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MALFORMATIO CONGENITA ARTICULI TALO-CRURALIS

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Congenital rigid flat foot (CRF) is a very uncommon condition diagnosed even more rarely in the newborn and infants. In some cases the diagnosis is first made when the child begins to stand. The reason for this relatively late diagnosis is that flat feet in babies is a frequent finding and only seldom requires treatment. This holds true only for the statical group of plano-valgus feet, which in children under three years of age may be considered as physiological. These exhibit free movement in both the ankle and subtalar joints.

CRF in the newborn is a serious deformity which is very difficult to treat. The diagnosis must be made as early as possible, preferably within the first weeks of life since a good result relies on immediate commencement of treatment (Giannestras 1967). These children have free movements in the ankle joint, but their subtalar joints are fixed.

In the literature studied on CRF it has been shown that these children have a vertical talus with dislocation of the talo-navicular joint and prominence of the head of the talus into the sole of the foot medially. The calcaneus is pulled up posteriorly, the metatarsus is dorsiflexed and the foot is in a position of valgus deviation. The angle between the talus and the calcaneus viewed from the side (tcs-angle) is large, and the angle between the same two bones viewed anteriorly (tca-angle) is large too. There is no alteration in the talo-crural joint.

One can reasonably understand why CRF is also called backward clubfoot when these deformities are compared with those of the congenital equinovarus deformity where the tcs and tca deviations are small, and where the head of the talus projects more or less laterally in comparision with the navicular bone (Osmond-Clarke 1956).

In some children with congenital clubfoot Fjeldborg (1971) has found a slight widening of the medial malleolus, but otherwise no abnormalities of the talo-crural joint. In other cases of congenital clubfoot in children Hjelmstedt (1973) has described differing flat-

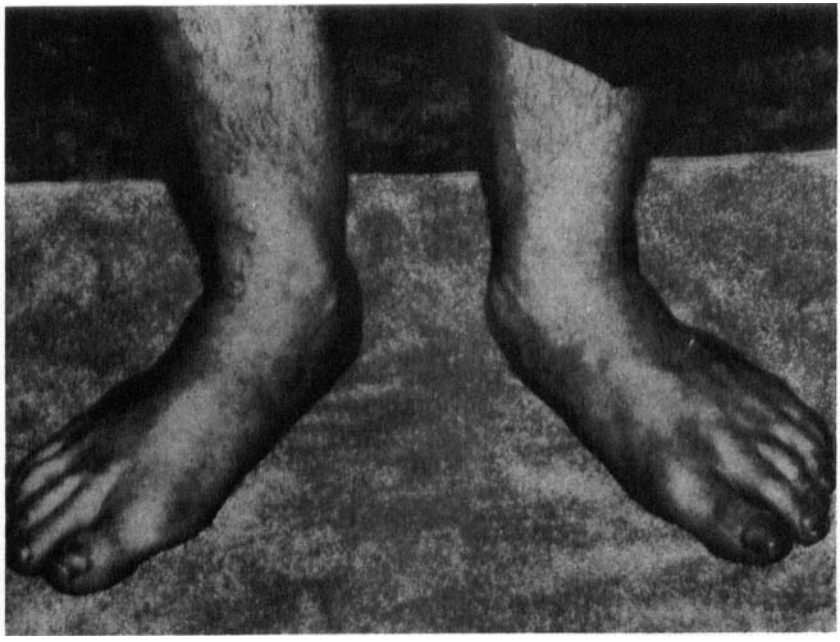

Figure 1. The propositus's feet from the front.

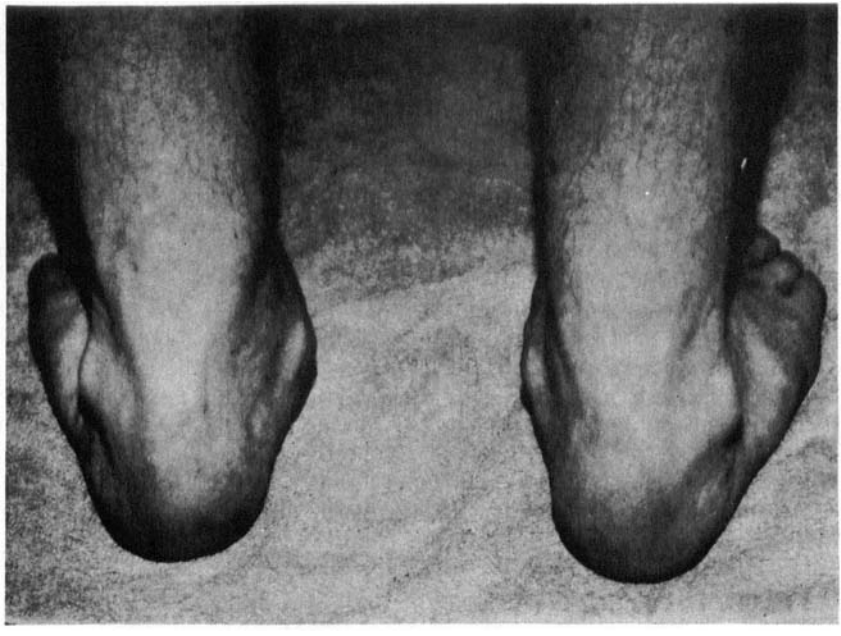

Figure 2. The propositus's feet from behind.

tening of the trochlea tali, obliteration of the ankle joint's recesses with reduction of the trochlear joint surface, and finally changes in the medial and lateral facets of the talus.

In a Danish family, I have found CRF combined with a previously undescribed congenitally inherited deformity of the bones which make up the ankle joint. Changes are found mainly in the ankle joint, but the joints between talus and calcaneus, and the joint between talus and the navicular bone are affected too in several cases.

In the following, the characteristic clinical and radiological appearances of the deformity are described, and it is shown how it presents itself at different ages. Finally, a family tree is shown. This has been worked out after personal reference to all surviving members of the family.

CASE REPORTS

The propositus (III, 22) was born on 4/7 1949 and prior to his presenting symptoms he had been a fit farmworker. At 23 years of age he was referred to the orthopaedic department in Aalborg because of weightbearing pain in both feet over many years. He is the third of four children. The mother had been well during the pregnancy and took no medicines. His parents are related only by marriage.

At the objective examination it was found that the patient's gait was normal. His feet were flat both with and without weightbearing. The toes were normal. The lateral malleolus was prominent. There was an obvious prominence on the inside of the foot just distal to the level of the medial malleolus (Figures 1 and 2). Movements in the talo-crural and subtalar joints were much reduced. With both straight and flexed knee there was only oscillatory movement around the neutral position in the talo-crural joint, whilst there was only a few degrees of pronation and supination in the subtalar joints. Clinically, there was consequently a fixed flat foot with a practically stiff ankle joint.

No other congenital abnormalities were found, especially in the bones and joints. The patient was mentally normal.

X-rays of the feet show deformities of the tibia, fibula and talus with the following changes in the ankle and subtalar joints (Figures 3 and 4). In the A-P projection, the tibia appears short compared to the fibula, the medial malleolus is greatly hypoplastic with the whole of the vertical portion missing. The lateral malleolus is thickened thus protruding to a considerable extent both medially and laterally. The talus is widened medially and the vertical joint surface which normally articulates with the medial malleolus is absent. Laterally the talus is excavated corresponding to the medial prominence of the lateral malleolus. The ankle joint seen frontally is thus changed from the normal hinge to a joint composed of a horizontal medial portion and a U-formed vertical lateral portion.

From the side, the distal extremity of the tibia is widened posteriorly by a bony process and the normal concave joint surface is changed to an anterior concave with a posterior convex curve stretching completely to the posterior edge of the tibia.

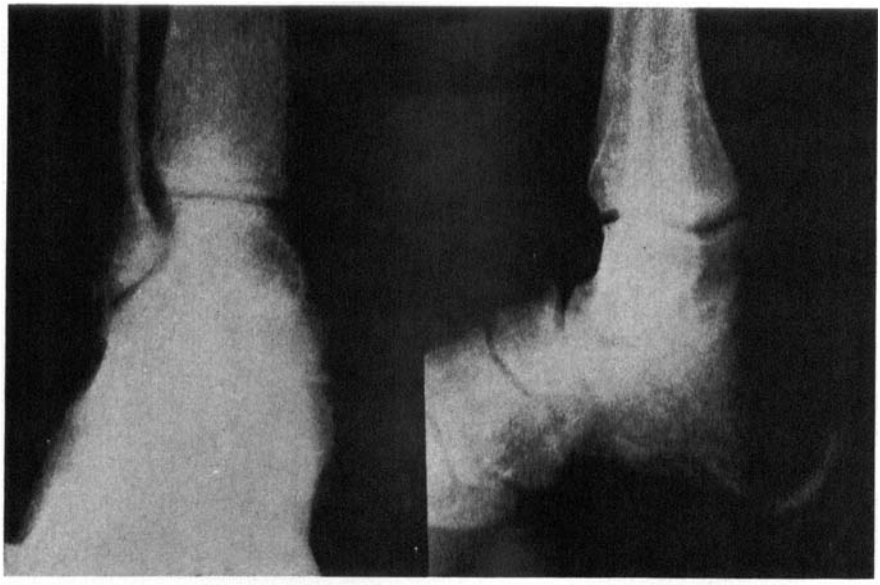

Figure 3. Right ankle and hind foot of the propositus (III, 22).

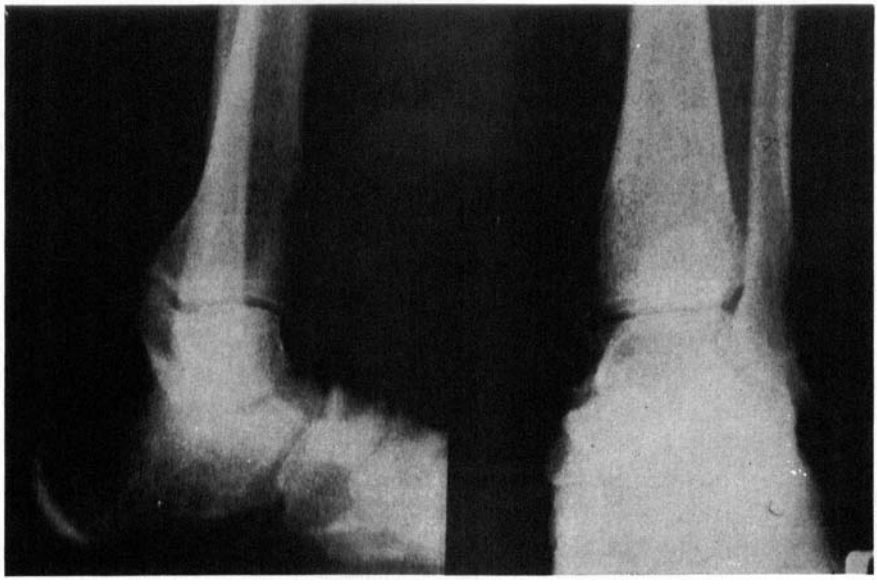

Figure 4. Left ankle and hind foot of the propositus (III, 22).

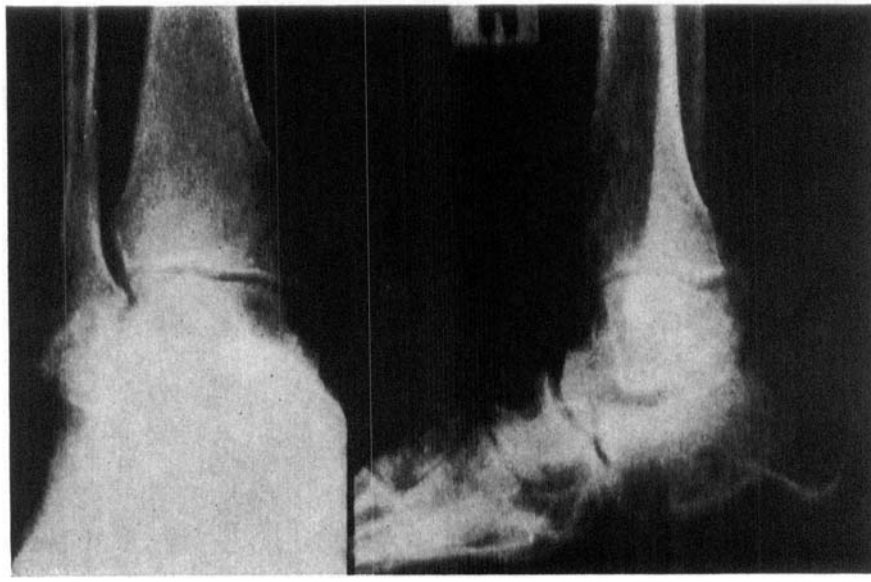

Figure 5. Right ankle and hind foot of the propositus's mother (II, 9).

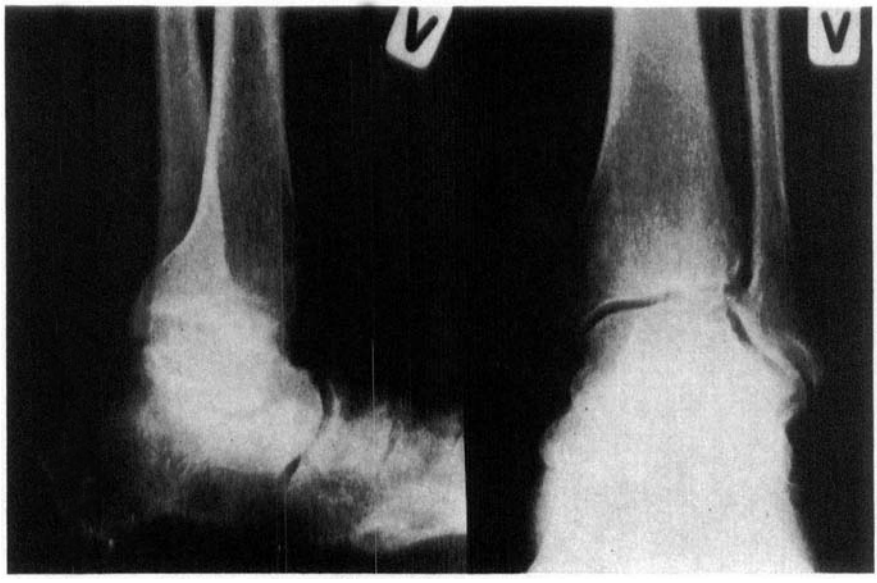

Figure 6. Left ankle and hind foot of the propositus's mother (II, 9).

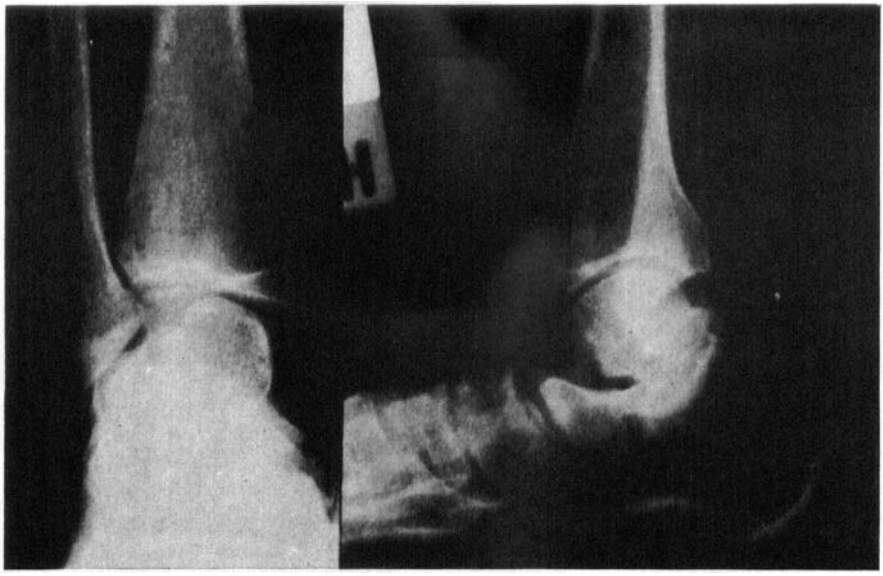

Figure 7. Right ankle and hind foot of the propositus's sister (III, 24) at the age of 19 years.

Both the body and neck of talus are deformed, the body being high and short whilst the posterior process of the talus is large and solidly combined to the body. Proximally the joint surface is curved corresponding to the tibial surface and extending completely out onto the posterior process. The neck of the talus is also short and the talus stands upright but not vertically. There is no dislocation between talus and the navicular bone. The joints between talus and calcaneus are not clearly seen, probably due to the medial and distal displacement of the talus compared to the calcaneus. Extra projections plus tomography were taken which were unsuccessful in showing a clear picture of the subtalar joints. It is unlikely that a bony bridge exists between the two bones and there are definitely no bony bridges between other tarsal bones.

X-rays of the lumbar spine, the pelvis, the hips, femora, the knees and hands were taken, all of which showed normal structure. Chromosomal studies were normal.

The propositus's mother (II, 9) was born on 25/7 1915 and was, in her youth, examined in the orthopaedic department because of symptomatic flat feet. No X-rays were taken at that time. At 58 years of age she was again referred to the department for examination because of weightbearing pain in her feet. The symptoms were so severe that the patient had in fact applied for a full pension. She has been a good observer of the family's defect, and she knew that her father had deformed feet of the same type as herself and other members of the family.

With clinical examination it was found that both feet were flat and fixed and the ankle joints were practically stiff like the propositus. Figures 5 and 6 show

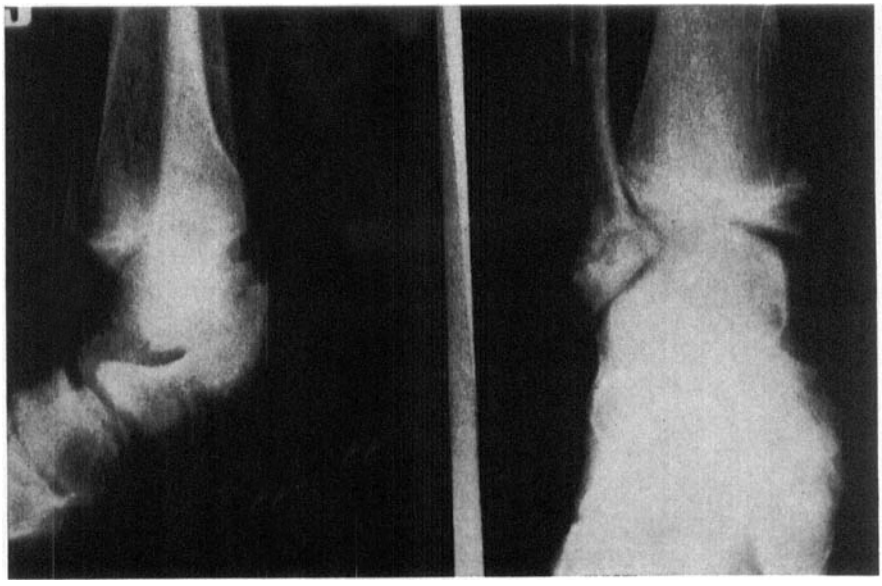

Figure 8. Right ankle and hind foot of the propositus's sister (III, 24) at the age of 13 years.

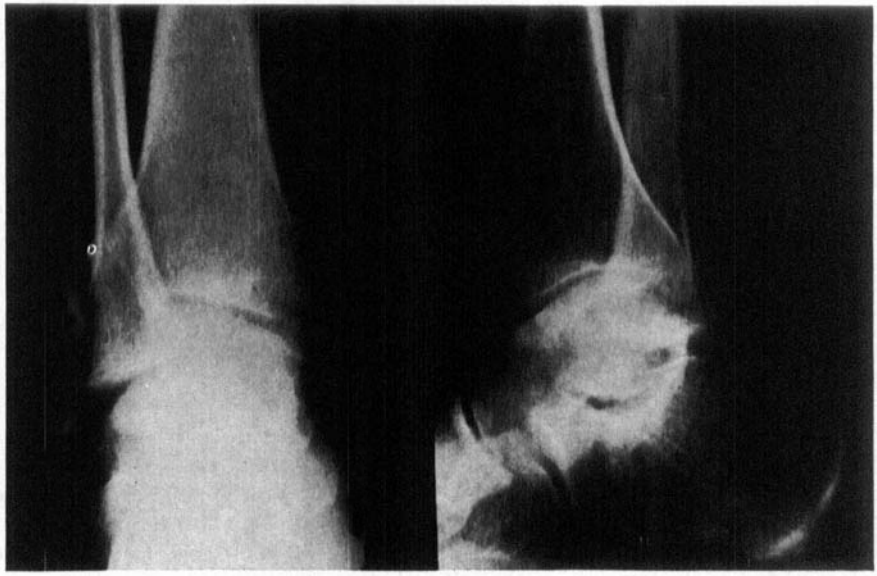

Figure 9. Right ankle and hind foot of the propositus's cousin (III, 1) at the age of 32 years.

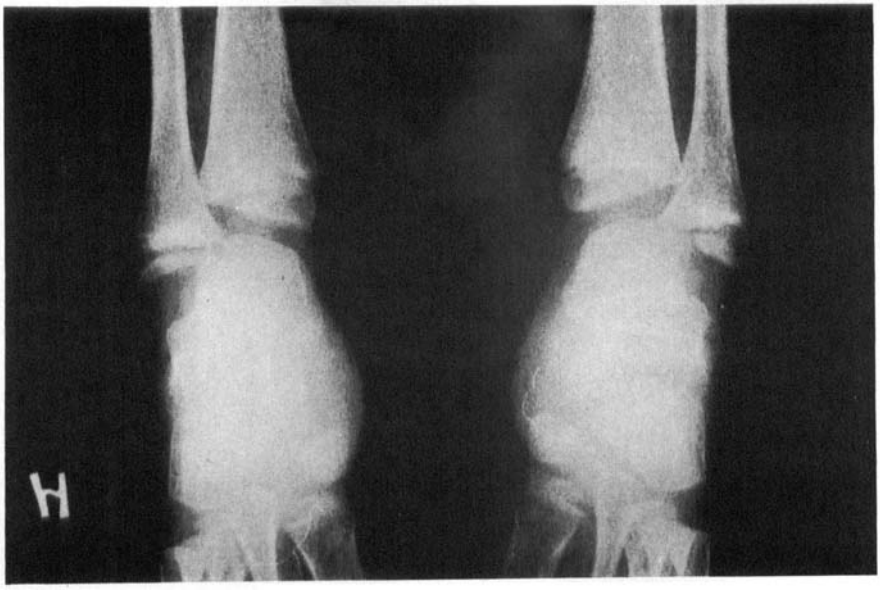

Figure 10. Right and left ankle and foot of the propositus's cousin (III, 1) at the age of 6 years.

symmetrical changes of the talus, and in and around the ankle and subtalar joints. The changes were the same as her son's but less marked. The upright talus with the short neck was clearly seen. The subtalar joints were shown better than in the propositus. There were some slight arthritic changes in the talo-crural joints but not in the subtalar joints. Heel spurs were seen on the calcaneal tuberosities.

The propositus's sister (III, 24) was born on 14/1 1951. She had both clinically and radiologically the same type of changes as her mother and brother, but here again the changes were less marked than in the propositus (Figure 7). X-rays were taken when this patient was 13 years old. The epiphyseal lines and centres are clearly seen (Figure 8). In the A-P projection the tibial centre of ossification is hour-glass shaped. The fibular epiphysis is wedge-shaped with the base lateral. In the lateral projection the tibial epiphysis is at the anterior tibial edge where it is seen to be wedge-shaped with the base anterior. The wide talus is seen to project proximally to a considerable degree in the central part of the ankle joint.

The last patient to be described is a male cousin of the propositus (III, 1); he was born on 4/4 1931. At the time of examination he was 38 years of age and had typical clinical and radiological changes in both feet (Figure 9). He was X-rayed at 6 years of age revealing clear radiological abnormalities of both the distal tibial and fibular epiphyses (Figure 10). Both ossification centres are wedge-shaped with the base to the periphery. The wide talus and the short tibia are also seen. No lateral views were taken at the same time.

Figure 11 shows the family tree. There are without doubt ten people possessing the described deformity. The degree of manifestation varies within very narrow

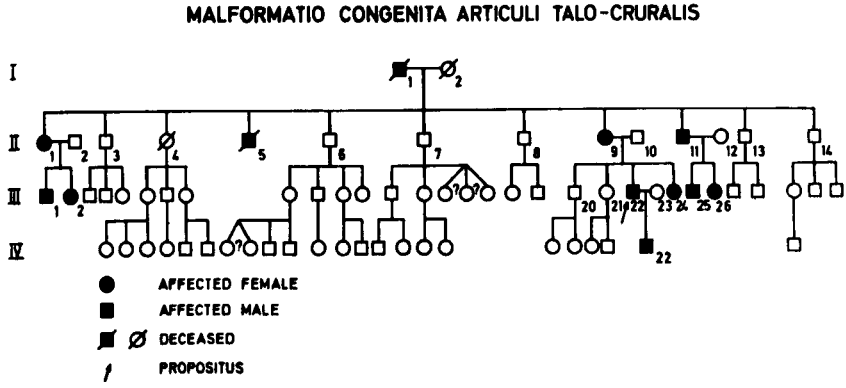

Figure 11. Pedigree.

limits. The pattern of the family tree suggests that the condition is an autosomal dominantly inherited abnormality.

DISCUSSION

This inherited abnormality is presumably caused by a genetic mutation. The time at which the mutation took place cannot be determined and the cause cannot definitely be established.

After careful study of the X-rays in Figures 8 and 9, where the epiphyseal lines and centres are distinct, one can formulate a hypothesis for the mechanism in the development of the anomaly. It could be suggested that the high, wide, short, and erect talus is of primary consideration in causing the abnormality. It can be postulated that the increased talus height can mechanically impede the growth of the distal tibial epiphysis thus explaining the relatively short tibia. The wide talus, which is most prominent medially presumably presses the medial malleolus bud medially and proximally, thereby mechanically hindering the normal development of the malleolus. The short tibia allows for medial expansion of the lateral malleolus bud, thus causing an indentation on the lateral side of the talus, the counterpressure of which could interfere with the fibular epiphysis. The high talus can exert greatest pressure on the tibial epiphysis posteriorly, thus stopping the longitudinal growth earlier posteriorly than anteriorly. At the same time the posterior tibial edge will be pressed backwards. In patients with a large posterior process attached to the talus one can imagine that at an early stage of development of the foetus a kind of nearthron is developed posteriorly between the talus and tibia.

In this way the normal hinge joint is converted to a saddle joint (Figures 3 and 4). In the patients without this posterior process there is no possibility for this joint formation to occur (Figure 7).

It is clear that the severest and most obvious changes are found in the talo-crural joints, suggesting that the cause of the faulty development is a primary dysplasia of the distal tibial and fibular epiphyseal lines and centres. The cause of such a primary dysplasia is not known, but in this connection it could be mentioned that the hereditary process for this deformity is the same as for chondrodystrophia foetalis.

Finally, it is possible that the development of the ankle joint and talus as a whole is primarily abnormal so that there are, from the beginning, anatomical abnormalities in the talus, the tibia, and the fibula.

The X-rays of the feet and the ankle joints give a rational explanation of the limited movements in the talo-crural and subtalar joints. The erect talus explains the flat feet.

SUMMARY

After a short introduction on flat feet in the newborn, clinical and radiological description is given of a previously undescribed congenitally inherited deformity in and around the talo-crural joints. Clinically there is a rigid flat foot with obvious limitation of movement in the ankle joints. Radiologically there are changes in the talus, the tibia and the fibula. The aetiology may be considered as a gene mutation. The pathogenesis is discussed. It can be a consequence of a mechanical influence on the distal epiphyseal centres of the tibia and fibula, due to a primarily pathological centre of ossification of the talus together with an abnormally positioned talus with regard to the positions of the calcaneus and the navicular bone. It may also be due to a primary dysplasia of the distal tibial and fibular centres of ossification. Finally, the condition could be due to a combination of these two mechanisms.

The deformity seems to be an autosomally, dominantly inherited defect.

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