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FAMILIAL INFANTILE OSTEOCHONDROSIS DEFORMANS TIBIAE. IDIOPATHIC TIBIA VARA

A Case Report

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The angulation deformity of the tibia due to an idiopathic growth disturbance of the medial part of the proximal tibial epiphyseal cartilage and epiphysis is a well known pathological entity. This condition, described by *Erlacher* (1922) has been reported in isolated cases (*McCurdy* 1922, *Mau* 1924, *Nilsonne* 1929, *F. Langenskiöld* 1929, *Blount* 1930, among many others) or large series (*A. Langenskiöld* 1952, *Michail et al.* 1959, *de Moraes & Perricone* 1959, *Golding & McNeil-Smith* 1963) by several authors. In 1937, *Blount* gave an extensive account of this localized growth disturbance, and he distinguished an *infantile* and an *adolescent* type of the disease. He further proposed the term "osteochondrosis deformans tibiae", and he presented the first description of the pathology of the disturbed function of the medial part of the epiphyseal cartilage. *A. Langenskiöld & Riska* published recently (1964) the most extensive series of osteochondrosis deformans tibiae, by giving an account of 71 cases observed and treated by them. This condition is by no means uncommon, and except about 170 cases published prior to 1964 (*Langenskiöld & Riska* 1964), there are, no doubt, several other unpublished cases. The etiology of the osteochondrosis deformans tibiae is largely unknown. It was first believed that the condition represents an atypical form of rickets (*Hass* 1934) or epiphysitis of the Perthes type (*McCurdy* 1922, *F. Langenskiöld* 1929, *Barber* 1942), but also infection (*Valentin* 1922, *Lülsdorf* 1931) dyschrondroplasia (*Sloane et al.* 1936), alteration of the lines of force on the medial portion of the upper tibial epiphyseal line (*Golding & McNeil-Smith* 1936) and congenital disturbance of the ossification (*Rocher &*

Figure 1. X-rays films of the knee region of case no. 1.

Roudil 1930) have been considered as etiological factors of this deformity. *Nilsonne* (1929) was the first to recognize the idiopathic character of this deformity. Genetical factors are known to be responsible for congenital or acquired skeletal deformities of different types and angulation at the level of a bony spur is frequently observed with hereditary deforming cartilaginous exostoses, which may cause some confusion between that condition and the idiopathic tibia vara (*Blount* 1937). A racial tendency of the condition has also been maintained by *Bateson* (1966) based on the fact of the large series of *Langenskiöld & Riska* (1964) in a Finnish and that of *Golding & McNeil-Smith* (1963) in a Jamaican population.

The authors have recently observed an idiopathic infantile osteochondrosis deformans of the tibia in 4 members of the same family, which have been treated in our Department. A study of these cases from the genetic viewpoint was considered of interest, and a short account of these cases is presented here.

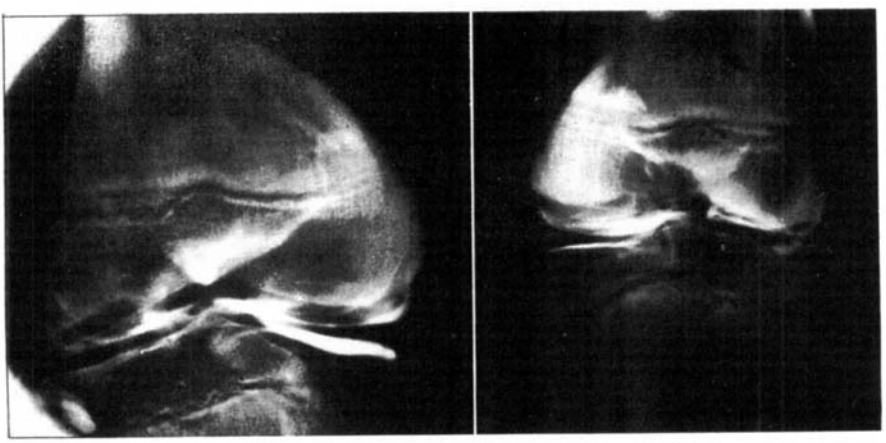
THE CASES

Case Nos. 1 and 2. Two seven years old boys, identical twins, were submitted to the out patient department for bow legs. The boys were born by normal delivery and were healthy earlier. Already at the age of 2 yeears they were examined by the pediatrician for bow legs, but expectancy was suggested until the age of 6 or 7 years. Leg deformity increased, however, considerably during these years. An elder sister of the patients had been operated earlier in the Department for bow legs (case no. 3). The clinical and x-ray (Figures 1 and 2) examination showed pro-

Figure 2. X-rays films of the knee region of case no. 2.

nounced changes characteristic of bilateral idiopathic tibia vara, and the patients were admitted to the Department for operative treatment. Arthrography of the right knee in both patients was performed before the operation, and the arthrograms disclosed almost normal conditions in both cases (Figure 3). A correction osteotomy was performed on the right tibia, and one month later the left tibia was operated on in the same way. A satisfactory correction of the deformities was obtained primarily with healing of the osteotomies after immobilization in plaster within approximately 3 months.

Case No. 3. A girl, 12 years old, sister to the boys of case nos. 1 and 2. Tibia osteotomy was performed on the left leg elsewhere at the age of 4 years because of a pronounced varus deformity. Eight years later the deformity of the leg was still



a *b*

Figure 3. Left knee arthrography of the patients of case no. 1 (a) and no. 2 (b).

Figure 4. X-rays films of the knee region of case no. 3.

pronounced, and a varus deformity had developed on the right leg too. The patient was, therefore, submitted to the Orthopaedic Department. The x-ray examination disclosed a bilateral idiopathic tibia vara (Figure 4). An osteotomy of the left tibia combined with a Femister's epiphyseodesis of the lateral aspect of the proximal epiphyseal cartilage of the left tibia and fibula was performed. Six months later a lateral epiphyseodesis of the tibia and fibula was performed on the right side. A satisfactory clinical and X-ray result was obtained 2 years later. The patient is new 22 years old, married and has two healthy children. There is no deformity of the legs and the patient has no trouble from her knee at the present time.

Figure 5. X-rays films of the knee region of case no. 4.

Case No. 4. Concerned a 10 year old boy, brother of the earlier patients. No deformity of the tibiae was observed by the parents or during the routine medical examination at school. In connection with the radiological examination of the members of the family a slight idiopathic osteochondrosis of both the tibiae could be diagnosed (Figure 5). The clinical examination revealed a slight varus deformity of the tibiae needing no surgical treatment at the present time.

DISCUSSION

The reported cases provide good evidence for the occurrence of idiopathic tibia vara in these 4 members of the same family. The X-ray examination of the knees in the other members of the family, the pedigree of which is demonstrated in Figure 6, disclosed no deformities of the tibia in any of them. The familial occurrence of the deformity strongly suggests that it is under genetic control. The absence of manifestations in the parents makes a dominant transmission unlikely. The observed distribution of unaffected: affected in the sibship 2: 4 (or rather 2: 3 since the affected identical twins should be counted as a single genetic event) is compatible with a recessive inheritance. However, until further pedigree material has been collected no definite conclusions concerning the mode of inheritance can be drawn.

No case of idiopathic tibia vara with genetic transmission has been reported earlier. In the publication of 3 cases by *Evensen & Steffensen* (1957), two of the cases involved cousins, but no genetic analysis was performed by these authors. Medial tibial torsion of a hereditary type, resulting in varus deformity of the legs, has been reported by *Blumel et al.* (1957), but the reported cases had neither the clinical nor the X-ray characteristics of the idiopathic tibia vara. Familial osteochondritis dissecans of the knee with associated tibia vara has also been reported by *Tobin* (1957), but in this publication the clinical reports and the X-rays did not suggest the idiopathic tibia vara.

An interesting point in the present cases was the normal appearance of the knee and of the tibia epiphysis in arthrographies, in the first two cases. *Evensen & Steffensen* concluded from arthrography studies of

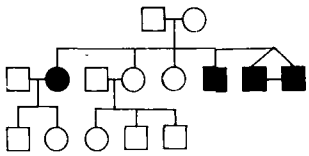


Figure 6. Pedigree of the family. Affected individuals are indicated by black symbols.

their cases that a deformation of both the osseous and the cartilaginous elements of the medial part of the tibial epiphysis occurs in idiopathic tibia vara. This observation is in disagreement with the concept of *Blount* (1937) about the shape of the proximal end af the tibia in this condition. The arthrography findings in our cases (Figure 3) support the view of *Blount* and are in disagreement with *Evensen & Steffensen*'s statement. An explanation for the divergence of findings in this respect might be sought in the development of the proximal part of the tibia in this condition with progressing age as it is demonstrated schematically by *Langenskiöld & Riska* (1964). It seems that a deformation of shape of the entire medial tibial condyle may occur in cases of infantile idiopathic tibia vara with prolonged development.

SUMMARY

Four cases of idiopathic osteochondrosis deformans of the tibia were observed in the same sibship. Two of the affected individuals were identical twins. These observations suggest that this localized growth disturbance is under genetic control. This localized skeletal growth disturbance occurred in this family probably as a result of the above, though at present the mode of inheritance cannot be determined with certainty.

RESUME

Quatre cas d'ostéochondrose idiopathique déformante du tibia ont été observés dans la même famille. Deux des sujets étaient des jumeaux vrais. Ces observations font supposer que de trouble localisé de la croissance est sous contrôle génétique. Ce trouble squelettique localisé de la croissance est probablement héréditaire bien qu'il ne soit pas possible de déterminer avec certitude la forme de cette hérédité.

ZUSAMMENFASSUNG

Vier Fälle von ideopathischer Osteochondrosis deformans der Tibia wurden in derselben Verwandtschaft beobachtet. Zwei der ergriffenen Personen waren identische Zwillinge. Diese Beobachtungen legen es nahe, dass diese örtliche Wachtumsstörung unter genetischer Kontrolle vor sich geht. Diese lokalisierte Wachstumsstörung des Skelettes entstand in dieser Familie wahrscheinlich als ein Erngebnis dieser Kontrolle, obwohl zur Zeit die Art der Vererbung nicht mit Sicherheit festgestellt werden kann.

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