

MADELUNG'S DEFORMITY ITS ETIOLOGY AND PATHOGENESIS

By

BERNHARD PAUS

Oslo

The genuine Madelung's deformity occurs spontaneously; a "symptomatic" form may arise after fracture or disease in the lower end of radius. The cause of the genuine Madelung's deformity is supposed to be either 1) rachitis, 2) constantly recurring injury, for instance, because of heavy work, or 3) heredity. As regards rachitis, no rachitic symptoms were present in any of my cases. With respect to heavy work, I have seen the deformity in two 6 year old children. They cannot, in all likelihood, have been subjected to the above type of injury.

Heredity or familial occurrence is mentioned in about 40 per cent of the cases hitherto published. Here in Norway *Solberg* (1905) has described the deformity in two brothers, and *Bergan* (1942) in three sisters. Recurrence throughout three generations is reported in several cases.

In 1942 I gave an account of a Norwegian family of farmers in which at least 27 cases of Madelung's deformity had occurred in the course of six generations. The investigation was carried out in 1940. On partial follow-up of the family 10 years later, it was found that three more cases had occurred, so that the total number is now 30. Respecting 5 members, who were not examined by me, conflicting information has been given and they are entered as uncertain cases, although it is most probable that they were deformed. I should here like to mention that I have always tried to obtain information concerning each family member from several sources. In many cases I have checked by personal examination the information received.

Usually a preponderance of females is found among the deformed persons, but in this family the proportion was 21 males to 9 females.

(Paper read at the meeting of the Norwegian Society of Surgeons 20/5 1950).



Fig. 1.

Madelung's deformity in a 76 year old man.

This preponderance of males is not due to a corresponding surplus of males in the family as a whole.

In the whole group of families, fig. 2, it is seen that there are 15 in which one of the parents had the deformity. In 10 of these the deformity is found in the offspring, while in 4 we cannot exclude the possibility of its presence or of its development. For the fact is, that the deformity may be difficult to detect in children, and in these 4 families the children either died in infancy or are still small children. There then remains only 1 family of the 15 of which it can be said with certainty that, although one of the parents was deformed, none of the offspring had the deformity. In this family, however, there were only two children. On the other hand, there are about 40 families where both parents are normal. In none of these has the deformity been observed among the offspring. The deformity does not skip a single generation. Only a dominant, autosomal, non-sex-linked hereditary factor can explain this mode of occurrence.

The pathogenesis of the deformity is usually said to be a premature fusion of the volar-ulnar part of the distal epiphyseal disc of the radius

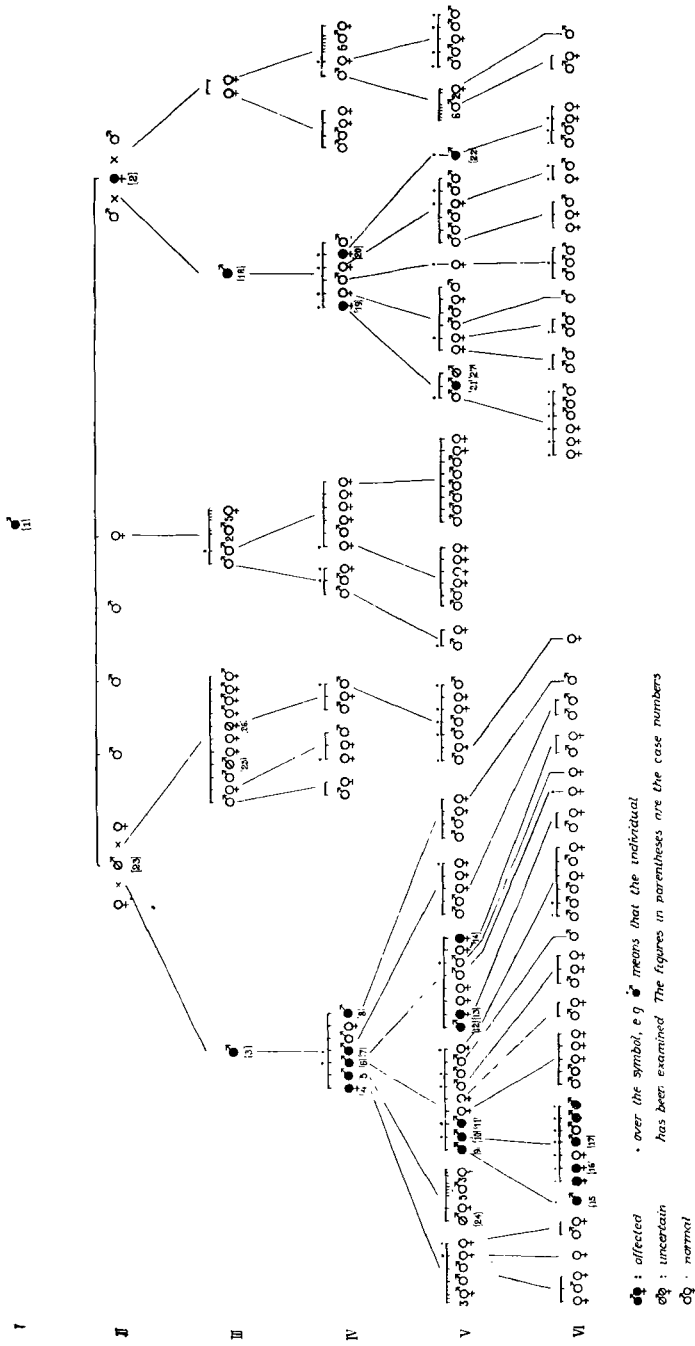


Fig. 2.
Pedigree of the affected family.



Fig. 3.

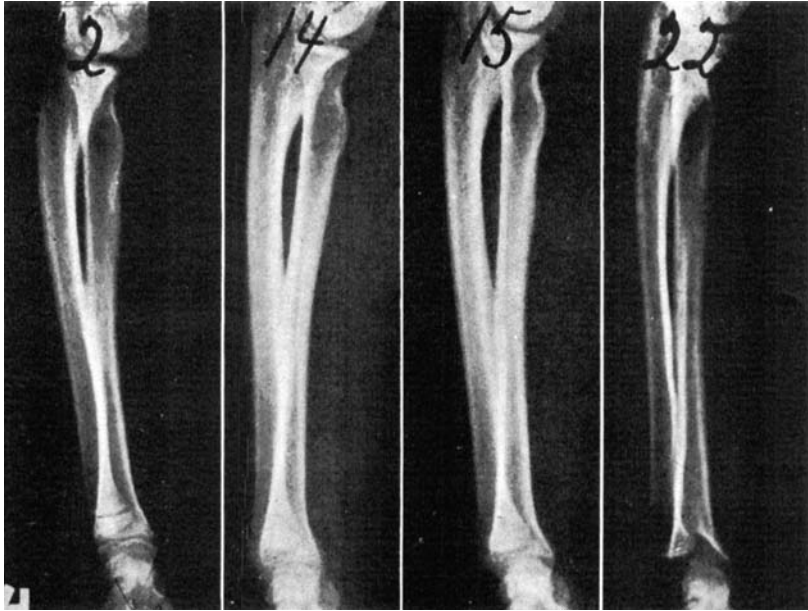
Girl at the age of 12, 14, 15 and 22.

The deformity is obvious even when the distal epiphyseal disc of the radius is open. There is no premature fusion of the volar-ulnar part of the disc, but the entire disc has fused between the ages 14 and 15.

while the rest of the disc continues to grow. *Björkroth* has reported a highly illustrative case of acquired Madelung's deformity in a 17 year old boy with fibrous osteitis in the volar-ulnar part of the disc, whereby the growth of these parts was checked.

In order to study the pathogenesis, I have followed-up 4 of the youngest deformed persons for 10 years (Family 5 in generation VL), figs. 3, 4, 5, 6.

The 4 series of radiograms show that the deformity is present although the disc is entirely open. Therefore, we must reject the theory that the cause is a premature fusion of the volar-ulnar part of the disc. Neither can this theory elucidate the shortening of the whole radius. On the other hand, much can be explained by assuming that a *reduction* takes place instead of a *stoppage* of growth. Such a reduction would affect the *whole* disc, even though it would especially concern the volar-ulnar part. This would account for the shortage of the radius with its ulnar and dorsal curvatures. As a result of the volar bend of the distal end of the radius, the distal end of the

*Fig. 3 b.*

Same girl as in fig 3, side-view.

ulna projects dorsally with, possibly, a dislocation of the distal radio-ulnar joint. The radiograms show that this ulnar projection is most marked when the volar bend of the radius is most pronounced, compare fig. 1 with the series of the children. A reduction of growth also seems to exist in the ulna.

The radiograms cannot reveal whether too early fusion of the epiphyseal disc has occurred. Usually the distal disc of the radius fuses between the ages of 18 and 22. In the girl, fig. 3, the entire disc is still open at the age of 14 but it has fused at the age of 15. In the boy, fig. 4, however, the whole disc is open at the age of 16.

Neither do the radiograms show anything special with regard to the first stages in the ossification of the carpal bones. Usually this proceeds in the following order: the capitate bone, the hamate, the distal epiphysis of the radius, and the triquetral bone. The ossification begins at the age of three years. These four centres were already formed in both hands of my two 6 year old boys. Therefore, I can say nothing as to the order in which they appeared.

The proximal row of the carpal bones forms an angle, with the semi-ulnar bone at the apex. Doubt has been expressed whether the angular formation is primary or secondary to the changes in the

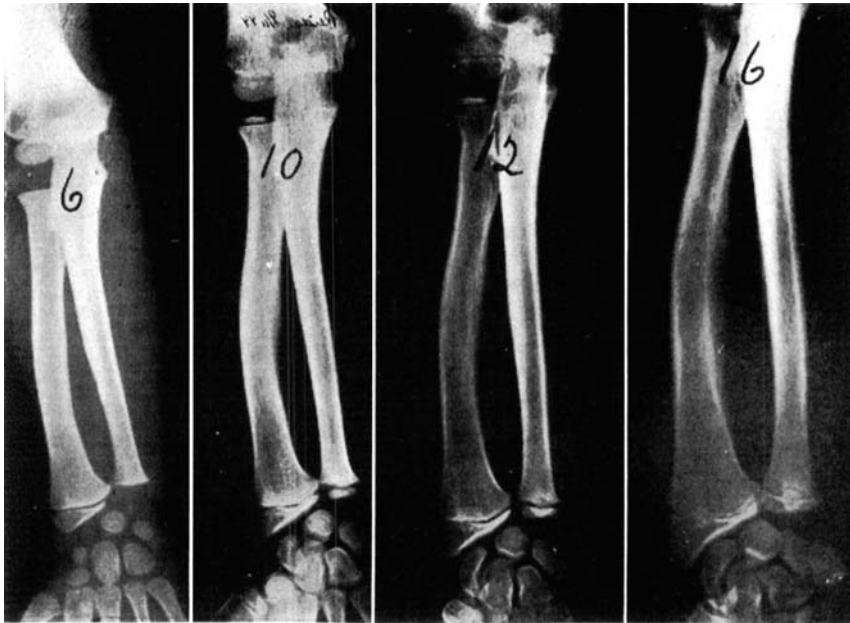


Fig. 4.

Boy at the age of 6, 10, 12 and 16.

The deformity is already seen at the age of 6, with a short and plump radius. The deformity is seen although the whole distal epiphyseal disc is open; it is open—in its entirety—also at the age of 16.

The angulation of the proximal carpal row cannot be seen at the age of 6, but is seen at the age of 10. There is a bony prominence at the ulnar side of the distal disc of the radius; it is already present at the age of 6.

radius. It may be supposed that the first step is the formation of an angle, whereby an uneven pressure against the distal articular surface of the radius is produced. This pressure is supposed to check the growth of the opposing part of the radius. However, it may be objected that it is not certain that an uneven pressure will inhibit the growth. Also, when we study the radiograms, it appears that even at the age of 8 years there exists no angular deformation, in spite of certain deformity. The angle is demonstrated at the age of 10. This ought to prove that the angular formation of the carpal row is secondary to the deformation of the radius.

The deformity is usually said to appear at or immediately before the age of puberty. My cases show that it starts much earlier.

The deformity is regularly accompanied by low stature. In this material all the deformed persons were remarkably small compared

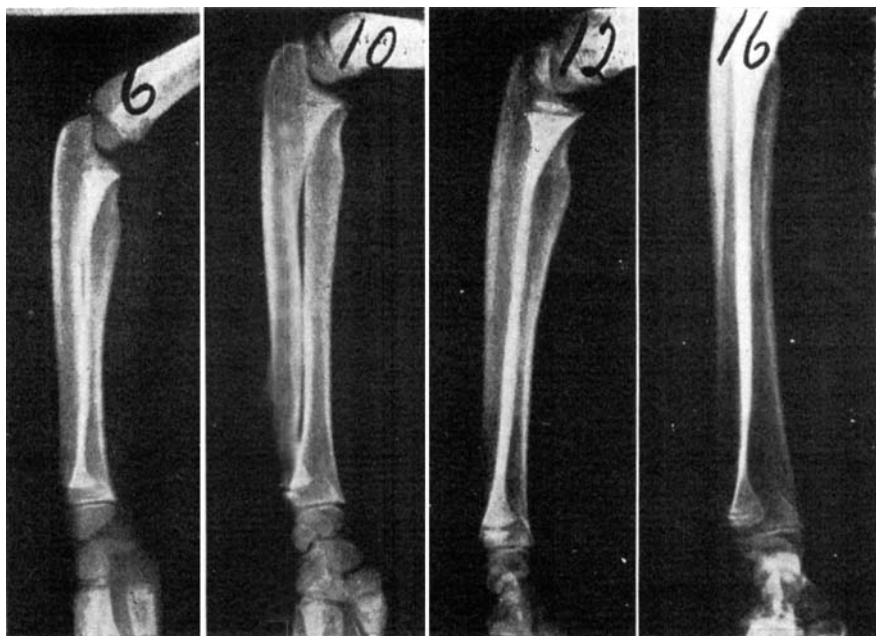


Fig. 4 b.

Same boy as in fig. 4, side-view.

to the non-affected members of the family, in adult age 155-165 cm tall. Unfortunately, I have had little opportunity of X-raying even the lower extremities, not to mention the whole skeleton. When the lower extremities were photographed, I never observed anything unusual.

SUMMARY

30 cases of Madelung's deformity have occurred in the course of 6 generations within one family. The deformity appeared only when it was also present in one of the parents. The cause of the deformity seems to be a dominant, autosomal hereditary factor.

The pathogenesis is apparently a reduction of growth in the entire distal epiphyseal disc of the radius, but especially in its volar-ulnar part. The cause is not a premature fusion of this part, as the deformity is clearly seen even when the entire disc is open.

The angular deformation of the proximal carpal row is secondary to the changes in the radius.

In the present material the deformity has already been found in full course of development at the age of 6 years.

The deformity is accompanied by a general restriction of growth.



Fig. 5.

Boy at the age of 8, 10 and 12.

The whole distal disc is open. The angulation of the proximal carpal row is not certain at the age of 8, but it is at the age of 10.

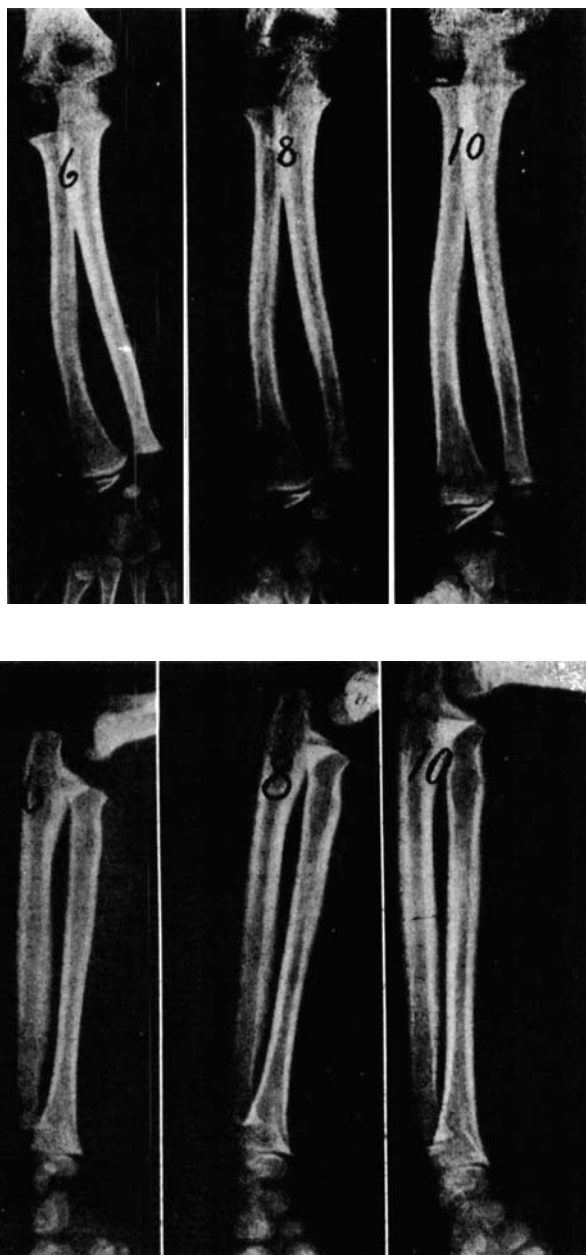


Fig. 6.

Boy at the age of 6, 8 and 10.

As in fig. 4 the deformity is already seen at the age of 6. The angulation of the proximal carpal row is not seen at the age of 6 or 8, but 10. The bony prominence at the ulnar side of the disc is already seen at the age of 6.

RESUME

30 cas de déformité de Madelung se sont manifestés dans une famille au cours de six générations. La déformité n'est apparue que dans les cas où l'un des parents l'avait. La raison doit en être un facteur héréditaire autosomal dominant.

La pathogénèse doit être recherchée dans une inhibition de la croissance de tout le disque distal de l'épiphyse du radius et particulièrement dans la partie ulnaire de la paume de la main. La raison n'en est pas la fermeture trop précoce de cette partie, car la déformité apparaît distinctement lorsque tout le disque est ouvert.

La position angulaire de la rangée proximale du carpe est secondaire aux modifications du radius.

Dans ce matériel d'observation, la déformité a été constatée à l'âge de six ans. Cette déformité est accompagnée d'une courte croissance en longueur.

ZUSAMMENFASSUNG

30 Fälle von Madelungs Deformitet traten innerhalb einer Familie im Laufe von 6 Generationen auf. Die Deformitet trat nur auf wenn einer der Eltern ebenfalls diese Deformitet hatte. Die Ursache muss daher ein dominanter autosomaler Erbfaktor sein.

Die Pathogenese muss auf einer Wachstumshemmung in der ganzen distalen Epiphysenscheibe, aber besonders in ihrem volar-ulnaren Anteil, beruhen. Die Ursache ist nicht eine zu frühzeitige Schliessung dieses Teiles, da die Deformitet deutlich sichtbar ist, selbst wenn die ganze Scheibe noch offen ist.

Die Winkelstellung in der proximalen Handwurzelreihe ist eine sekundäre im Verhältnis zu den Veränderungen im Radius.

In diesem Materiale wurde die Deformitet bereits im Alter von 6 Jahren nachgewiesen.

Die Deformitet ist von einer Verkürzung des Längenwachstums begleitet.

LITERATURE

- Bergan, F.*: Nordisk Medicin 1942:15:2403.
Björkroth, T.: Acta orthop. Scand. 1932. Vol. II.
Paus, B.: Videnskaps-Akademiets Skr., Oslo 1941, No. 8.
Solberg, M.: Tidsskr. D. Norske Lægeforening 1906:26:195.