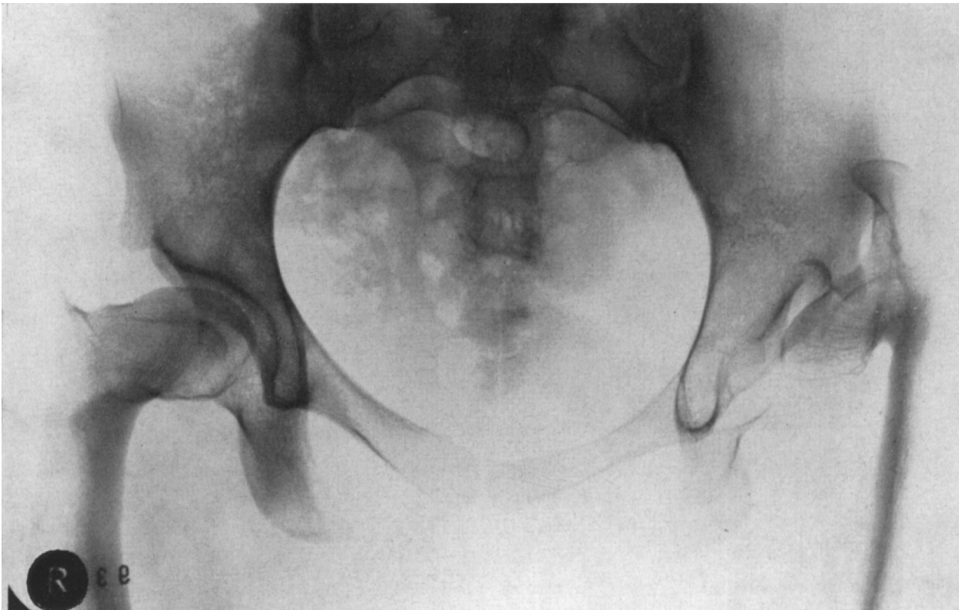


Fig. 9. Bilateral congenital coxa vara

OTHER FAMILY MEMBERS

- I. 1 and 2. F. K. and H. M. Both parents died at the age of 52 from unknown cause.
- II. 1. F. Ew. No somatic abnormalities. Dies at the age of 20. Has two normal daughters (III.1 and III.2).
- II. 2. F. Er., 53 years old. No somatic abnormalities. Has a normal son (III.3).
- II. 3. F. Ed., 50 years old. Acute rheumatic fever when 18. Normal somatotype. Five normal children (III.4 to III.8).
- II. 4. F. Em., 47 years old. Normal. Five normal children (III.9 to III.13).
- II. 5. Proband.
- II. 6. Female child. Died when 7 months old. Nothing else known.
- III. 14. D. L. S., 23 years old. Married woman. Somatic features: height 167 cm, weight 60 kg. Has a male child of 6 months (IV.1). Radiographic examination: pelvis, no abnormalities; knees, normal; normal patellae; feet, no abnormalities.
- III. 15. D. L. J., daughter, 18 years old. Unmarried. Somatic features: height 154 cm, weight 54 kg. Radiographic examination: pelvis, asymmetric aspect; severe dysplasia of both hips, with bilaterally severe coxa vara (cf. Fig. 9). Subluxation of the right hip. Bilateral hypoplasia of the descending parts of the pubic arches. Absent ischiopubic syn-

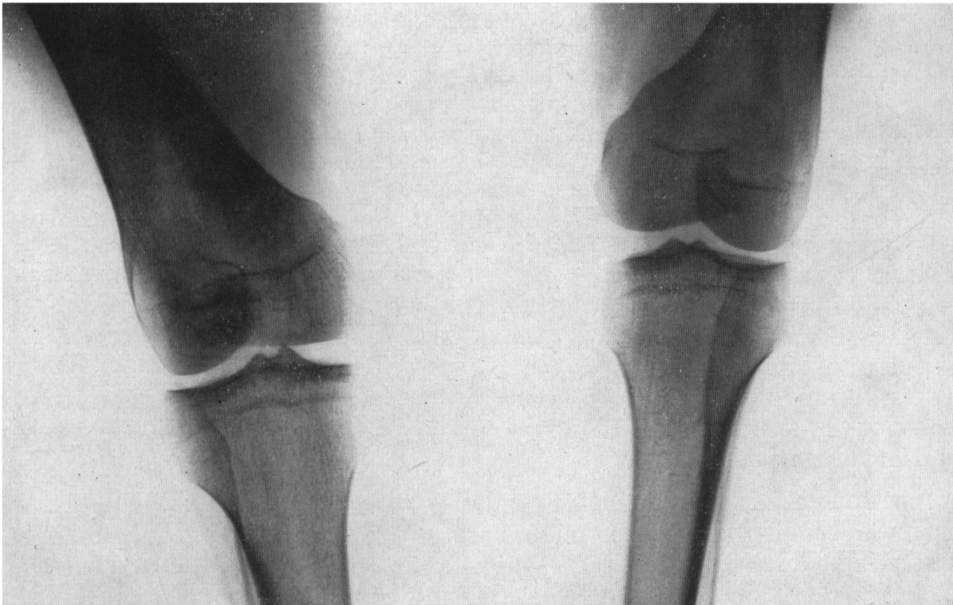


Fig. 10. Bilateral aplasia of the patellae

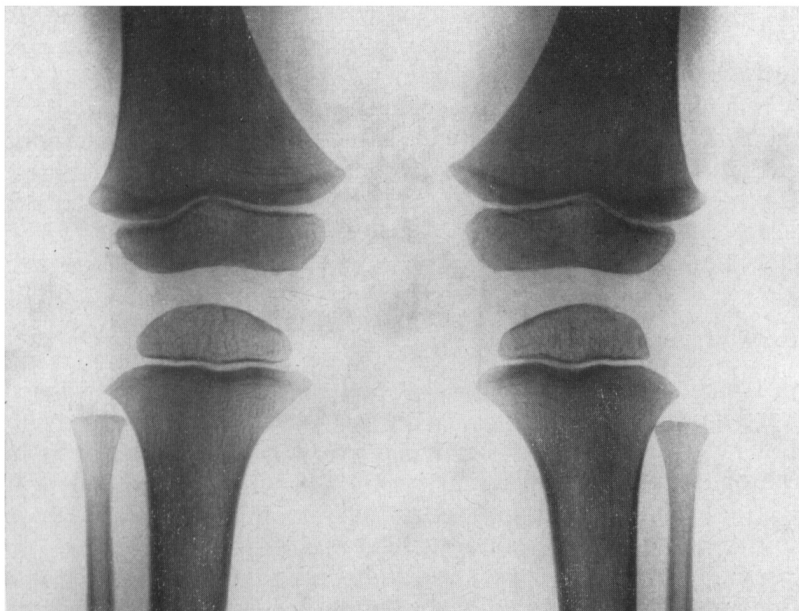


Fig. 11. Bilateral absence of bone nucleus of the patellae



- stosis, bilaterally. Knees, bilateral absence of patellae (cf. Fig. 10). Feet: platypody, arachnodactyly, five metatarsals present, no synostosis.
- III. 16. D. L. C., female child, 14 years old. Somatic features: height 158 cm, weight 47 kg. Radiographic examination: pelvis, knees, feet: normal.
- III. 17. D. L. P., male child, 4 years old. Somatic features: height 109 cm, weight 19 kg. Radiographic examination: pelvis, normal; knees, bilateral absence of the bone nucleus of patellae (cf. Fig. 11), which should normally be visible at this age; feet, no abnormalities.
- IV. 1. Male child, 6 months old. Nothing known.

### Discussion

The syndrome does not consist of one type of lesion only, but is a combination of several kinds of skeletal disorders. It seems to be constituted essentially by the following major abnormalities:

- 1) Bilateral congenital hip dysplasia with coxa vara, hypoplasia of the descending part of the pubic arch and moderate femur hypoplasia;
- 2) Bilateral patella aplasia;
- 3) Bilateral talo-calcaneal synostosis and oligodactyly, due to oligometatarsy.

There are no other somatic nor visceral associated abnormalities.

The proband is born in Wernrode, near Nordhausen, a town in Thuringia, East-Germany, 50 miles W. of Halle. The region was heavily bombed in the second world war, especially on account of rocket research installations. Thus, the proband completely lost all contact with other family members.

In our search for other cases in this region, we sent a questionnaire to four important Medical Services (Internal Medicine, Radiologic Department, Service of Orthopedics, Pediatric Department), of three academic centers in this area (Weimar, Erfurt, Jena). From these twelve services, we received nine replies; however, without positive results.

The major abnormalities will now be discussed briefly.

#### HIP DYSPLASIA, WITH CONGENITAL COXA VARA

The prevalence of this anomaly has been estimated at 1/25 000 live births (Johanning, 1951).

Congenital hip dysplasias may be associated with abnormal development of the ischiadic arch of the pelvis, with femur hypoplasia and congenital foot deformities.

Congenital dislocation of the hip with coxa vara may also occur in cleidocranial dysostosis, and some other rare conditions.

In 55 cases of congenital coxa vara, Johanning (1951) observed 5 cases with genuine hypoplasia of the upper end of the femur, and one case with bilateral absence of the fibulae with a talosynostosis.

Familial observations on dyschondroplastic anomalies with coxa vara with dominant inheritance have been described by Arslanian and Madjalani (1949), Le Mesurier (1951), Johanning (1951) and Almond (1956).

Coxa vara has been described in MZ twins by Greve (1944) and other authors.

#### PATELLA APLASIA

This anomaly was first described in 1820 by Chatelain. The prevalence of patella aplasia as isolated anomaly is unknown. The condition does not seem to be rare, as was thought for time. Little, in 1897, already noted the association with absence of thumb-nails. This entity was further, better individualized, after 1933, as the nail-patella syndrome.

This nail-patella syndrome has five cardinal features:

- 1) Hypoplasia, aplasia and luxation of the patella;
- 2) Iliac horns;
- 3) Dystrophy of the finger- or thumb-nails;
- 4) Malformation of the elbow;
- 5) Renal insufficiency of nephrotic type.

The entity here described is thus entirely different from the nail-patella syndrome. Patella aplasia is sometimes associated with short stature, and also with congenital hypotrichosis.

Patella aplasia, as an isolated anomaly, may occur as a simple autosomal dominant trait.

#### TARSAL SYNOSTOSIS

This rare condition usually occurs only as a sporadic entity, without associated abnormalities (Borgnis, 1952). The talo-calcaneal and calcaneo-navicular forms of tarsal synostosis are more frequent than the talo-navicular forms.

Only some cases may be caused by hereditary factors, since the lesions are symmetrical, bilateral and associated with other congenital anomalies. Familial cases were reported by Bersani and Samilson (1957). Symmetrical carpal and tarsal synostosis have been reported by Geyer (1958), Kewesch (1934), Klapp and Gebhard (1952), Weber (1954).

Familial synostosis in the carpal and tarsal bones was also described by de Vol-dere (1960), and Slaby et al (1964).

Bilateral tarsal synostosis has been described together with dominant hereditary bilateral dysplasia and synostosis of the elbow joint, together with brachymesophalangism, brachymetacarpus and synostosis in fingers and carpus (Fuhrmann et al, 1966). It is suggested that this may represent a distinct hereditary entity.

The association of synostosis of carpalia and tarsalia with symphalangism of the fingers has also been described by some authors.

## References

- ALMOND H. G. (1956). Familial infantile coxa vara. *J. Bone Joint Surg. (Amer.)*, **38**: 539.
- ARSLANIAN J., MAJDALANI E. (1949). *Arch. Franc. Pediat.*, **6**: 100.
- BERSANI F. A., SAMILSON R. L. (1957). Massive familial tarsal synostosis. *J. Bone Joint Surg. (Amer.)*, **39**: 1187.
- BORGNIS G. (1952). Symmetrische Fusswurzelsynostose. *Z. Orthop.*, **81**: 620-627.
- CHATELAIN (1820). Cit. by Touraine, 1955.
- DREWIS J. (1966). Congenital tarsal synostosis. *Radiology*, **6**: 225-31.
- FUHRMANN W., STEFFENS C., ROMPE G. (1966). Dominant erbliche doppelseitige Dysplasie und Synostose des Ellenbogengelenks mit symmetrischer Brachymesophalangie und Brachymetakarpi sowie Synostosen im Finger. *Humangenetik*, **3**: 1-64.
- GEYER E. (1958). Beitrag zu den Synostosenbildungen der Hand und Fusswurzel. *Z. Orthop.*, **90**: 395-408.
- GREBE H. (1944). Coxa vara bei eineiigen Zwillingen. *Arch. Orthop. Unfallchir.*, **43**: 309.
- JOHANNING K. (1951). Coxa vara infantum. *Acta Orthop. Scand.*, **21**: 273.
- KEWESCH E. L. (1934). Über hereditäre Verschmelzung der Hand- und Fusswurzelknocken. *Fortschr. Röntgenstr.*, **50**: 550-563.
- KLAPP B., GEBHARD W. (1952). Symmetrische Synostosenbildung an Hand und Fuss. *Z. Orthop.*, **81**: 637-640.
- LE MESURIER A. (1951). Developmental coxa vara. *J. Bone Joint Surg. (Amer.)*, **33**: 487.
- LITTLE E. M. (1897). Congenital absence or delayed development of the patella. *Lancet*, **2**: 781.
- SCHREIBER A. (1968). Symmetrische Fuss und Handwurzelsynostosen in 4 Generationen. *Z. Orthop.*, **104**: 197-202.
- SLABY O., VESELSKY J., BRYCHTA M. (1964). Familial synostosis of autopodium and a discussion of its causes. *Plzen. lek. Sbor. (Cz)*, **23**: 41.
- SOEUR R. (1959). Le pied plat contracturé. *Rev. Chir. Orthop.*, **45**: 277-289.
- WRAY J., HERNDON C. N. (1963). Hereditary transmission of congenital coalition of the calcaneus to the navicular. *J. Bone Joint Surg. (Amer.)*, **45**: 365.

### RIASSUNTO

Viene descritta una famiglia con una probabilmente nuova sindrome ereditaria, che comporta displasia bilaterale congenita dell'anca con coxa vara, aplasia della rotula e sinostosi del tarso. Questa associazione è probabilmente trasmessa come carattere dominante incompleto, come possibile risultato di una nuova mutazione. In questo caso si potrebbe considerare responsabile dei molteplici difetti scheletrici un singolo gene pleiotropico. Da un esame bibliografico, si può concludere che un'associazione nosologica di questo difetto plurimo finora non è mai stata descritta.

### RÉSUMÉ

Les auteurs décrivent une famille atteinte d'un syndrome héréditaire probablement nouveau, avec dysplasie bilatérale congénitale de la hanche, coxa vara, aplasie de la rotule et synostose du tarse. Cette association est probablement transmise comme un caractère dominant incomplet, probable résultat d'une mutation récente. Dans ce cas, un gène pléiotropique unique pourrait être considéré comme responsable des nombreux défauts du squelette. Sur la base de la littérature, on peut conclure qu'une association nosologique de ce défaut multiple n'a jamais été décrite.

ZUSAMMENFASSUNG

Beschreibung einer Familie mit einem wahrscheinlich neuen Erbsyndrom, das sich in angeborener Dysplasie der Hüften mit Coxa vara, Patella-Aplasie und Synostosis des Tarsus äussert. Dieser Symptomkomplex wird wahrscheinlich als unvollständiges dominierendes Merkmal, möglicherweise als Ergebnis einer neuen Mutation übertragen. In diesem Falle wäre ein einziges pleiotropes Gen für den mannigfaltigen Skelettfehler verantwortlich zu machen. Ein Überblick über das Schrifttum gestattet die Behauptung, dass eine krankhafte Association dieses mehrfachen Defektes bisher noch nie beschrieben wurde.

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