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CHONDRO-OSTEO-DYSTROPHY (BRAILSFORD) AND ITS RELATION TO OTHER BONE DISEASES[1])

A REPORT OF SIX CASES

BY

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Achondroplasia (Parrot) or chondrodystrophia foetalis (Kaufmann) is a long recognized form of congenital deformity with very typical clinical symptoms and, as we now know, a characteristic roentgen picture. However, cases have been described recently which in some respects are reminiscent of achondroplasia but which in others are not at all typical of this disease. The first well known case was published in 1925 by Silfverskiöld (from the Orthopedic Hospital in Stockholm) under the name of "Forme fruste of chondrodystrophia with changes simulating several of the known local malacias". Later the Scandinavians Wahren (also from the Orthopedic Hospital), Dale, R. Hanson, Sundt, Nilsonne (Orthopedic Hospital), Stören and Sundal described similar cases as "atypical achondroplasia, multiple disturbances in growth, familial bone disease", etc. In his book "The Radiology of Bones and Joints" (Ed. 2, 1935) Brailsford, on the basis of about 30 cases from the literature and 12 of his own, introduced a new disease concept and a new name for these "atypical" cases, namely, chondro-osteodystrophy. Just lately, in his dissertation "Studien über hereditäre Epiphysenstörungen", Ribbing has discussed in detail the particular connection between cases of this kind and the so-called osseous aseptic necroses (local malacias).

Since the primary material is yet relatively small and the related problems are not completely solved, case reports are

[1]) This article was written in 1938 but not printed before now.

still of value, for which reason I shall describe a number of new cases observed at the Orthopedic Hospital in Stockholm and make a few comments thereon.

Since Ribbing does not mention Brailsford's series and since, in my opinion, he has not collected the features characteristic of this disease (which he calls achondroplasia atypica), I shall first give a short description of chondro-osteo-dystrophy (Brailsford), which agrees essentially with Brailsford but which also contains some of my own viewpoints as gained directly from a study of the original reports of the cases collected by Brailsford and of a number of others of similar nature. There are quite a number of cases published under different names which are not mentioned by this author but which probably belong to the same category. The Kaschin-Beck disease spoken of in the national Russian literature has not been sufficiently investigated roentgenologically; however, its typical, endemic form probably does not belong, but the sporadic, atypical, familial cases described by Goldstein and Nikiforov agree clinically and roentgenologically with Brailsford's. A great many cases of so-called generalized platyspondylisis, because of their roentgenologic, clinical and familial features, should also be grouped with chondro-osteo-dystrophy, that is, unless one uses the term platyspondylisis in such a wide sense as Buchman.

The characteristic findings in these cases is more or less extended disturbances in the laying down and growth of the epiphyses with irregular ossification both in the primary and secondary centres of ossification. Roentgen examination shows multiple, irregular bony nuclei in the epiphyses, irregular and uneven epiphyseal lines and often expanded metaphyses. Later pressure deformities appear in these faultily organized regions and remain after the cessation of the active stage at the time of fusion of different epiphyseal nuclei and epiphyses with diaphyses. According to Hirsch, the development is divided into a latent, florid and healing stage with deformity as a definitive sequel. Clinically, nothing abnormal is observed in the children at birth (normal proportions). In severe cases, the first symptom is often kyphosis in the dorsolumbar region, which may appear

as soon as the child begins to sit or when it begins to walk at the age of one to two. The knee, ankle and wrist joints are often swollen as in rickets, for which this disease is often mistaken clinically. At the age of five to six, the patients who are severely affected have a very characteristic appearance; they are relatively dwarfed due to a definite shortening of the trunk in comparison with the extremities, which are of relatively normal length. The head often sits close down on the chest which protrudes anteriorly. The back shows marked kyphosis in the dorsolumbar region and the joints of the extremities are swollen. The mental development is normal. The bodily growth seems to cease entirely at this time, the children growing only a few centimetres in several years. There are no inflammatory symptoms and generally no pain in the affected epiphyses and joints. At this time, or later around the age of puberty, there is generally some subjective and objective limitation of motion (but never complete ankylosis) in the joints of the lower extremities, especially the hip joints, which later increases or remains the same but never decreases. There is often a flexion contracture in the hip joints which, in its turn, if the back is not already fixed in a kyphotic position, leads to a compensatory increase in the lumbar lordosis. At some time from the age of twenty to thirty, depending upon the degree of deformity in the articular extremities, symptoms of atrophic arthritis (arthritis deformans) begin to appear with troubles, sometimes irritation, in the joints, and a greater or less degree of invalidism. The more severe cases remain dwarfs with a short, compressed trunk and a protruding chest, often a short neck and sometimes an occipital kyphosis due to the pressing of the base of the skull down over the upper cervical vertebrae, and a prominent dorsolumbar kyphosis or marked lumbar lordosis. It is often of familial occurrence; of Brailsford's 42 cases, 15 were related and distributed in six families. The characteristic roentgen findings in the different stages are as follows. In the infant all the articular cartilage seems to be greatly thickened with increased distances between the bony outlines in the joints. The ends of the diaphyses are irregular and thickened and the bony nuclei

in the epiphyses are first seen as multiple, anomalous islands of bone which gradually fuse. The cancellous tissue is extremely coarsely and irregularly reticulated, the compact tissue is thin, the joint contours are irregular and the articular ends are often expanded. There is generally no definite delay in the appearance of the osseous nuclei in the metacarpus and metatarsus, but there are numerous anomalies and variations, and the growth and molding are slow and irregular. The most typical changes are found in the spine and hip joints. Before puberty the true pelvis is narrowed with wide joint spaces; after puberty the articular spaces are decreased, the true pelvis is compressed but the false pelvis is broad. The changes in the bone structure brought about by the defective ossification are particularly evident in the hip joints where the static strain is greatest; the growth is inhibited and irregular; the femoral head and neck are deformed and the acetabulum adapts itself to their changed shape. There are instances in which the head and neck are never ossified, as in severe coxa vara congenita, but there are also cases with slighter lesions in which the dystrophic disturbance seems to cease in early childhood, after which normal ossification takes place.

Different degrees and types of changes also appear in the spine. The most serious lesions correspond so well with those in my first case that the reader is referred there for their description; there are both diffuse changes in the whole spine and local defects in the region of the first lumbar vertebra. These cases generally have other serious disturbances with fragmentation of practically all the epiphyses in the body and, according to Brailsford, these children often do not attain full maturity but die in the uterus or in early child-hood. In less severe cases the vertebral bodies are regular in size and similar in shape, but compressed and often stippled. Before the epiphyses for the rims appear, the anterior parts of the bodies are "tongued" with defective ossification in the superior and inferior corners, and later they are irregularly outlined. Even in slight cases local defects, generally in the first lumbar segment, can be found, as well as general compression of the bodies.

According to the seriousness and extent of the lesions, Brailsford divides the cases into four different groups. A) Generalized form with fragmentation of practically all the epiphyses in the body and the marked changes in the spine just described; dorso-lumbar kyphosis and other symptoms appear at an early age. B) Moderate generalized form in which the active phase appears to stop before puberty. The frequent disturbance in the ossification of the metaphyses leads to shortening of the extremities and pressure deformities of the ends of the extremities, vertebrae and pelvis. Most of the epiphyses are fragmented. C) Changes confined mainly to the spine and hip joints, and less often to the knee joints. The vertebrae are compressed, laterally expanded, stippled and sometimes tongued. There is defective ossification of the femoral necks and heads but otherwise but slight signs of dystrophy of the bones of the extremities—the most common being a separate epiphysis for the base of the second metacarpal. D) Lesions confined to the spine which shows the generalized changes just described and occasionally local defects in the first lumbar vertebra.

The roentgen findings are typical but, if only one joint is photographed, they may be confused with those in cretinism, localized osteochondritis (local malacia), achondroplasia or multiple chondroma. I shall return to the differential diagnosis after I have described my cases.

The *first case* was a boy aged 9 on admission to the Orthopedic Hospital in October of 1936 (Record 3155/36). Two sisters aged 17 and 19 were healthy and showed no signs of deformity. No hip or spinal disease was known in the family. Besides having the usual children's diseases (measles, chickenpox), he had been healthy and well-shaped until about the age of 4 when rather suddenly he began to limp and to become tired after walking or standing a long time. He complained occasionally over moderate pains radiating down toward the knees, mostly on the left side. There was no fever or feeling of general ill health. The symptoms became aggravated, and on Sept. 1, 1934 at the age of 7 he was admitted to the surgical ward of the Falu Hospital (Dr. J. Waldenström, physician-in-chief) where he remained until Feb. 6, 1935. He then showed about the same clinical picture as on admission to the Orthopedic Hospital two years later which is described below. Because of a flexion contracture of about 30 degrees in both hip joints which improved only slightly after

staying in bed, on Sept. 27, 1934 a subspinal tenotomy was done on the flexor muscles of the hip joints and the legs were extended as much as possible under deep general anesthesia, after which a large plaster bandage was placed from the feet to the inferior part of the chest. After two months the plaster was removed and gymnastic therapy with walking exercises was instituted. On discharge on Feb. 6, 1935 the gait was somewhat improved and the flexion contractures were almost gone, but the marked obliquity of the pelvis and lumbar lordosis in the upright position remained. After leaving the hospital his legs and back became progressively weaker and he finally could not walk more than fifteen minutes without sitting down and resting. There was no real pain. He was admitted on Oct. 29, 1936 to the Orthopedic Hospital in Stockholm.

Clinical examination revealed good general condition and a healthy appearance. There were no external signs of endocrine disturbance. The mental development was perhaps slightly above normal. Nothing abnormal was found in the internal organs. The reflexes were normal except for slightly exaggerated knee jerks on both sides. Otherwise he was normal neurologically. The Wassermann reaction in the blood was negative. Examination of the blood calcium showed 11.1 mg. per 100 cc. Calcium analysis of the urine (Dr. E. Jorpes, Karolinska Institutet) showed the daily excretion to be normal. The sedimentation rate was normal. The temperature was afebrile. The boy was rather tall for his age—140 cm. The proportion between the trunk and extremities was about normal; in the upright position the finger tips did not reach the knee joint space. The proportions between different parts of the extremities were also normal. For other details, see the photograph (fig. 1).

He walked with a very marked lumbar lordosis (about 45 degrees) and in a waddling manner. The Trendenlenburg test was positive on both sides and most marked on the right. When standing, if possible he leaned forward on his hands; both hip joints were flexed to 45 degrees, the right knee to 25 degrees and the left knee somewhat less. When he sat down in a relaxed position there was a marked semicircular kyphosis with the apex in the inferior dorsal region which could be entirely straightened out actively. There was no tenderness over the spinous processes and compression of the spine in the long axis elicited no pain. The dorsal musculature was good. Lying down, he held the left leg slightly adducted and inwardly rotated in the hip joint and there was also a very pronounced lumbar lordosis. When this lordosis was straightened out, by flexing one of the legs to the maximum in the hip joint, there appeared a flexion contracture of 40 degrees in the right hip joint and of 35 degrees in the left which could not be forcibly corrected. The functional shortening (measured between the medial malleoli) of the left leg was 2 cm. and the real shortening (measured from the spina iliaca anterior superior to the medial malleolus) of the same leg was ½ cm. and entirely

distal to the region of the femoral neck in the line of the tip of the trochanter to the external malleolus. The hip musculature was somewhat atrophic generally and the thigh musculature fairly normally developed.

Passive movements in the hip joints showed flexion, adduction and inward rotation to be normal on both sides. On the right side there was a shortage of 40 degrees in extension to midposition and on the left 35

Fig. 1.

degrees. Abduction was moderately restricted on both sides (to 25 degrees from midposition). The outward rotation on the right side was normal but slightly limited on the left. The active movements were of just as great range as the passive; the endurance and strength were considerably reduced.

The articular ends in the knees were not expanded. On the right side extension could not be effected right to midposition, but 5 to 10 degrees remained, otherwise the mobility was normal.

When weight was placed on the feet, there was a slight valgus position, otherwise nothing abnormal. The movements in the ankle joints and toes were normal; the dorsal flexion in the metatarsophalangeal joints was about 90 degrees on both sides.

The free movements were of normal range and strength in all the

joints of the upper extremities. The hands were somewhat short and stubby with short fingers and the four ulnar digits were nearly equal in length. The fingers could be overextended nearly 90 degrees dorsally in their proximal joints and nearly 45 degrees in their distal joints, otherwise the movements were normal.

The chest, neck and nape were of normal configuration. The shape

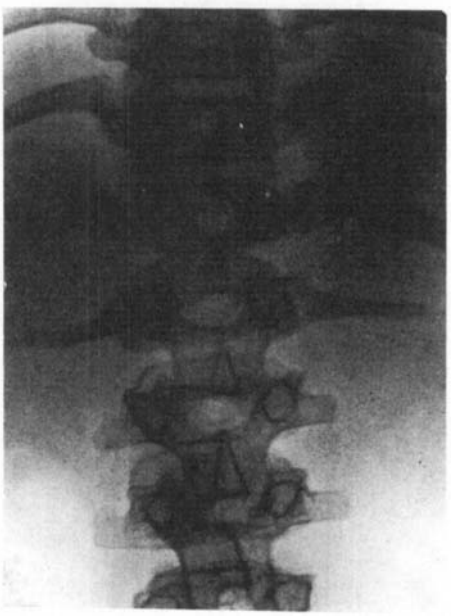

Fig. 2.

of the head was normal and, except for a broader nasal bridge than usual, the face was also normally shaped.

Roentgen examination (Oct. 29, 1936). Frontal views of the spine (fig. 2) showed both generalized and local changes; there was less density than normal, the cancellous structure was very coarse and irregular and the vertebral bodies in the dorsolumbar region were indefinitely delimited. The intervertebral spaces were also irregular, poorly defined and distinctly higher than normal. The intervertebral joints did not, as usual, lie on two straight lines diverging inferiorly, but were irregularly disposed; the articular processes were poorly developed. The bodies were distinctly flatter than normal but broader, as though compressed; in some places their height was even less than one-third of their breadth. In lateral views (fig. 3) it was seen that the vertebral bodies differed in

shape, size and position. The eleventh dorsal and first lumbar segments were much smaller than their neighbours because of defective development in the upper and lower parts of the anterior portions, the middle parts jutting forward like a thorn. These two vertebrae were also slightly displaced dorsally in relation to those lying above. Both these factors, the wedge shape and the backward subluxation, caused a marked kyphos

Fig. 3.

which was not yet fixed; furthermore, because of the extended changes it was relatively soft and not so pointed as in the following case. The other bodies were not of the normal rectangular shape; in the lumbar region they had rounded corners and curving outlines; in the dorsal region they had a very characteristic appearance (which Brailsford likens to an old shoe facing ventrally); they were flattened in the centre and expanded anteriorly with a small projection in the middle part. The outlines of the bodies in the dorsal region were distinct, fairly sclerotic and well defined from the very thin cancellous tissue. The processes were small but well defined and the extremities of the transverse processes were not cupped (a frequent finding in other cases). The cervical spine showed no marked changes. Comparison with the roentgen pictures taken in the Falu Hospital two years earlier in September of 1934, which were

kindly lent me by Drs. J. Waldenström and E. Saul, showed that the spinal lesions had remained unchanged since then.

In order to bring out the course the changes in the pelvis had run, I shall begin with the roentgen findings in the Falu Hospital on Sept. 1, 1934 (fig. 4). The pelvis seemed rather compressed over the hip joints, the opening becoming almost square. The alae ilii were slightly projecting (this is not as well seen in the copy as in the original roentgenogram).

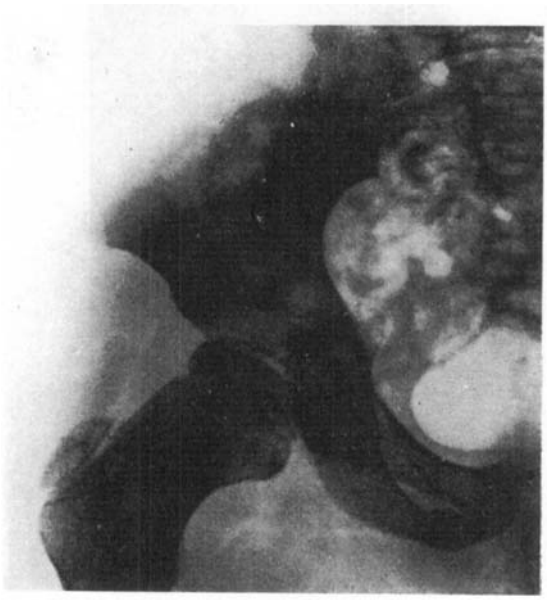

Fig. 4.

The sacro-iliac joints and the symphysis were distinctly broader than normal. The ischium and pubis were united in the inferior border of the foramen obturatum but the junction was somewhat expanded and blurred in outline. The calcium content was less than normal in the whole pelvis, especially around the hip joints where the structure was mushy and coarse. The acetabuli seemed somewhat shallow and flat; the distal portions appeared to be displaced medially (see the tear figure), while the superior portions seemed normally rounded. The floors of the acetabuli were rather thick. Their articular margins were wavy and not sharply outlined in the centre. The Y cartilage was ossified. The epiphysis of the right head showed a blurred structure. In the centre of the articular surface a segment about 3 mm. in height was seen to be fragmented and of fairly low calcium content. However, the peripheral parts of the arti-

cular surface, of which a good centimetre can be seen superiorly and laterally and infero-medially, were normally outlined. Lateral to this fragmented segment (zone 1 in fig. 5) was a very "porous" zone (2) about 7 mm. broad, separated from the first by small clearings in the centre. Lateral to this porous zone there was still another, slightly sclerotic zone (3) a few millimetres wide which fused with the epiphyseal line in the centre (4). The head seemed somewhat displaced laterally but not upwards; the distance from the floor of the acetabulum to the head was distinctly increased in the inferior part of the joint, to nearly twice that from the roof of the acetabulum to the superior part of the head. The neck was a little expanded with a slightly projecting supero-medial extremity and a straight superior border; there was a very slight coxa

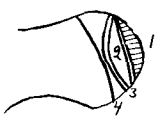

Fig. 5.

vara deformity. The left side showed the same picture but centrally the changes were less in extent. The fragments were also very pale but the zones in the head were not so distinct and there was no definite sclerosis. With Lauenstein's projection the right side showed the same central position of the changes, in this case limited laterally by a narrow band of sclerosis parallel with the epiphyseal line. On the left side it was distinctly seen that the head was not compressed but normally rounded. This projection also showed clearly the same zones as the antero-posterior view showed in the right side: innermost beside the joint was a fragmented zone poor in calcium, then a band a few millimetres wide entirely lacking in structure and casting no shadow, which merged laterally with a zone with blurred contours, and finally a slightly sclerotic peripheral zone.

The photographs taken at the Orthopedic Hospital on Oct. 29, 1936 showed that no great change had taken place; the different zones could not be distinguished so clearly in the epiphysis of the head, but the structure was more indistinct generally, especially in the region of the socket. The distance between the head and the tear figure was very great, about 3 times the height of the superior joint space. In the so-called cystoscopy position (corresponding to Lauenstein's projection with the patient lying on his back and the legs flexed about 60 degrees and abducted 45 degrees in the hip joints) the disturbances looked unchanged; in this projection the joint space seemed equally wide throughout its length (fig. 6). The roentgenograms taken on Feb. 22, 1937, that is two

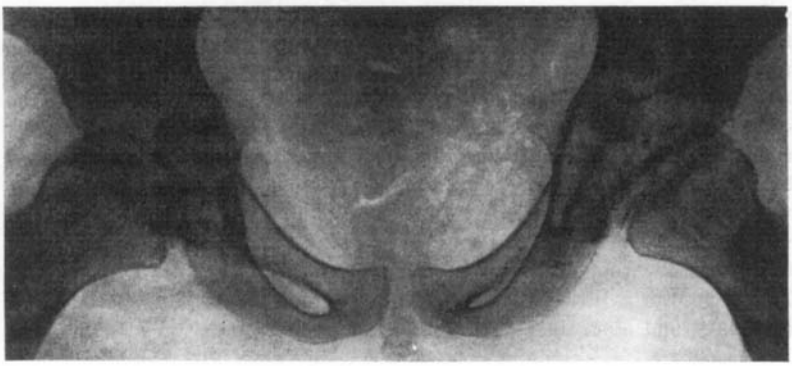

Fig. 6.

years and seven months after the first, showed the same picture but perhaps the outlines af the head appeared more clearly. In the cystoscopy position a distinct sclerotic peripheral zone 2 to 3 mm. broad was seen on the right side.

Pictures of the knees (Oct. 29, 1936) showed the epiphyses to have a coarsely reticulated cancellous tissue and a thin cortex. The ends of the diaphyses were slightly splayed out; the articular borders of both the femur and tibia were slightly wavy but sharp; the patella was normal and the fibula was of normal length.

The compact tissue in the feet was rather thin and the cancellous tissue somewhat "mushy". An extra epiphysis was found on the tip of the external malleolus consisting of one nucleus about half the size of a pea and another the size of a pinhead. The apophysis of the calcaneus was somewhat denser than normal. A pseudo-epiphysis was seen distally on the first metatarsal, fragmented into three parts. Another was seen distally on the proximal phalanx of the big toe where the lateral corner of the trochlea formed its own little nucleus. No sesamoid bones were visible. Nilsonne's index was negative, that is the distal extremity of the first metatarsal did not extend beyond a line touching the articular surface of the second metatarsal and perpendicular to the long axis af this bone.

In the shoulders the head of the humerus was normal. The glenoid cavity was irregularly pitted, very flat and poorly delimited from the neck.

The elbows showed considerable fragmentation of the trochlear nucleus. A small excavation was found laterally on the radial diaphysis right beside the epiphyseal line. The olecranon was also fragmented but the various nuclei were of good density.

The hands showed coarse trabeculation in the radial and ulnar epi-

physes. The styloid process of the ulna consisted of several small nuclei. The bones of the carpus were coarsely reticulated. The naviculare was unusually small; one side was increased in density and the other only laid down as two extremely small, dense spots. Remains of pseudo-epiphyses were seen distally on the first metacarpal and the proximal phalanx of the thumb.

The skull, clivus and sella turcica were normal roentgenologically.

The treatment at the Orthopedic Hospital consisted of complete recumbency in a plaster bed. The legs were hung up in plaster splints in such flexion in the hips that the lumbar lordosis became normal. This angle of flexion was then successively decreased and the flexion contractures thus slowly released. While in the hospital he suffered from transient troubles in the elbow and knee joints combined with fever and a rise in the sedimentation rate to 25 mm. per hour, but these soon disappeared and left no traces.

The other cases all came from one family; of 8 members, 4 had the same type of skeletal disturbances. The parents were living and well and had no symptoms in the back or joints. No hip or back deformities were before known in the family. The first of the children was a man of 32 who was healthy and of average height but his 9 year old son (case VI) showed slight lesions in the spine. The second was a man of 30 with no other subjective symptoms than slight tiredness in the loins after hard work. Clinically, a slightly increased lumbar lordosis was found, but an otherwise normal back. There were no symptoms or deformities in the extremities. Roentgenologically his pelvis and spinal column were normal. The third child was a man of 28 with typical, pronounced skeletal changes (case IV). The fourth was a man of 26 with the same changes (case III). The fifth was a woman of 22 with slight but typical lesions (case V). The sixth was a man of 20 with pronounced changes (case II). The seventh was a big woman of 18 without deformities or subjective troubles, and the eighth was a girl of 16, of average height who also had no deformities or symptoms (the last two have not been examined roentgenologically).

Since the past history was about the same for the three brothers, only that of the youngest will be reported in detail.

Case II (record 10023) age 20. At birth he weighed over 4 kg. and appeared to be quite normal. He was a strong infant. He had no diseases worthy of mention during childhood and adolescence. The rigidity in the hip joints began when he was about 5 or 6 years old. It was then seen that he could not run like other children but he had no pain. He was therefore not sent to school until he was 8 years old. Because of this rigidity, the parents took him at the age of 9 to the Orthopedic Hospital in Stockholm. Unfortunately the records and roentgen pictures from this examination are no longer available, only the diagnosis "coxa vara from Perthes' disease?" being recorded. No treatment was thought indicated. From that age on he did not grow normally but remained small and somewhat "saddle-backed" with a slight hump at the junction of the dorsal and lumbar spine. There was stiffness and restricted motion in the hip joints but it caused him no inconvenience or pain to walk and move about; even football gave him no other trouble than a slight tiredness after an hour. He walked without a cane but preferred to bicycle. He also felt some stiffness in his shoulders, not being able to lift his arms up to the vertical plane properly. Because of his decreased earning possibilities, the Pension Department arranged for his training as a watchmaker. However, when he was 19½ years old (autumn of 1936) he had acute (rheumatic) polyarthritis with fever and pains in the left hip, left knee, right elbow, back of the neck and joints of the jaw, for which reason he was admitted to the Orthopedic Hospital in the middle of January of 1937. He had been away from home the whole autumn and, according to all the members of his family, during this time his back had collapsed considerably, causing a great increase in the previously insignificant hump.

Clinical examination on Jan. 14, 1937 revealed all the signs of acute polyarthritis involving the joints of the left hip, both knees, left wrist, right elbow and both sides of the jaw. His general condition was greatly affected; he was pale, thin and perspired greatly. At first his temperature was between 38 and 39° C. There was a high sedimentation rate (80—120 mm. per hour). The internal organs were normal. There were no external signs of endocrine disturbance or of healed rickets. The Wassermann reaction in the blood was negative. There were no neurologic symptoms.

In addition to the swelling, exudate and tenderness over these joints, the following findings were made. The head was of normal shape and appearance, the face was not deformed in any way (see the photograph, fig. 7) and the neck was of normal length and freely movable. The trunk was unusually short but the extremities were of about normal length and proportions (body length about 148 cm.). The back showed a large hump with an angle of about 120 degrees in the inferior dorsal and upper lumbar region, the apex involving a little over three vertebrae. Inferior

to this gibbus there was a very slight lordosis in the lumbar region. Superiorly the back was quite flat with a small lordotic curvature in the inferior cervical region. There was no direct tenderness over the spinal processes and no indirect tenderness. The dorsal muscles were fairly tense on both sides but not tender. The whole dorsolumbar spine was completely fixed.

On examination of the pelvis, both trochanters were palpated more

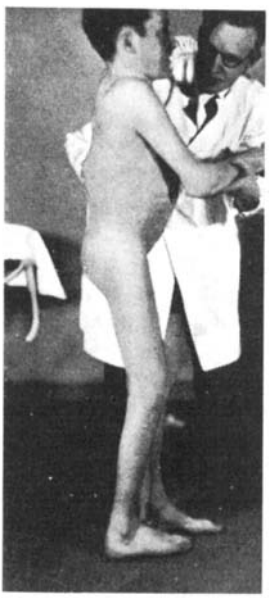

Fig. 7.

dorsally and proximally than normal; the distance from the tip of the trochanter to the interspinal line was only 1.5 cm. on the right side and 0 cm. on the left. The left leg was held slightly adducted, inwardly rotated and flexed in the hip. Maximal extension in the hip joint showed a flexion contracture of 25 degrees to remain, from which position the leg could only be flexed 40 degrees. No rotation was possible and there was only rocking in the direction of abduction and adduction. On the right side there was a flexion contracture of 15 degrees and the leg could be further flexed 110 degrees. Rotation could only take place as rocking and there was altogether 25 degrees of abduction and adduction.

The knee joints showed, besides acute symptoms of irritation, a shortage of extension of 55 degrees on the left side and of 15 degrees on the right. The articular ends were slightly expanded.

Examination of the feet revealed the toes to be about equal in length

and somewhat club-shaped, and the presence of a fairly pronounced metatarsus latus. The longitudinal arches were well preserved. There was no restriction of motion in the metatarsophalangeal joint of the large toe or in other joints.

The shoulders showed fairly pronounced atrophy of the muscles of the thoracic girdle, especially in the supraspinal and infraspinal regions. In simultaneous movement on both sides, the abduction in the shoulder joints was 100 degrees, the elevation forwards 120 degrees and the rotation moderately restricted, mostly outwards.

The hands were of normal shape and mobility with no swelling in the joints (the capacity of hyperflexion in the proximal joints of the fingers was normal—30 degrees).

Roentgen examination (Jan. 14, 1937). Photographs of the spine showed the prominent kyphosis to be caused by defective development in the anterior parts of the twelfth dorsal and first lumbar vertebrae, giving them a wedge shape. Moreover, both these segments were slightly displaced dorsally in relation to those lying above. There were also more generalized lesions in the spine; the bodies showed the same characteristics as in the preceding case, including the rounded corners and semicircular, curved outlines in the lumbar region. They were flatter than usual but broader ventrally and wider sagittally. Beside the intervertebral spaces their borders were indistinct and irregular, as though divided in several lines; there were also numerous semicircular excavations partly bordered by a sclerotic zone. The anterior part of the intervertebral spaces, especially in the inferior dorsal region, was filled up with cloudy condensations with lamellae parallel to the outlines of the bodies. In the place of the normal marginal epiphyses were ill-defined, small dense spots which were not fused with the bodies. These changes did not correspond to Schmorl's nodes ("knorpelknötchen") nor to kyphosis dorsalis juvenilis (Scheuermann), but were more diffuse disturbances in the growing zone and were most pronounced in the inferior dorsal region. The height of the intervertebral spaces was somewhat decreased. All the processes were small and slightly irregular. In the cervical spine the intervertebral spaces were of normal height. The bodies were regular but somewhat flatter than normal anteriorly causing a decrease in the normal lordosis.

The pelvis had a very characteristic appearance (fig. 8) which was found in all the diseased members of the family but not in the normal brother examined. The alae ilii were broad and projecting but short, the spina iliaca anterior superior was situated unusually low, the inferior part of the ilium was narrow and the whole pelvis seemed compressed over the hip joints, making the pelvic inlet more square than normal. All the cases had protrusio acetabuli to a greater or less degree. The sacroiliac joints were perhaps somewhat broad; the outlines of the symphysis were indistinct.

Photographs of the right hip showed a slender femoral neck with perhaps a smaller angle than normal (note the position of rotation, however). The head showed changes which in their localization—the centre third—agreed entirely with those in the previous case. The superior (lateral) articular surface of the head was not affected but below it

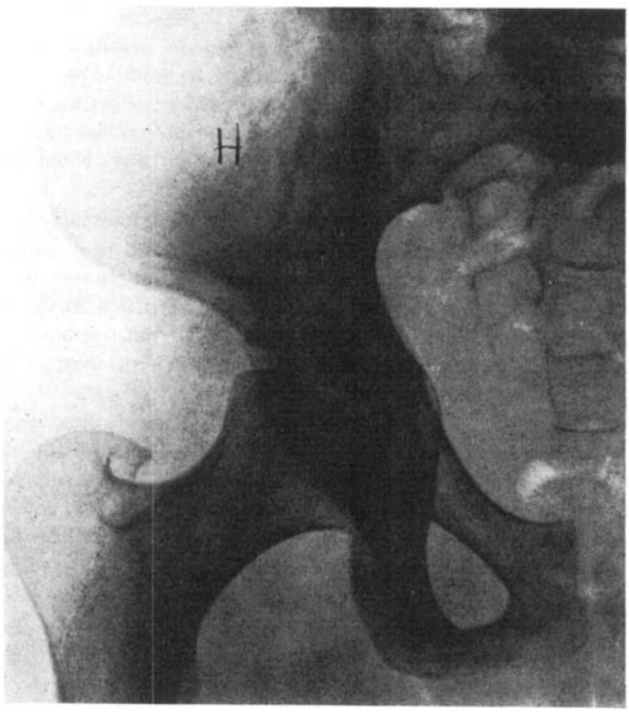

Fig. 8.

(medially) was an area in which the structure was obliterated and where no joint space could be distinguished. In the inferior part of the head it was again possible to follow the outlines of the joint. The structureless region involved the central articular surface and extended more than one centimetre into the head, practically to the old epiphyseal border. It was separated from the normal part of the head by a slightly sclerotic zone nearly a centimetre wide which merged with the normal structure with no sharp line of demarcation. In the opposing region in the centre of the acetabular floor there were also no distinct outlines next the joint and the same lack of normal bone structure. The roof of the acetabulum was slightly sclerotic. On the left side the changes were similar but more

extensive; the peripheral parts of the head were also normal on this side. The changed region was delimited just as indistinctly from the normal structure by a zone of sclerosis. The tear figure was seen only as a narrow sclerotic band doubly outlined in its upper part. The cystoscopy projection also showed distinctly the central position of the changes.

The knee joints were normal.

In frontal views of the feet all the heads of the metatarsals were seen slightly flattened from before backwards. In the middle of the articular surface of the first metatarsal was a shot-sized rarefaction bordered by an extremely narrow sclerotic zone; the joint contour was unbroken over it but appeared to be raised up in a mound sagittally. Nilsonne's index was negative.

Study of the shoulders revealed the head of the humerus to be slightly flattened and broadened medially and from above and to have even joint outlines. The articular surface of the cavity was shallower and flatter than normal and its contour not regular and sharp but wavy. The neck of the scapula ways clumsy and poorly removed from the body. The epiphysis of the tuberculum minus was not fused with the rest of the bone.

The elbow joints were normal.

In the hands the epiphyseal line distally on the radius was normal but the fusion was not complete. The styloid process of the ulna was unusually large, provided with an extra bony nucleus and not yet united with the ulna. The hands were otherwise normal.

The skull was normal and showed no compression of the base. The clivus and sella turcica were normal.

Case III (record 12186). This was the 26 year old brother of the preceding patient. According to the 1927 records made on his first visit to the Orthopedic Hospital, the father reported that the spinal curvature was first noted when the patient was 3 or 4 years old but combined with no fever or other disease. He was treated by a quack for "rickets" when young. At the age af 15 his back became more crooked and also bent to one side, for which reason he was taken to the Orthopedic Hospital in 1927. He then exhibited an untender, fixed kyphosis with a gibbus in the inferior dorsal and upper lumbar spine (see photograph, fig. 9). Diagnosed as "gibbus after spondylitis tuberculosa", he was given a leather corset which he wore one year. From then on his back gave him no trouble except that he felt tired after long walks. Since the age of 12 his hips had felt weak and he limped slightly on the right leg which was somewhat shorter. They caused him no pain until the summer of 1936, when he was 25 years old, at which time the left hip joint began to hurt on movement, the knee joints to feel weak and the anterior part of the thigh to hurt. He had no fever and no feeling of general ill health. He

was treated at a hospital with the usual physical methods, but with only a slight and transient effect. He was again given a corset at Apelviken's Coast Sanatorium in February of 1937.

Clinical examination on Jan. 14, 1937 showed a good general condition. There were no signs of endocrine disturbance. The internal organs were normal. The sedimentation rate was normal. A slight rosary and a sug-

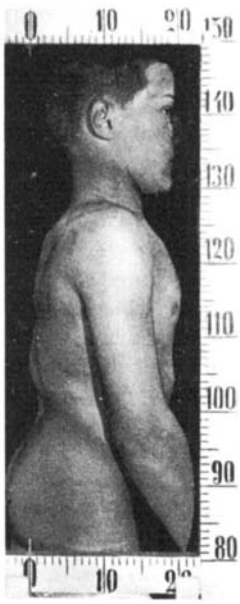

Fig. 9.

gested pectus carinatum indicated the possibility of past rickets. The general configuration of the body corresponded extremely well with that of the brother's just described; the trunk was short, the extremities were of normal length, and there was a long gibbus with its apex over the first lumbar vertebra. Slight acrocyanosis of the hands and feet was noted.

A very slight flexion contracture was found in the right hip; there was about 100 degrees of flexion only, 10 degrees altogether of abduction and adduction and restriction of the rotatory movements to rocking. The left hip showed the same flexion contracture but the leg could only be flexed 45 degrees and both abduction and adduction and rotation were restricted to rocking. The right leg was held slightly adducted in the hip, while the left was somewhat more abducted; the tip of the right trochanter thus felt displaced upwards in comparison with the left, and the

right leg was functionally shortened (there was no real shortening). The feet could not be brought together entirely, 15 cm. remaining between the inner malleoli.

There was no expansion of the articular ends and free mobility in the knee joints. The feet were normal.

There was moderate restriction in the mobility of the shoulder joints, especially of abduction and forward elevation.

Examination of the hands showed a shortage of 10 degrees in extension in all the middle joints of the fingers and some expansion of the articular ends, especially that of the middle phalanx of the third finger.

Roentgen examination (some of the roentgenograms were kindly placed at my disposal by Dr. R. Hanson, Apelviken). The marked kyphosis in the spine was caused by lack of the entire anterior part of the first lumbar vertebra which like a little wedge-shaped hemivertebra jutted from behind between the twelfth dorsal and second lumbar segments; moreover, it was slightly displaced dorsally in relation to those lying above. These three bodies were practically fused into a block concave anteriorly, the middle disk of which could only be seen indistinctly anteriorly. The other lumbar vertebrae were flatter but much deeper (in the sagittal direction) and somewhat broader than normal; in general they resembled very much those previously described, having rounded corners and semicircular, curved horizontal borders (in the lateral views). Next to the intervertebral spaces their contours were faint and irregular and showed numerous uneven rarefactions as in the previous case. The middle vertebrae were also not as high as usual, especially in the neighbourhood of the gibbus. There was spina bifida occulta of the fifth lumbar segment. All the processes were small and poorly developed. In the lumbar region there was a fairly pronounced scoliosis with the convexity to the left. In the dorsal region the borders of the bodies showed the same disturbances in the growing zone. These vertebrae were also flatter and broader than normal and likewise the cervical vertebrae, the anterior parts of which were particularly flat causing a slight kyphos instead of the normal lordosis. There were no other disturbances.

The sacro-iliac joints were normal. There was hardly any translucent zone between the bony outlines of the symphysis, which were moreover faint and blurred. The ischio-pubic junction at the lower border of the foramen obturatum was slightly expanded but the bone was of normal reticulation. This patient had the same shape of pelvis as his brother. The hip joints also showed the same localization of the pathologic changes—the central articular surface of the head. (Fig. 10). On the right side the femoral neck was slender, somewhat short but otherwise normal. The lateral, superior and anterior part of the femoral articular outline seemed normal over only 1 cm. projecting out laterally to the acetabular corner. Central to this centimetre, the outlines of the head

could not be distinguished at all until distal to the centre where they again appeared in the last 3 cm. The central portion was fairly normal in density but the reticulation was blurred and not of normal structure. The opposing part of the floor of the acetabulum was likewise not normally outlined beside the joint and had the same diffuse reticulation. Scattered within this region of the head and acetabular floor were more sclerotic areas alternating with round, cystic areas of resorption. The

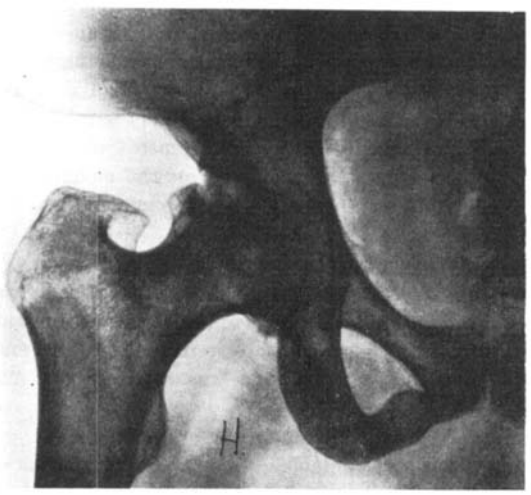

Fig. 10.

superior part of the acetabular roof was fairly sclerotic but it likewise had no distinct joint outlines. The tear figure was preserved but very narrow. If the head is reconstructed from the remaining parts in the periphery, it is seen that centrally, around the fovea, about one-third was lacking but there did not seem to be any corresponding upward displacement of the head in relation to the pelvis. On the left side the changes were of the same type but there was more diffuse involvement of the whole head; the normal borders were only retained a little over one centimetre superiorly and anteriorly, thereafter the joint space was only suggested distally as a line of rarefaction 2 mm. broad, the distal part of which was very indistinct. The whole joint region was diffusely sclerotic and contained areas of cystic resorption up to the size of a hazel-nut which appeared more clearly in the cystoscopy projection. If we again attempt to sketch out the normal contour of the head, it is seen that a slice about 5 mm. broad was missing from practically the

whole head except the infero-medial corner. A little spur lengthened the upper claw of the acetabulum.

The knee joints were normal.

In both feet the head of the first metatarsal had a sagittal eminence in the middle of its articular surface, the articular boundaries were somewhat sharpened and there was a spur on the dorsal aspect of the left foot (as in hallux rigidus). On the right side a rarefaction surrounded by a sclerotic zone was seen on top of the eminence. Nilsonne's index was negative.

Photographs of the left shoulder showed that the head of the humerus was compressed from above medially and as though slightly flattened out on the shaft. The glenoid cavity was shallower than normal, its reticulation irregular and its articular boundaries finely waved; the neck of the scapula was poorly removed. There was a dense spot the size of a hazelnut with no distinct bone structure lying on the medial aspect near the surgical neck. The right shoulder showed the same changes but no shadow of this kind.

The elbow joints were not photographed.

Photographs of the hands showed the distal articular surface of the radius to be extremely oblique and leaning towards the ulna (about 30 degress); the ulna was so short that it did not reach forward to the joint. The styloid process of the ulna was rather large. There were no other pathologic features.

The skull showed no lesions.

A comparison between roentgenograms taken six months apart showed that no change had taken place in the hip joints or spine during this interval.

Case IV (record 10022). This was the 28 year old brother of the preceding patient. The past history was the same as in the foregoing case. In 1925, at the age of 16, he came to the Orthopedic Hospital for the first time together with his youngest brother. The records and roentgen pictures are no longer available. The diagnosis was "bilateral coxa vara after Perthes' disease? and slight scoliosis". In the autumn of 1936 he began to suffer from tiredness and pain after working localized in the upper arms, back and thigh muscles. He could not walk more than one kilometre at a time because of rapidly progressing tiredness.

Clinical examination on. Jan. 19, 1937 revealed an appearance and bodily structure very like those of his brothers (photograph fig. 11). He had a short trunk and a relatively large head. There was a marked gibbus of about 150 degrees whose apex involved three vertebrae. Otherwise the back was practically flat, completely fixed and not tender. He limped very slightly on the right side but did not use a cane.

There was a slight flexion contracture in both hip joints, greater in the right (25 degrees) and his right leg could be further flexed 65 degrees and his left 100 degrees. Abduction was possible to an angle of 35 degrees between both legs, mostly on the left side, and adduction to midposition. Rotation was restricted to rocking on the right side but there was somewhat more outward movement on the left; both legs were held slightly outwardly rotated. There was a slight valgum deformity in the joints

Fig. 11.

of both knees, but the mobility was normal. When weight was placed on the feet there was a slight metatarsus latus but otherwise nothing abnormal.

The scapular musculature on both sides was rather atrophic. In simultaneous movement of both arms he could only abduct 90 degrees, lift forwards 120 degrees and backwards 10 degrees. The rotation was restricted to about one-quarter of the normal outwards and two-thirds inwards. Otherwise the arms and hands were strong and well-shaped. The elbow joints showed a slight shortage of extension, of 15 degrees on the right side and less on the left, but otherwise complete mobility.

Roentgen examination. The general type of the vertebral bodies in the dorsolumbar region was the same as in the preceding cases. The kyphosis was due to defective development of the supero-anterior part of the second lumbar vertebra giving the body a wedge shape; this segment was also slightly displaced dorsally in relation to those above. The intervertebral disks were narrow. Furthermore, frontal views showed

scoliosis with the convexity to the left and the apex at the third lumbar vertebra.

The pelvis had the same typical shape as in the foregoing cases; there was marked protrusio acetabuli on both sides (fig. 12). The symphysis was one centimetre broad and rather irregularly outlined. The ischiopubic junction was normal.

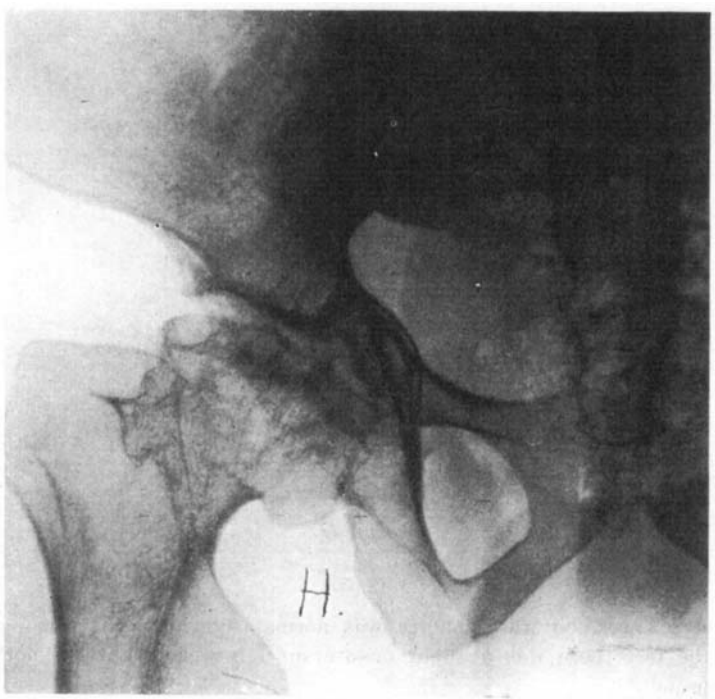

Fig. 12.

The right femoral neck was short and broad but there was no real pressure deformity. The head was clumsily shaped; it seemed rather compressed from above medially. The joint space could be followed nearly the whole way but the contour of the head was indistinct. Right under the surface, lying in the upper two-thirds of the head, were several rounded areas of resorption the size of a pea to a hazel-nut, surrounded by sclerotic zones. The inferior third of the head was fairly distinct. The tear figure appeared only as a narrow line. Spurs were seen on the lateral corner of the acetabular roof and slight sclerosis in its lateral part. In the middle of the floor of the acetabulum directly opposite the

fairly local protrusion, there was an indistinct, nearly bean-sized, rounded spot, surrounded by a zone of rarefaction over a millimetre broad. On the left side the neck was normal. There the head also appeared flattened from the medial side but, as in the preceding case, the normal rounded outlines were retained farthest laterally superiorly and in the inferior part. The joint space was indistinct and narrower than normal but could be distinguished the whole way. As on the right side, the upper two-thirds of the head contained alternating areas of sclerosis and resorption; there was an osteophytic deposit on its inferior corner. The cystoscopy projection also showed clearly the central localization of the process; on both sides the superior and inferior peripheral areas, that is those situated anteriorly and posteriorly respectively on the head, were fairly unaffected.

The left shoulder joint showed a distinct flattening of the humeral head from above medially, but the contour and joint space were normal.

The left elbow joint showed nothing abnormal; full extension was hindered by the unusually prominent olecranon.

In the left foot the heads of the second, third and fourth metatarsals were slightly flattened from before backwards. The distal joint surface of the first metatarsal was likewise flatter than normal with a slightly projecting lateral corner; the opposing corner of the first phalanx was also somewhat pointed. Nilsonne's index was negative.

Case V (record 207/37). This was the 22 year old sister of the preceding patients. She had never had subjective symptoms but her back had always been crooked and one hip projected more than the other.

Clinical examination on Jan. 19, 1937 revealed the patient to be strongly built but under average height due to a slight shortening of the trunk, the extremities being of normal length. She looked healthy and had no external signs of endocrine disturbance. She was four months pregnant.

Her back showed no kyphosis but a fairly pronounced scoliosis with the convexity to the left in the lumbar region and to the right in the dorsal, which could not be corrected passively. The right hip projected more than the left and likewise the left inferior part of the chest. The mobility in the back was moderately restricted. She walked without limping.

In the hip joints flexion was normal, extension (or overextension beyond the midposition) was slightly restricted on the right side and practically impossible on the left, abduction and adduction were about normal in range, inward rotation was normal, outward rotation on the left side moderately restricted and on the right normal.

The knee joints and feet were normal. The joints of the upper extremities were normal.

Roentgen examination. The dorsolumbar spine showed no kyphosis but only a marked scoliosis. The vertebral bodies and intervertebral spaces showed the same general changes as in the foregoing cases; the marginal epiphyses in the inferior dorsal and superior lumbar region were fragmented in places but otherwise normal.

The pelvis was of normal shape. The sacro-iliac joints were normal. The symphysis was ½ cm. broad with very jagged and indistinct borders, and the surrounding area was rather diffusely sclerotic. The ischio-pubic junction was expanded but the bones were fused. The femoral neck was slender on both sides; there was a slight valgus position on the right side. The outlines of the head were normal on the right side except for an area hardly 2 cm. long around the fovea centralis where the normal bony structure was obliterated to a depth of ½ cm. There was no change in the structure of the head in the neighbourhood of this area (no reactive sclerosis or the like). In the opposing region in the floor of the acetabulum the joint contour was likewise lost. The same changes were found on the left side but they involved a somewhat larger area, 3 cm. long and 1 cm. deep in the upper part and situated practically in the centre. It was not so cup-shaped as on the right side but more irregular. Its reticulation was blurred and there were a few pea-sized round clearings. The opposing part of the floor of the acetabulum was likewise somewhat blurred centrally but the outlines were well preserved in the superior and inferior part. There was also no reactive change in the surrounding parts in the head on this side. The height of the joint space was normal. The cystoscopy projection showed the same picture with the changes centrally localized.

Photographs of the left shoulder joint showed the head of the humerus to be perhaps flattened from the medial side but an otherwise normal picture.

Case VI (record 232/37). 9 year old boy, son of the eldest, healthy brother. There were no subjective symptoms at all but his family thought he was a little "saddle-backed".

Clinical examination on Jan. 19, 1937 revealed a healthy-looking boy of average height for his age (118 cm.) and no disproportion between his trunk and extremities.

There was a slightly exaggerated lumbar lordosis in the back which was soft and movable and not tender. The hip joints were likewise freely movable and not painful.

Roentgen examination. In the dorsolumbar spine the vertebral bodies were regularly shaped with normally sharp corners (fig. 13). The reticulation of the cancellous tissue was not normally distinct. The intervertebral spaces were abnormally high (1 cm. in the lumbar region while the adjacent bodies were 2 cm. high in the anterior part; in the middle dorsal

region the corresponding figures were 6 to 7 mm. and 14 mm.). The vertebral bodies were wider sagittally and transversally than normal; in the lumbar region their sagittal diameter was 3 to 3.5 cm., transverse 3.7 to 4.5 cm. and vertical only 1.5 to 2 cm. (Compare these figures with those given by Buchman as normal, making all reservations for their correctness and his other conclusions as well).

The pelvis was of normal configuration, the sacro-iliac joint was

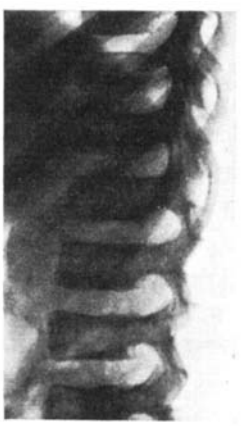

Fig. 13.

normally broad, as was also true of the symphysis, and the ischium and pubis were normally fused. The hip joints showed quite normal conditions and the femoral heads were regularly shaped and outlined.

These cases may be summarized as follows. The patients, normal at birth and with no signs of endocrine disturbance, developed normally until the age of 4 to 6 when tiredness and stiffness began to be felt in the back and hips, and sometimes in the shoulder joints as well. From then on a more or less pronounced disturbance of growth was noted in the spine, clinically manifested in decreased growth of the latter (short trunk but normally proportioned extremities) as well as a kyphosis at the border between the dorsal and lumbar spine. Roentgenologically, the vertebral bodies were flatter but broader than normal with first high, then decreased intervertebral spaces; their outlines became irregular through the very extended gener-

al disturbances in development. The kyphosis was due to local lesions with defects in the anterior corners of the vertebral bodies and a wedging of the vertebrae which were also slightly subluxated dorsally. The pelvis had a characteristic appearance in all the cases; the alae ilii were broad but flat and extended, and as though compressed over the acetabular region; even protrusio acetabuli was often present. As a rule, disturbances in ossification were found in the sacro-iliac joints, in the symphysis and in the junction between the superior ramus of the ischium and the inferior ramus of the pubis. Exactly the same changes in the hip joints were presented by all the cases, ranging from those without clinical symptoms but with typical changes (case V) to the most serious cases (II, III, IV) with great limitation of motion and pain. They were always situated centrally on the articular surface of the head around the fovea centralis and generally appeared at the age of 7 (case I) as a calcium-poor, fragmented segment which out towards the capital epiphysis was surrounded by a very osteoporotic zone. Where this zone bordered on normal bone there was sometimes a slight sclerosis. The changes showed no progress, but remained unchanged for several years (for 2 years and 7 months in case I). About the age of 20 (cases II and V) the fragmentation was lacking; instead, the whole region involved showed a very slight, diffuse structure with no regular outlines beside the joint itself. Later (cases III and IV) secondary changes of the type found in chronic arthritis appeared in the head and acetabulum (which adapted itself to the head) with sclerotic and resorptive areas. In my most severe case, on one side it seemed as though a bean-sized body had been "sequestrated" centrally in the site of the joint space, but it had not the appearance and situation of those found in osteochondritis dissecans. The neck showed no changes. Besides these lesions in the back and hips, several of the cases exhibited restricted mobility in the shoulder joints, roentgenologically seen to be due to faulty development of the glenoid cavity which was shallow and poorly delimited from the neck of the scapula; in addition, the head of the humerus was flattened and showed signs of disturbed ossification. Fur-

thermore there were a number of roentgen-anatomic variations and anomalies such as coarsely reticulated and often fragmented epiphyses (e.g., trochlea humeri, olecranon), extra epiphyses on the tip of the lateral malleolus and styloid process of the ulna (which were also fragmented), increased density in the apophysis of the calcaneus, fragmentation or increase in density of the navicular bone of the hand and pseudo-epiphyses (often fragmented) distally on the first metatarsal and the opposing proximal phalanx.

Two of my cases had acute rheumatic polyarthritis after the appearance of the lesions (lessened local resistance?).

The question is, can these cases be classified under Brailsford's term chondro-osteo-dystrophy? As far as the spinal changes are concerned, the roentgen pictures from case I practically correspond with several of Brailsford's pictures and descriptions (including one of Dale's cases) and, in consideration of the difference in age, also show a great resemblance to cases II and V. The latter in their turn correspond with cases III and IV. Case VI can be grouped with Brailsford's slight cases and should be included because of the familial features—otherwise the clinical and roentgenologic findings alone were hardly enough to permit the diagnosis chondro-osteo-dystrophy.

Cases with such a kyphosis not of inflammatory or traumatic origin are very rare; Gebhardt could collect no more than 7 instances of congenital kyphosis in 1910, of which only 2 were in the dorsolumbar region. But then it was only a question of localized kyphosis and not, as in these cases, of generalized disturbances in the spine as well.

According to Donath and Vogel, in achondroplasia the vertebral bodies in the dorsolumbar region are invariably wedge-shaped, the deformity ranging from only an insignificant difference in height between the anterior and posterior parts in slight cases to entire lack of the anterior part in the most severe —pictures which very much resemble cases II to IV. Unfortunately Donath's and Vogel's publication contains no original pictures but only profile sketches, but they report that the vertebral bodies were otherwise normal, that is, there were no changes

in contour. Their investigations seem to have been given relatively little attention and this finding of wedge-shaped vertebrae is not mentioned as typical of achondroplasia in many later publications, for example, those of Mesz, Fliederbaum and Markuszewiez. In the cases of achondroplasia roentgenologically examined at the Orthopedic Hospital during later years, this wedging has been clearly seen but only to a slight degree and the vertebral bodies have been otherwise normal and not of the shape and type found in the above cases. Further points in the differential diagnosis between achondroplasia and these "atypical" cases, are to be found in Silfverskiöld's articles.

While the changes in the spine in my cases are typical and agree entirely with those described by Brailsford, this is not true of those in the hip joints. According to Brailsford, characteristic in chondro-osteo-dystrophy is a picture like coxa plana with flattening of the head and deformity of the neck and, in some cases lack of the head and even part of the neck, more resembling severe coxa vara congenita (generally a more irregular deformity of the head and a varus position of the neck and not, as in coxa plana, a head flattened medially from above and a short thick neck). Although the cases I have described were very like one another, they showed another picture; the changes were localized to the centre of the articular surface of the head and acetabulum around the fovea centralis; in the severe cases they involved nearly the whole of the head but it was always the peripheral parts which were best preserved and most normal. There was no real pressure deformity; in the severe cases a piece of the head in the changed region looked as lacking but there was no flattening. The neck was unchanged in all the cases except for a slight shortening and thickening on one side in the most severe case. Moreover, the changes were very like in extent and development on both sides in each case. The acetabulum showed the same changes as the head and seemed to have adapted its shape to that of the head. On looking at the roentgen pictures in case I, one thinks first of coxa plana, but closer study reveals characteristic differences. First and foremost there is the localization; coxa plana always begins, as

H. Waldenström has shown, in the antero-superior part of the head, while only at a later stage and only in severe cases is the postero-inferior part and neck attacked and changed; in these cases the localization was central on the apex of the head. Moreover, the diseases run a different course; coxa plana has a characteristic development: first comes the evolution period with the initial stage (lasting 6 months to a year), when only the slight flattening of the head and the increased distance from the head to the acetabular floor appear which, when the resorption progresses, passes on to the fragmenting stage (2 to 3 years) with first increased and later decreased calcium content in the fragments. After the evolution comes the healing period (1 to 2 years) when the epiphysis again becomes homogeneous and the calcium returns, and then the growth period when the joint receives its permanent shape. After the first "acute" stage (evolution) in coxa plana, the surfaces bordering the joint are deformed, it is true, but they are always sharply outlined, because the articular cartilage is always preserved (until secondary arthritis makes its appearance). No such development could be discerned in our cases and, not even at the age of 20 to 25, had any restitution of the structure taken place. Coxa plana generally begins at the age of 5 to 10; bilateral cases to occur but are relatively rare, and then the changes are always of different ages on both sides, that is the developmental stage is never alike on both sides, as it was in all our cases. Familial distribution has been noted in coxa plana but rarely.

The changes in the hip joints deserve special consideration, since they differ both from the ordinary changes in chondro-osteo-dystrophy and from coxa plana. On the other hand, they correspond closely with those in cases published by Kreuz, Freund, Müller and Hetzar and Ribbing. Kreuz' and Freund's cases much resemble each other; the changes in the hip joints were carefully described and agree with my cases. The other bones do not seem to have been systematically examined. However, it appears from their reports that spinal changes were found ("Rundrücken" and scoliosis in one case of an 11 year old girl and a short, scoliotic trunk and normally long extre-

mities in the other, a 15 year old boy), but it is impossible to determine definitely whether they are of the same kind as in my cases. The cases published by Müller and Hetzar are comprised of three brothers and sisters with changes in the hip joints of the type described above. There were also changes in the spine with roentgen pictures showing the same characteristics as in my cases, including rounded vertebrae with curved borders, as well as osteochondritis dissecans in the elbow, knee and shoulder joints. Osteochondritis dissecans in one knee joint combined with typical generalized chondro-osteo-dystrophy is also described by Wright in a 9 year old boy. Judging from the description, Sundt's case also had the same type of changes, both in the back and hip joints (and in several small joints) but roentgen pictures and complete descriptions are lacking. Ribbing also describes similar changes in the hip joints in his thesis. He relates the occurrence in 23 related persons of "anatomic variations and anomalies" (e.g. pseudo-epiphyses distally on the proximal and middle phalanges with an asymmetric trochlea, disturbances in the ossification of the navicular bone, an independent nucleus in the styloid process of the ulna, fragmentation of the epiphysis in the elbow joint) and in 6 brothers and sisters of these 23, the presence of "destructive changes resembling osseous aseptic necroses". These changes in the hip joints and spine were of the same kind as in my cases although not so severe but, on the other hand, they also occurred in other parts of the skeleton. When classifying his cases he distinguishes them from achondroplasia atypica, since, in addition to the roentgenologic disturbances, he considers the latter disease characterized by disproportionate dwarfism with slight micromelia of the upper extremities but normal length of the lower extremities, which often show a marked valgus position, as well as great crookedness of the spine and atrophic musculature. In principle, Ribbing's cases show the same local skeletal changes (see above), but since the patients were well developed individuals with strong muscles, he thinks it necessary to coin a new term for them, namely, "hereditary multiple epiphyseal disturbances". Brailsford's term chondro-osteo-dystrophy inclu-

des cases with slight but typical roentgenologic skeletal changes which often lack any mentionable external deformity. These deformities depend entirely, of course, on the degree of the skeletal disturbances. In the slight cases there are no primary deformities and but slight restriction of motion in the joints, and consequently insignificent if any fault in their position; that is there are no secondary deformities. In the severe cases, on the other hand, deformities appear, some primary through the serious skeletal changes, e.g. kyphosis from wedge-shaped vertebrae, and some secondary through contractures in changed joints and the consequent abnormal placing of weight on other, unchanged joints, e.g. genu valgum in pronounced contractures in the hip joints. When there are contractures and restriction of motion in the joints, there is always muscular atrophy as well.

In practical orthopedic work, therefore, one should not attach too great significance to the external appearance and deformities in differential diagnosis, at any rate, not as much as to the roentgenologic findings. That this is true is shown by a comparison between the "well developed" arms and the atrophic legs of an individual where the roentgenologic disturbances are found primarily in the hip joints, and a comparison between members of the same family with roentgenologic changes of the same kind but of different degree, e.g. case V and cases II to IV, which showed about normal proportions and "disproportionate dwarfism" respectively.

In my opinion, therefore, all the cases cited above, both those previously published and those now described, should be collected into one group which, as long as we have no more adequate term, might just as well be given Brailsford's name, chondro-osteo-dystrophy, a term certainly to be preferred to achondroplasia atypica. Perhaps we may, as suggested by Jaffe and Hirsch, add "eccentro-chondroplastica" to indicate the development of the epiphyses from multiple osseous nuclei. To the characteristic features in the hip joints reported by Brailsford we are now in a position to add, on the basis of our cases, another easily recognized and typical finding, namely, perifoveal

necrosis of greater or lesser extent. The etiology, pathogenesis and connection with local osteo-chondritis (osseous aseptic necroses or local malacias) has been discussed by Ribbing who has adopted Lehmann's theory published in 1922 on constitutionally weak epiphyses and has extended this with interesting anatomic examinations with special reference to osteochondritis dissecans.

Treatment is not of much effect in these cases except in the early stages. Then ordinary orthopedic measures can hinder or at least decrease pronounced pressure deformities in the affected joints, especially in the hips and spine. Since in several cases the pronounced gibbus in the dorsolumbar spine did not appear, or at any rate did not become fixed until fairly late (case I), it should be possible to circumvent it through orthopedic superintendence. If it is already present, attempts can be made in relatively early cases to correct it through successive redressement followed by Albee fixation as recommended by H. Waldenström for tuberculous gibbus. In long-standing cases there remains only symptomatic treatment of the subjective symptoms and training in suitable occupations. However, I should like to point out that Sundal in 1936 described a case with typical roentgenologic changes of the above kind and clinically proportional dwarfism, that is not only shortening of the trunk, without signs of endocrine disturbances which, after treatment with injections of prolan, showed distinct improvement both clinically and roentgenologically. This method of treatment should therefore be given further testing.

ZUSAMMENFASSUNG

Brailfords Begriff der Chondro-osteodystrophie und deren Einteilung wird kurz erörtert.

Beschreibung von 6 eigenen Fällen, darunter 4 Geschwister und 1 Geschwisterkind. Alle zeigen eine normale Entwicklung bis zum Alter von 4—6 Jahren, wo im Rücken und in den Hüftgelenken, bisweilen auch in den Schultergelenken, Ermüdung und Steifigkeit auftreten. Danach zeigt sich eine Wachstums-

störung im Rückgrat — mehr oder weniger ausgesprochener Zwergwuchs mit kurzem Rumpf, aber normal langen Extremitäten, sowie Kyphose zwischen Brust- und Lendenwirbeln. Röntgenologisch sind flache und breite Wirbelkörper mit unregelmässigen Konturen charakteristisch, ausserdem oft Keilwirbel infolge von Defekten in den vorderen Rändern. Das Becken ist ebenfalls typisch mit niedrigen, breiten, auseinandergebogenen Alae ilii, die über der Acetabularregion gleichsam zusammengedrückt sind (oft eine wirkliche Protrusio acetabuli), ausserdem Verknöcherungsstörungen. Die Hüftgelenke zeigen bei allen Ptt. gleichartige Veränderungen, die zentral um die Fovea capitis lokalisiert sind, und die im Alter von 7 Jahren in einer kalkarmen Fragmentation bestehen; etwa im Alter von 20 Jahren fehlt die Fragmentation in dem nach wie vor undeutlich abgegrenzten Gebiet. In ein paar Fällen fanden sich ähnliche Veränderungen im Schultergelenk. Ausserdem kommen zahlreiche röntgenologische Variationen und Anomalien vor mit fragmentierten, bisweilen verdichteten, oft Extraepiphysen.

Differentialdiagnostische Besprechung der Fälle unter besonderer Berücksichtigung von Rückgrats- und Hüftgelenksveränderungen. Die ersteren stimmen völlig mit den von Brailsford u. a. beschriebenen überein. Die letzteren unterscheiden sich davon und bilden eine deutlich abgegrenzte Gruppe. Nähere Beschreibung dessen, was für diese Gruppe typisch ist und wodurch sie sich insbesondere von der Coxa plana unterscheidet. Hinweis auf ähnliche, früher veröffentlichte Fälle, besonders die Ribbing'schen »hereditären multiplen Epiphysenleiden«. Kurze Auseinandersetzung, warum alle diese Fälle als gleichartig angesehen werden, und warum die röntgenologischen Veränderungen entscheidend sind.

Durch Zusammenstellung der beschriebenen Fälle mit einigen früher veröffentlichten kann man also den von Brailsford beschriebenen Gruppen von Veränderungen bei Chondro-osteodystrophie (die in Ermangelung einer mehr adäquaten Bezeichnung als Sammelname akzeptiert wird) eine Gruppe mit mehr oder weniger ausgedehnter zentraler, perifovealer Nekrose des Caput femoris angliedern, die sich durch Lokalisation, Aussehen und

Verlauf sehr deutlich von anderen Hüftgelenksleiden unterscheidet.

Die Behandlung soll in einer orthopädischen Prophylaxe gegen die Ausbildung hochgradiger Deformitäten bestehen, eventuell in Hormonbehandlung trotz des Fehlens äusserer Zeichen einer endokrinen Störung.

SUMMARY

A brief account is given of the concept set up by Brailsford as chondro-osteodystrophy and its classification.

Description is given of 6 cases observed by the author; 4 of these patients are children of the same parents, and 1 is their cousin. They all showed normal development up to the age of 4—6 years, when tiredness and stiffness began to appear in the back and hip-joints, sometimes also in the shoulder-joints. After this, disturbances of growth became manifest in the vertebral column: more or less pronounced dwarfishness with short trunk and extremities of normal length, besides kyphosis of the dorsal and lumbar spine. Roentgenologically characteristic are the flattened and broad vertebral bodies with irregular contours; often the vertebral bodies are wedge-shaped from defects in the anterior margins. Also the appearance of the pelvis is typical: low, broad and flaring alae ilii, apparent compression of the hip over the acetabular region (often true protrusion of the acetabulum), and disturbances of ossification. In all the patients the hip-joints show the same kind of changes localized round the fovea capitis, at the age of 7 years consisting of calcium-low fragmentation; at the age of about 20 years no fragmentation can be made out in this poorly defined blurred region. In a couple of cases similar changes were found in the shoulder-joint. Furthermore, there are many other roentgenographic variations and anomalies with fragmentary, sometimes condensed, often additional epiphyses.

The differential diagnosis is reviewed with special reference to the changes in the vertebral column and hip-joints. The former fully agree with the findings described by Brailsford

and others. The latter, on the other hand, differ from previous findings and constitute a well-defined group. A detailed account is given of the features typical of this group, and through which this condition is differentiated in particular from coxa plana. Reference is made to similar cases reported previously, especially Ribbing's "hereditary epiphyseal lesion". The reasons are given why all these cases are looked upon as being of the same nature, and why the roentgenological changes are decisive.

Through the cases here described and correlated with other cases, published previously, it is practicable in addition to the groups of changes described by Brailsford as chondro-osteodystrophy (in the want of any better, this designation is adopted as a collective term) to add another group with central, perifoveal, necrosis of the head of the femur, more or less extensive, which in localization, appearance and course is well differentiable from other lesions of the hip-joint.

The treatment ought to consist in orthopedic prophylaxis as to development of pronounced deformity, possibly also hormonal therapy—even in the absence of external signs of endocrine disturbances.

RÉSUMÉ

Il est rendu brièvement compte de la conception établié par Brailsford sur la chondro-ostéodystrophie et sa classification.

Description de 6 cas observés par l'auteur, dont 5 enfants d'une même famille (4 frères et soeurs et 1 cousin). Tous les enfants s'étaient développés normalement jusqu'à l'âge de 4 à 6 ans, où ils commencèrent à ressentir de la fatigue et de la raideur dans le dos et les articulations des hanches, parfois aussi dans les épaules. Par la suite, des troubles de croissance se manifestèrent dans la colonne vertébrale, avec signes plus ou moins prononcés de nanisme, tronc court et extrémités de longueur normale, ainsi que cyphose entre la colonne dorsale et lombaire. Comme traits caractéristiques radiographiques, on relève: corps vertébraux plats et larges, aux contours irréguliers, souvent vertèbres en coin en raison de défectuosités sur les bords antérieurs. Le bassin est également

typique avec alae ilii bas, larges, courbés, qui sont également comprimés dans la région acétabulaire (apparaissent souvent comme une véritable Protusio acetabuli) ; d'autre part, troubles de l'ossification. Chez tous ces malades on constate le même genre de modifications localisées autour de fovea capitis, consistant à l'âge de 7 ans en une masse fragmentaire pauvre en chaux. Vers l'âge de 20 ans, on ne voit plus de fragmentation dans cette région indistinctement délimitée. Dans deux des cas on trouva des modifications similaires dans l'épaule. D'autre part, beaucoup d'autres variations et anomalies radiographiques avec épiphyses fragmentées, parfois condensées, souvent supplémentaires, ont été constatées.

Examen des diagnostics différentiels par rapport en particulier aux modifications de la colonne vertébrale et de la hanche. Les premiers concordent entièrement avec les faits décrits par Brailsford et d'autres. Les derniers diffèrent et forment un groupe bien distinct. Il est donné un compte rendu détaillé des caractéristiques de ce groupe, en indiquant ce qui le différencie spécialement de Coxa plana. L'auteur se réfère aux cas publiés antérieurement, notamment aux »lésions héréditaires multiples de l'épiphyse« de Ribbing. Il indique ensuite les raisons pour lesquelles il considère que tous ces cas sont analogues et pourquoi les modifications radiographiques sont décisives.

En comparant les cas décrits ici avec quelques-uns de ceux publiés antérieurement, on peut ainsi ajouter aux groupes des modifications décrites par Brailsford comme chondro-ostéodystrophie (à défaut d'une désignation plus adéquate, ce terme est pris dans un sens collectif), un groupe nouveau avec nécrose centrale, périfovéale de caput femoris, d'une extension plus ou moins grande qui, par sa localisation, son aspect et son évolution se différencie nettement des autres lésions de la hanche.

Il convient d'appliquer un traitement prophylactique orthopédique pour empêcher que se développe un haut degré de difformité en l'accompagnant éventuellement d'un traitement aux hormones, même s'il n'y a pas de signes apparents de troubles endocriniens.

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For further references see Brailsford's book named above.