OSTEOPSATHYROSIS: REPORT OF A CASE
WITH ROENTGENOGRAMS OF ELEVEN DIFFERENT FRACTURES
IN THE SAME PATIENT *

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The accompanying case of osteopsathyrosis was treated and studied in the surgical outpatient department of the Children's Hospital, Boston. The accompanying series of roentgenograms gives a graphic fracture history of the patient from the time he was 9 months old until his death, at 7½ years.

In spite of the large number of cases reported in the literature, osteopsathyrosis, or fragilitas ossium, is a relatively uncommon cause of pathologic fractures. Griffith¹ and Ostheimer² had collected 193 cases in all up to 1913, and since then a few cases have been reported. In 1913 one case was reported by Schwarz and Bass,³ and another by Bamberg and Huldshinsky.⁴ In 1914 Bookman⁵ reported one, and the following year Kienböck⁶ collected thirty-seven cases from the literature (from 1888 to 1914) and added two cases of his own. Blaine⁷ reported one case in 1916, and, in 1917, Hess⁸ presented four cases and Mixsell⁹ two. In 1920 Ruggeri¹⁰ reported one, and in 1921 Vander

* From the surgical service of the Children's Hospital, Boston, under the direction of Dr. James S. Stone. The roentgenogram reproduced in Figure 12 is used through the courtesy of the Massachusetts General Hospital.

1. Griffith, J. P. C.: *The Diseases of Infants and Children*, Philadelphia, W. B. Saunders Company **2**:432, 1919.

2. Ostheimer, Maurice: *Fragilitas Ossium*, J. A. M. A. **63**:1996 (Dec. 5) 1914.

3. Schwarz, Herman, and Bass, M. H.: *Osteogenesis Imperfecta: Report of a Case with Study of Its Metabolism*, Am. J. Dis. Child. **5**:131 (Feb.) 1913.

4. Bamberg, K., and Huldshinsky, K.: *Ueber angeborene Knochenbrüchigkeit*, Jahrb. f. Kinderh. **78**:214, 1913.

5. Bookman, A.: *The Metabolism in Osteogenesis Imperfecta with Special Reference to Calcium*, Am. J. Dis. Child. **7**:436 (June) 1914.

6. Kienböck, R.: *Ueber infantile Osteopsathyrose*, Fortsch. a. d. Geb. d. Röntgenstrahlen, Hamb. **23**:122, 1915.

7. Blaine, E. S.: *Idiopathic Infantile Osteopsathyrosis*, Am. J. Roentgenol. **3**:438 (Sept.) 1916.

8. Hess, J. H.: *Osteogenesis Imperfecta*, Arch.Int. Med. **19**:163 (Feb.) 1917.

9. Mixsell, H. R.: *Osteogenesis Imperfecta with Report of Two Cases*, Arch. Pediat. **34**:756 (Oct.) 1917.

10. Ruggeri, E.: *Idiopathic Osteopsathyrosis*, *Pediatrics* **28**:953 (Oct. 15) 1920.

Veer and Dickinson¹¹ reported one. There have doubtless been others that I have overlooked. In other words, more than 200 cases of osteopsathyrosis have been reported. Contrasted with this apparently large group, we find that in a series of 45,000 births in the Göttingen Frauenclinic, Birnbaum¹² found only one case of fragilitas ossium. Again, in a series of 1,610 long bone fractures (including the clavicle) treated at the Massachusetts General Hospital, only two fractures due to fragilitas ossium are recorded.¹³

REPORT OF CASE

History.—The family history was of no significance. The father, mother and five other children were living and well. There was no history of bone disease, fractures or blue sclerae anywhere in the family. The patient was a full term baby and was delivered normally. He was breast fed for fourteen months. He had had measles, but no other illnesses, except for the fractures he had sustained. His lower, central incisor teeth were present at birth. About a year ago they had come out, and none had replaced them.

When 9 months old, the patient fell, fracturing the left femur just above the condyles (Fig. 1). A plaster spica was applied in the surgical outpatient department of the Children's Hospital, and union resulted, with practically no deformity.

When 21 months old, he was jumping on the porch steps, when he began to cry and to hold up his right leg. He did not fall, but he would not use his right leg after that. The roentgen ray revealed a fracture at about the junction between the upper and middle thirds of the right femur, with slight anterior bowing (Fig. 2). He was admitted to the hospital, where a plaster spica was applied with a pad over the site of the deformity. At that time both femurs showed some anterior bowing, which Dr. James S. Stone then attributed to rickets (Fig. 3). A small cystic rarefaction was noted at the lower end of the left femur.

Soon after discharge from the house, in the following month, the roentgen ray revealed shadows about the site of the fracture, which were rather more extensive than might be expected from callus formation alone (Fig. 4 A). Three months later he was again admitted because of a definite tumor mass that had formed at the point of fracture. Because of the possibility of new growth, the tumor mass was excised. Pathologic examination, however, demonstrated the specimen to be callus and not new growth (Fig. 4 B). At about this time, the patient complained of inability to flex the right forearm, but there were no clinical signs of fracture. No roentgen-ray examination was made. After immobilizing the arm with an internal angular splint for a few days, all symptoms disappeared.

When 2 years and 9 months old, the patient complained of some pain in the left thigh. The roentgen ray disclosed slight raising of the periosteum along the middle third of the left femur (Fig. 5). A plaster spica was applied and kept on for two months, after which time all symptoms abated.

11. Vander Veer, E. A., and Dickinson, A. M.: *Fragilitas Ossium*, Ann. Surg. 74:629 (Nov.) 1921.

12. Birnbaum, R.: *A Clinical Manual of the Malformations and Congenital Diseases of the Foetus*, Philadelphia, P. Blakiston's Son & Co., 1912, p. 246.

13. MacDonald, R. F.: Unpublished series of pathologic fractures.

When 3 years old, he fell to the floor from a standing position. His left arm swelled considerably; but the swelling disappeared within a few days, and the function seemed practically normal again. A roentgenogram, taken one week later, revealed a fracture of the left radius, with some lateral displacement, but no overriding (Fig. 6.). Good union was obtained with antero posterior splints.

When he was 4 years old, he fell again, and a green-stick fracture of the left radius resulted (Fig. 7). This fracture united quickly without deformity, after splinting.

Fig. 1.—Fracture of the distal end of the left femur at 9 months (the patient's first fracture).

At 4 years and 5 months, he fell from the davenport, alighting on his right arm. The upper third of the right humerus was fractured with comparatively little displacement (Fig. 8). Good union was obtained by means of a traction splint, after a triangle in the axilla had failed.

When nearly 5 years old, the boy fell from a cart onto his right shoulder, fracturing the right clavicle about the middle (Fig. 9). This break united without deformity, a Velpeau bandage being used. At the same time, roentgeno-

grams were made of both tibias, because of the bowing they presented (Fig. 10). This bowing was interpreted by Dr. Percy Brown as being due to rickets.

When he was nearly 7, the patient fell on the left arm, breaking both bones of the forearm (Fig. 11). He was treated in the outpatient department of the Massachusetts General Hospital, and good union was obtained, although marked bowing of both bones remained.

At 7 years and 3 months, he fell over backward, his weight falling on his hyperextended palms. Swelling appeared over the flexor tendons of the left wrist, and flexion and extension became painful. The swelling and tenderness

Fig. 2.—Fracture of the right femur at 1 year and 9 months.

disappeared in two or three days, but some ecchymosis remained. Slight mobility was detected on palpation, and the roentgen ray demonstrated that a small crack had been opened up at the site of the last radial fracture (Fig. 12). This injury was treated with a posterior splint and pads and adhesive straps to correct the bowing, with fairly good results. Roentgenograms of all the long bones, the skull and the chest were taken at the same time (Figs. 13, 14, 15, 16 and 17). The long bones of the extremities all showed anterolateral

bowing, with marked thickening of the cortex of the diaphysis and relative thinning at the epiphysis. The chest showed nothing remarkable, and the skull showed only moderate thinning.

At 7½ years, the patient tripped over a rug and fell on his right arm. The roentgen ray revealed no evidence of fracture. Nine days later he again fell on the floor at home, breaking the right humerus at about the junction of the middle and upper thirds. The roentgen ray disclosed a clean, right-angled break, with practically no displacement (Fig. 18). He walked into the outpatient department that morning. The arm was immobilized with coaptation

Fig. 3.—Appearance of the left femur at the time of fracture of the right femur, shown in Figure 2. A cystic area is seen in the lower third.

and an internal angular splint. He was kept under observation until late in the afternoon, and, as there was no evidence of other injury, he was allowed to go home. During the evening he vomited several times and was seen by his local physician, who made a diagnosis of fractured skull, sending him to the hospital. On the way to the hospital, he is said to have died from intracranial hemorrhage. Necropsy was not performed.

Physical Examination.—At no time did examination reveal anything remarkable about the patient except bowing of the extremities and slight deformities due to the various fractures. As shown in Figure 19, the boy was well proportioned and of about normal size, with a slightly flattened chest and a rather

prominent abdomen. All of the extremities showed moderate outward bowing. The lower, central incisor teeth were absent, and the alveolar process was much thinned out at that point. There was no evidence of the presence of permanent incisors in the alveolar process to replace the missing teeth. The sclerae were of normal color and did not show the dark bluish tint which has often been described in patients with osteopsathyrosis.

Summary.—The patient was a boy, aged 7½ years, with negative family history, who had sustained multiple long bone fractures, and who died following what was probably a fractured skull. Eleven fractures were demonstrated by the roentgen ray, and there were two other probable fractures which were not

Fig. 4.—*A*, appearance of femur one month after the fracture shown in Figure 2, showing the excess of callus formation at the site of fracture; *B*, four months after the fracture. The apparent tumor at the site of fracture proved to be callus.

roentgenographed. The fractures were distributed thus: left radius, 4; right humerus, 2 (3?); left femur, 2; right femur, 1; right clavicle, 1; left ulna, 1, and skull, 1 (?).

All of the fractures were the result of comparatively slight trauma, and all, with the exception of those immediately preceding death, united readily. The deformities resulting from fractures were insignificant; but the long bones of all the extremities showed a certain amount of outward bowing, from infancy. One of the early fractures showed excessive callus formation, and in one other case a small cystlike rarefaction of the bone was noted.

TERMINOLOGY

Many names have been applied to this condition which is characterized by congenital bone fragility. Osteopsathyrosis, fragilitas ossium and osteogenesis imperfecta are the terms in common use in the English literature. "Idiopathic osteopsathyrosis" is accepted by most of the

Fig. 5.—Fracture of the left femur at 2 years and 9 months, shown here by raising of the periosteum. Both femurs show considerable bowing and flaring at the epiphyseal line.

continental writers. It has been pointed out that "fragilitas ossium" accentuates the chief clinical symptom of the disease, while "osteogenesis imperfecta" suggests the etiologic factor of imperfect bone formation.² Practically all writers use osteopsathyrosis and osteogenesis imperfecta

synonymously, but Griffith¹ reserves the latter name for cases of multiple intra-uterine fractures, together with imperfect development of membrane bone.

CLASSIFICATION OF CASES

Nichols¹⁴ divides *fragilitas ossium* into two groups: (1) symptomatic fragility, in which fragility is due to one of various known pathologic processes, as rickets, old age, osteomyelitis, tuberculosis,

Fig. 6.—Fracture of the left radius in its upper third at 3 years.

tabes, syringomyelia, paralysis, malignant disease, bone cysts or echinococcus cysts, and (2) "idiopathic fragility," which is identical with osteogenesis imperfecta. Osteimer describes two types of cases: (1) the fetal type found either at birth or shortly thereafter, with imperfect bone formation and multiple fractures (imperfect osteogenesis or periosteal dysplasia). Frequently these patients do not

14. Nichols, E. H., in Keen: *Surgery: Its Principles and Practice*, Philadelphia, W. B. Saunders Company 2:56, 1914.

survive after the first year. (2) The congenital type found in infants and children, at birth or later, with fractures recurring often until puberty or even adult life, associated with similar bone changes (idiopathic osteopsathyrosis). These patients, with care, will survive but are often badly crippled. Locke¹⁵ adds a third, the adult form, encountered as an idiopathic fragility of the bones after childhood.

Fig. 7.—Fracture of the left radius in its lower third at 4 years.

ETIOLOGY

No definite etiology for osteopsathyrosis has been determined. It occurs about equally in males and females. The hereditary factor has been recognized by many writers,¹⁶ and the incidence of osteopsathy-

15. Locke, E. A.: Oxford Medicine, New York, Oxford University Press 4:452, 1921.

16. Conard, H. S., and Davenport, C. B.: Hereditary Fragility of Bone, Bull. 14, Eugenics Record Office, Cold Spring Harbor, N. Y., November, 1915. (Includes a complete bibliography.)

rosis in persons with blue sclerae is sufficiently high to suggest that there is a definite relation between the two conditions.

The principal theories advanced as to the etiology are: (1) The disease is an affection of some central organ (osteotrophic organ) which furnishes some substance necessary for the normal formation and nutrition of bones; " (2) the disease is of nervous origin,¹⁷ and (3) it is due to some disturbance of nutrition affecting the entire bone system during fetal life.¹⁸

Fig. 8.—Fracture of the right humerus at 4 years and 5 months.

PATHOLOGY

The disease apparently affects only the bones, as no constant pathologic condition has been found elsewhere in the body.

Growth of the bones in length usually proceeds normally, but the bones may appear to be much shortened because of bowing and angulation resulting from fractures. There is no constant gross deformity.

17. Biggs, M. H.: Univ. Penn. M. Bull. **15**:488, 1902-1903.

18. Harbitz, F.: Beitr. z. path. Anat. u. z. allg. Path. **30**:605, 1901.

In some cases, the bones have been described as being thin and delicate; but in many cases (as in the case described herewith), the bones appear to be proportioned normally. The extent of deformity usually depends on the number of fractures previously sustained, although (as cited above) there is often an intrinsic bowing of the long bones, entirely independent of traumatic deformities. This bowing is usually not so pronounced as that of rickets, and is confined to the diaphysis, in contrast to the epiphyseal deformities in rickets.

Fig. 9.—Fracture of the right clavicle at 5 years.

The skull is usually normal in size and shape in the older cases. In the early, fetal cases, the skull may be only a membranous sac with islands of calcification.

Microscopically, the bone usually shows a cortex which is thin and friable with occasional islands of cartilage. Trabeculae are often imperfectly formed or absent. Nichols¹⁴ says that instead of osteoblasts depositing new bone on a cartilaginous matrix, the trabeculae are formed by calcification of cartilage cells. The dense bone formed is nonlaminated or imperfectly laminated, with no haversian canals but with large marrow spaces in which there are few trabeculae. The formation of periosteal bone is also abnormal and incomplete. Several writers have called attention to the fact that in this disease there is

increased absorption of bone but very scant bone proliferation. There is a difference of opinion concerning the relative amount of calcium salts present, it not being definitely determined that there is a lack of calcium in patients with osteopsathyrosis.

Fig. 10.—Marked anterior bowing and thickening of the midportion of the diaphyses of the bones of the lower leg at the time of fracture of the humerus, shown in Figure 9.

SYMPTOMATOLOGY

Aside from the tendency toward frequent fractures, there are practically no constant symptoms.

In some of the earlier cases, the appearance of the patients conforms more or less to type. They have a relatively large head, small chest,

Fig. 11.—Fracture of both bones of the left forearm at 7 years.

Fig. 12.—Refracture of the left radius at 7 years and 3 months, at the site of the previous fracture.

and a protuberant abdomen. The later cases, however, may show no recognizable peculiarities of form.

As has already been mentioned, in some patients there is a dark blue tint in the sclerae, which is said to be due to deficiency of the sclerotic fibrous tissue, allowing the pigment of the choroid to show through.

Character of the Fractures.—As the name suggests, the most characteristic symptom of the disease is the extreme brittleness of the bones

Fig. 13.—Characteristic deformity of the bones of the arm, three months before the patient's death.

and the ease with which fractures are sustained. The trauma is usually slight, and very often the signs of fracture are meager. Instances have been cited in which even the most careful handling of diseased infants or children caused fractures, and in other cases a mere handshake has been enough to break the forearm. Very little pain, tenderness or swelling is attendant on these fractures, mobility or deformity often being the only sign found. Frequently the child will show nothing

more than unwillingness to use the affected arm or leg. The tendency to fracture is bilateral, and both sides of the body are usually affected sooner or later. The long bones of the extremities, the clavicles and the ribs are the most common seats of fracture; but the other bones of the

Fig. 14.—Characteristic deformity of the femurs, three months before the patient's death.

body are occasionally involved. The femur is the one bone most commonly broken in osteopetrosis (Locke). Fracture of the skull is comparatively rare.

Kienböck states that the periosteum is rarely ruptured. The callus forms in the usual way and is variable in amount. It is generally said

that these fractures unite more readily than those in normal bone, but Kienböck asserts that the healing may not be complete for several months. In support of his contention may be mentioned the fact that refracture occasionally occurs at the site of previous injury, several months after union is thought to be complete. Such an instance is cited in the case described herewith. Certainly most of the fractures in this case were followed by apparently firm union in shorter time than that

Fig. 15.—Characteristic deformity of the bones of the leg, three months before the patient's death.

required for most normal bones. Kienböck also asserts that the callus may be considerably hypertrophied, an instance of which peculiarity was noted above, following one of the early femur fractures (Fig. 4 *B*).

The number of fractures varies greatly, but few patients have less than ten. Terry¹⁹ reports the case of a man, aged 42, with more than

19. Terry, W. L.: Hereditary Osteopsathyrosis, *Ann. Surg.* **68**:231 (Aug.) 1918.

sixty fractures. Mixsell records the case of Chaussier with 113 fractures. According to Emerson,²⁰ the tendency to fragility is lost if the patient lives to be 30; but he mentions one case in which the disease continued to the seventieth year.

Roentgen-Ray Appearances.—Locke¹⁵ has admirably summarized the roentgen-ray findings noted by various authors, as follows: “(1) a high degree of osteoporosis uniformly affecting the entire skeleton,

Fig. 16.—Appearance of the chest of the patient, three months before his death.

the variations in density giving a mottled appearance; (2) marked deformities of the long bones of the extremities, usually of the angular type, due to fractures, rarely also some actual bowing; (3) apparently normal size and shape of bones, except as altered by fractures and resulting callus formation; (4) often excess callus formation

20. Emerson, C. P., in Osler and McCrea: *System of Medicine*, New York, Oxford University Press 6:723, 1909.

with variable degree of calcification. A transverse line of decalcification is often seen at the point of previous fracture; (5) the epiphyseal line is straight, cartilage is always thin; (6) a very faint shadow is given by all the bones, frequently scarcely more dense than that of the surrounding soft parts; (7) the cortex is thin, irregular and deficient in salts; (8) the spongiosa of both epiphysis and diaphysis is extremely faint with entire absence of normal markings; (9) the medullary cavity

Fig. 17.—Appearance of the skull of the patient, three months before his death.

is very irregular in outline and dilated." These changes are considerably less marked in some of the milder cases, such as the one described above.

Metabolism.—Several interesting metabolic studies on patients with osteopsathyrosis have been made within the last few years. While these studies have not been conclusive, they have suggested that calcium

metabolism may have some bearing on this type of bone fragility, as well as on the deformities of rickets. They have also suggested a method of treatment which may prove to be helpful.

Bookman²¹ reported studies on a man, aged 27, with osteopsathyrosis, who had a healed fracture, five weeks before the time of the experiment. He found in this patient a markedly negative calcium balance (loss of 0.019 gm. of calcium monoxid per kilogram of body

Fig. 18.—Fracture of the right humerus at 7 years and 6 months. On comparing this illustration with Figure 8, it will be noted that the cortex of the diaphysis is relatively thinner than it was at the time the earlier roentgenogram was taken.

weight), and likens his to similar findings in osteomalacia. The negative balance is explained by the fact that in both conditions calcium from the already formed bone is being liberated and thrown out of the system.

21. Bookman, A.: The Metabolism in a Case of Idiopathic Osteopsathyrosis, *Arch. Int. Med.* **8**:675 (Nov.) 1911.

Schwarz and Bass³ studied the metabolism of a 7 months old infant with osteogenesis imperfecta, who had had no fractures subsequent to the fourth day after birth. They found a practically normal, or positive, calcium balance (with retention of 0.109 gm.).

Fig. 19.—Appearance of patient at 7 years and 3 months.

Bamberg and Huklschinsky's⁴ case showed a variation from a loss of 0.019 to a gain of 0.136 gm., the latter figure being obtained while the patient was under treatment with cod liver oil and phosphorus.

Bookman,⁵ in his studies on a 3 months old infant with osteogenesis imperfecta, also found a low calcium retention, which, however, varied with the course of the disease. He also found that the deficient reten-

tion of calcium was favorably influenced by the administration of cod liver oil and phosphorus, and still more by calcium lactate. He believes that the similar results obtained in rickets suggest an etiologic relationship to osteopsathyrosis.

Ruggeri¹⁰ studied the metabolism in a girl of 13, with a history of twelve fractures. He found that she had a daily high deficit in the phosphorus, calcium and magnesium balance. This balance was improved for a short time by the administration of thymus and suprarenal extracts, but gradually the condition returned to its original status within a few months.

PROGNOSIS

The outlook in cases of osteopsathyrosis is poor, in spite of the fact that a few patients have grown to adult life, apparently overcoming the bone fragility from which they suffered in earlier years. Most of the patients that survive are deformed by earlier fractures. Few fractures occur after the patient is in the early twenties, and after that time the prognosis is fairly good. Below that age, however, the mortality is high. The mortality is almost 100 per cent. in the fetal type, the majority dying in utero.

DIAGNOSIS

The diagnosis is usually perfectly clear, because there is no other congenital disease in which multiple fractures occur. Rickets and syphilis occasionally cause fractures in young children; but rarely more than two or three fractures occur in the same person as a result of these affections.

TREATMENT

The fractures are treated in the usual way, great care being taken to prevent distressing deformities, by preserving good alinement.

The metabolic studies already mentioned would point to the efficacy of cod liver oil and phosphorus, and in some cases they have seemed beneficial. Hess⁸ uses a dose of 4 c.c., twice daily, of either of the following preparations: phosphorus, 0.01 gm., and cod liver oil, 60 c.c., or pure tribasic calcium phosphate, 6 gm., and cod liver oil, 60 c.c. Czerny²² reports benefit from the use of 100 gm. daily of raw carrot juice, on the theory that osteopsathyrosis is an avitaminosis.

Care of the patient to prevent fractures is the most important single factor in the treatment of osteopsathyrosis.

22. Czerny, A.: *Berl. klin. Wehnschr.* **61**:310, 1919; *Deutsch. med. Wehnschr.* **45**:259, 289 (March 6) 1919.