

A CASE OF CONGENITAL ANKYLOSIS OF THE ANKLE JOINTS AND OTHER MALFORMATIONS

BY

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I think the following case may be of interest, as I have found no record of any precisely like it in the orthopedic literature.

E. J., a girl fifteen years old, born 20th December, 1918, was admitted to the Allm. and Sahlgrenska Hospital, Gøteborg, in 1934. She belongs to a family in which there has not, so far as known, been any instance of malformation, either on her father's or her mother's side. The mother has ten brothers and sisters, the father eight. She has a brother, two years older than herself, likewise in good health and without any physical defect. The mother has not had any abortions. The girl herself was born on time; the mother was well during the pregnancy; was not roentgen-treated. The only noteworthy circumstance was that during the last months of her pregnancy she continued to be very slim (paucity of liquor amnii), so that her friends used to say: »But *where* have you the child?« The girl was delivered in footling presentation; and the accoucheur (Prof. Brandt, of Oslo) is reported as having remarked that the position was »all wrong«.

When she was about one year old she was treated for congenital dislocation of both hip-joints; the dislocation being reduced, whereupon she was kept in plaster-of-Paris bandages for a year or so. Whe she was a couple of years old, both her achilles tendons were tenotomised. She has attended school regularly, and is at the top of her class.

On March 20th, 1934, she was admitted to the surgical dep't of the Allm. and Sahlgrenska Hospital for examination with a

view to possible orthopedic treatment. Her general health was good; she was lively, and her intelligence appeared to be considerably above medium. She was 143 cm. tall; her mammæ about normally developed; strong growth of hair on the pubis and in the axillæ (Figs. 1 and 2). Of her upper extremities there



Fig. 1.



Fig. 2.

was nothing to remark, except as regards the hands, whose position was one of marked ulno-palmar contracture, at an angle of about 130° with the forearm. The distal finger joints were slightly flexed, especially those of the fourth finger (Figs. 3 and 4). The wrists could be bent actively from the contracture position to an angle of about 90° with the forearm. Abduction and adduction, about 45° . She could not clench her hands

completely; there lacked about an inch and a half. The thumb of the left hand could touch all the other fingers except the third; that of the right hand only the fifth finger. Otherwise both hands were alike. The patient can both write and sew. *Roentgen examination* showed that the bones of the carpus were deformed



Fig. 3.

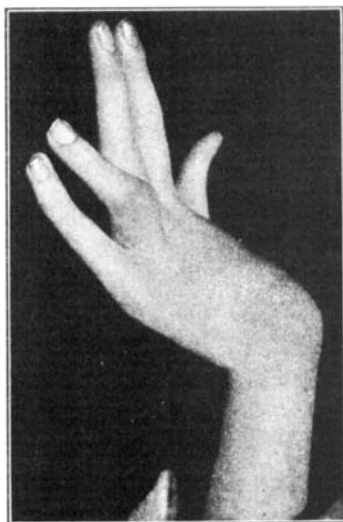


Fig. 4.

and without the normal anatomical separation. The fourth metacarpal was abnormally short (Figs. 5 and 6).

The girl walked with considerable difficulty, owing to extreme outwards rotation of her hip-joints; and this was the principal reason why she sought orthopedic treatment. The mobility in the hips was somewhat restricted; flexion about 50°; rotation about 30°. *Roentgen examination* showed that in the right hip the collum and caput were almost absent. Of the former only the base remained, and opposite to this was a peg-shaped formation, extending in the direction of the upper part of the acetabulum, which was worn slightly flat, with a sort of point projecting in the middle. The trochanteric portion was well developed (Fig. 7). In the left hip there were similar

changes, except that there the remnant of the collum was somewhat larger, and the acetabulum more hollow (Fig. 8).

Both feet were in marked club-foot—about equinovarus—position. When the patient walked, it was almost only the lateral side of them that came in contact with the ground. The great toes showed marked flexion contracture; especially that of the

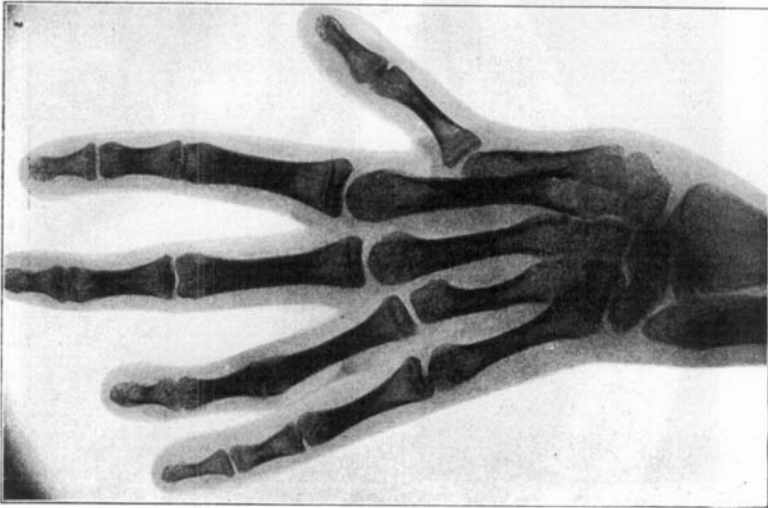


Fig. 5.

left foot, which pointed almost straight downwards. Also the other toes were in marked contracture position. Otherwise the two feet were alike. *Roentgen examination* showed ankylotic union, due to the presence of bridging, bony adhesions in the ankle joint, the talocalcaneal, and—in so far as the navicular bone existed (what could not be positively made out; see Figs. 9 and 10)—the talonavicular joints. The cuboid and cuneiform bones were missing. The malformations were absolutely alike in both feet.

On *March 22nd, 1934*, a rotation osteotomy was done on the proximal end of the left femur, the first metatarsophalangeal joint was resected, and the tendons of the flexors of the other toes tenotomised.

On *April 12th*, the patient was discharged, fitted with a plaster-of-Paris bandage with which it was possible for her to move about. In spite of this, her ability to walk had become considerably better, and she is going to come back later for a similar operation on the other side.

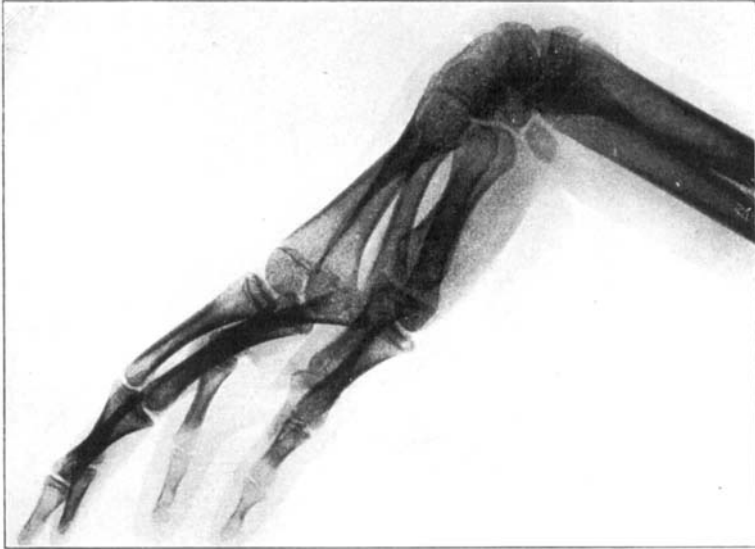


Fig. 6.

The most interesting deformity in this case—not least on account of its rarity—is the ankylosis of the ankle joints. Congenital union of bones—variously termed ankylosis, syndesmosis and coalition—has been observed in other parts of the skeleton. The literature contains descriptions of such deformities, for instance in the bones of the skull, the knee-joints, the spine, between the tibia and the fibula, between the radius and the ulna (the most usual form), etc.

Haglund, among others, has called attention to the necessity of being very careful not to take it for granted, in the individual case, that such a union of bones—ankylosis or coalition—is perforce congenital. As a matter of fact, it may be a case of an over-

looked inflammatory process with secondary fusion; and he gives a typical example of such secondary talonavicular coalition. In the case I have just described, though, it seems that there can hardly be any doubt about the abnormality being congenital. That conclusion is supported not only by the circumstance that

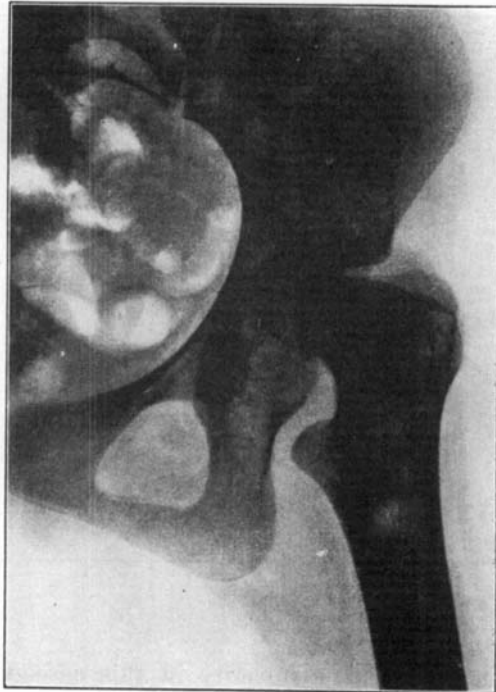


Fig. 7.

there was no history of any disease of the ankle-joints even during the patient's earliest infancy, but also by the fact that the condition was the same in both ankles, by the simultaneous existence of defects in most of the carpal bones, and by the other concomitant malformations.

As I have already said, there has not to my knowledge been any previous description of a case of congenital ankylosis of the ankle-joint; but congenital synostosis of other tarsal bones is

not very uncommon. The most usual is fusion between the calcaneus and the navicular, which was first described by Cruveilhier, already in 1829, and which has since been thoroughly studied bot clinically and roentgenologically, for instance by Slomann and Bentzon. Pfitzner thinks it is connected with the

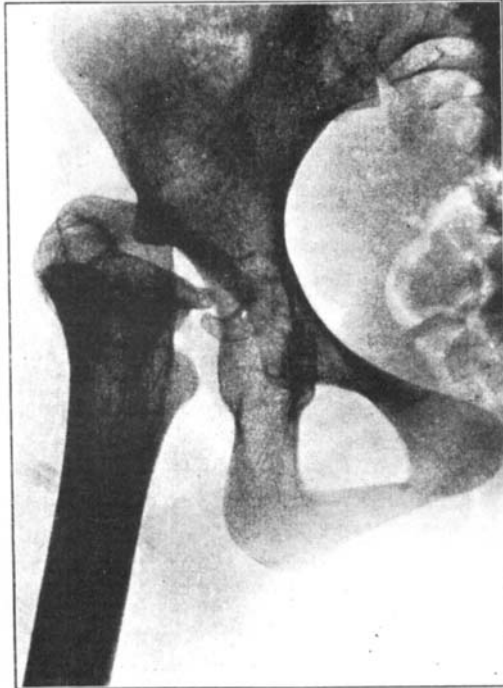


Fig. 8.

presence of a calcaneus secundarius. It can give rise to talipes planovalgus, and Bentzon has used osteotomy and correction with good result in several such cases.

Congenital coalition between the talus and the calcaneus was described for the first time in 1877, by Zuckerhandl, and later by Pfitzner, Korvin, and others. Bentzon, too, has seen cases of it, and believes that the os trigonum is the link responsible for the union. Also this synostosis can give rise to flat-foot troubles,

but otherwise it may be regarded as clinically of little importance.

Congenital union of the talus and the navicular was observed for the first time by Holland, in 1918, and has since been de-



Fig. 9.

scribed by Blenke, among others. Also this coalition is of slight importance, clinically.

A couple of years ago, Rey communicated a case of a synostosis, probably not before described, namely between the calcaneus and the cuboid. For the congenital origin in that case spoke, among other things, the circumstance that the patient in question also had bilateral »cleft hand«. The condition was discovered accidentally, and did not give rise to any symptoms.

Finally, Behr has, in 1933, described a case which, more than any of the foregoing, resembles mine. There was symmetric synostosis of all the tarsal and carpal bones, and, besides, of the tarsometatarsal, the radiocarpal and the carpometacarpal joints. Clinically, the deformity manifested itself in complete immobility of the wrists, both as regarded flexion and extension, together

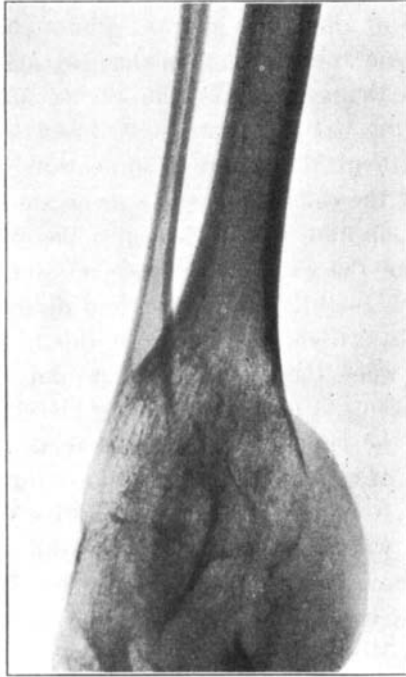


Fig. 10.

with a slightly restricted pronation and supination. The ankle joints could be flexed and extended, but neither pronation or supination was possible, even to the slightest degree: »der Fuss sass in der Malleolengabel wie in einem eisern festhaltenden Scharnier«. The feet were not, as in the cases of synostosis described in the foregoing, of the planovalgus type, but were of a distinct excavatus form. The function, both of the hands and the feet, was remarkably little impaired. The patient herself, who was thirty-three years old when the deformity was discov-

ered, looked at it rather as something that merely detracted, perhaps, from her otherwise good appearance. She was fond of walking-tours, and even climbed mountains now and then.

In the title of this paper I have, for the sake of brevity, termed my case merely: A case of ankylosis of the ankle-joints; but, as a matter of fact, there was also ankylosis both between the distal ends of the tibiæ and the fibulæ, of the talocrural joints, and of the talotibial, talocalcaneal and—possibly—the talonavicular articulations. As the roentgenograms (Figs. 9 and 10) show, the various bones were fused together into one single piece, without the slightest indication of interarticular spaces, and only the calcaneus had to a certain extent preserved its outline. But, in addition to this, also the other tarsal bones—the cuboid and the cuneiform—were missing. The deformity is thus one which combines inhibition and defect. The deformity of the hip-joints, which, according to the history given, had been dislocated when the child was born, and had been treated for that dislocation, is now in the main merely a defect, most pronounced on the right side, where there is hardly anything left of the caput or collum. The acetabular hollows are markedly deep. Clinically, the most prominent feature is the outwards rotation, which was deemed to constitute an indication for a rotation osteotomy. In the upper extremities, finally, it is evidently a case of congenital muscular and tendon contracture of both wrists, with a medium degree of secondary skeletal changes without ankylosis or deformation, and with surprisingly good function.

In all cases of congenital deformities there presents itself, of course, the question of the etiology. It will probably have to be admitted that in spite of all the work—also experimental—that has been devoted to its elucidation, the problem is far from being solved. To begin to discuss it, be it ever so briefly, on the basis of a single case, may therefore perhaps seem an idle task. Yet the analysis of the isolated case is necessary, because it is only by testing the various theories advanced on the basis of

those single cases that we may hope eventually to arrive at some likely explanation.

The deformity which in this special case seems to me, from an etiological point of view, to present the greatest hypothetical interest is the ankylosis of the ankle-joints. As we all know, an indication of the definite joints begins to make its appearance already at a very early stage of fetal life. According to Merkel they are present already by the tenth week, when the fetus has a length of only 30 mm. A lesion sustained by the fetus, and which prevents the normal differentiation of the joints from taking place, must therefore be able to make itself felt at a very early stage; at any rate so early that it cannot very well be a question of pressure effect due to an insufficiency of amniotic fluid, as the production of the latter can hardly have begun yet. The idea of a so-called »deformity due to intrauterine pressure« may therefore be altogether rejected.

Against the supposition of an, at any rate very hypothetical, intrauterine disease during a more advanced stage of fetal life that might cause ankylosis, speaks not only the fact that the deformity was bilateral, but most of all the simultaneous defect of nearly all the tarsal bones. The conclusion, therefore, must be that some harmful agent, of nature unknown, either endogenous or exogenous, has had an effect on the fetus during this early stage of its development.

Yet there is in the anamnesis one positive statement, which, in spite of what I have said in the foregoing, should not be overlooked, even though the discussion here is about the deformity of the ankle-joints; and that is the abnormally small quantity of amniotic fluid, of which there can probably not be any doubt. Murk Jansen has in recent years suggested that a great many congenital malformations may perhaps be explained if we assume them to be due to the harmful influence of a too small amnion cavity already during the first fetal months, in other words, before there can yet have been any production of amniotic fluid. This abnormal smallness of the amnion may, according to him, be either endogenous, due to hereditary factors, or exogenous, determined by either physical, chemical or in-

fectious noxæ. Kiewe, who has recently written an interesting, very complete and critical, paper on the genesis of congenital malformations, says that it is difficult to imagine that an abnormal pressure of the amnion during the first months of pregnancy should cause only local deformities; and he further points out that nothing has been positively proved in regard to the increase of the hydrostatic pressure of which it is supposed that it should produce an ischemia leading to disturbances of growth and to deformities.

And yet it seems to me that my case lends a possible support to Murk Jansen's theory in so far as the lack of sufficient amniotic fluid during the later period of pregnancy may indicate that there has already at an early stage been something wrong with the amnion, which has had a harmful effect on the child. In that manner one would at least be able to explain the different deformities in this case as due to one, common cause. The ankyloses, together with the bone defects and -malformations, would then have been due to the anomalous amnion directly; while the dislocation of the hip-joints and the contractures of the wrists would have been due, not directly to »amnion disease«, but to the oligohydramnios resulting from the latter.

With regard to the factors that in this case should have had a harmful influence, perhaps on the amnion, perhaps on the ovum itself, we have however not the slightest basis for surmise. Neither on the father's nor the mother's side are there, so far as we can find out, any kind of malformations that might point to an endogenous genesis; and that in spite of the fact that both parents had many brothers and sisters. With regard to the generations farther back I have not been able to examine into the question. As regards the mother, she had never had any abortions, nor any disease of her genitalia that might suggest alterations of her uterine mucous membrane. Kiewe, in the paper referred to above, asserts that precisely lesions of the uterine mucosa, with the resulting disturbances of fetal nourishment, probably play an important part as far as congenital malformations are concerned. If this is so, the problem is one of very great practical importance, considering the steadily increasing frequency of abortions; and this is a point to which the orthopedists

should undoubtedly pay every attention when studying the anamnesis in cases of congenital deformities.

SUMMARY

The author describes and discusses a case of congenital, symmetrical ankylosis of the ankle-joints and proximal tarsal bones, combined with symmetrical contractures of the wrists and defective formation of the proximal ends of both femurs.

ZUSAMMENFASSUNG

Der Autor beschreibt und diskutiert einen Fall von kongenitaler, symmetrischer Ankylose der Knöchelgelenke und der proximalen Tarsalknochen, kombiniert mit symmetrischen Kontrakturen der Handgelenke und defekter Bildung der proximalen Enden der beiden Femura.

RÉSUMÉ

L'auteur décrit et discute un cas d'ankylose congénitale, symétrique dans les articulations talo-cruvales et des os tarsiens proximaux, en relation avec des contractions symétriques des poignets et une formation défectueuse de l'extrémité proximale des deux fémurs.

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