

Sclerosteosis Involving the Temporal Bone: Clinical and Radiologic Aspects

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To my former teachers, Dr. Samuel J. Crowe, Dr. Stacy R. Guild, Dr. John E. Bordley, Dr. Howard P. House, and Dr. John R. Lindsay.—G.T.N.

Sclerosteosis is one of the rare, potentially lethal, autosomal recessive, progressive, craniotubular sclerosing bone dysplasias. Syndactyly of the second and third or other fingers is evident at birth. Hyperostosis and sclerosis are most prominent in the skull and tubular bones, and are frequently associated with excessive height and weight. The typical facial deformity is apparent by the age of 5 years. The changes involving the temporal bone include a marked increase in overall dimensions, extreme sclerosis, and narrowing and constriction of the external ear canal, middle ear cleft, internal acoustic meatus, and fallopian canal. Impairment of hearing, as a rule bilateral, is a frequent presenting symptom which may manifest in early childhood. Initially it is an expression of interference with sound conduction; later it may become associated with a loss of sound perception. Impairment of facial nerve function is another salient feature which occasionally is present at birth. As a rule, it manifests initially as a unilateral, recurrent paresis, eventually progressing to a bilateral permanent partial loss of facial nerve function. Since impairments of hearing and facial nerve function are two of the salient features, present at birth or in early childhood, the responsibility for recognizing the disease often falls upon the otolaryngologist. The clinical and radiologic features permit not only early recognition of the disorder but also differentiation from similar bony dysplasias. Hyperostosis and sclerosis of the skull lead to thickening and distortion of the calvaria, cranial base, and foramen magnum resulting in reduction of the intracranial volume, interference with the cerebral blood flow, resorption of cerebrospinal fluid, and gradual increase of intracranial pressure. Severe headaches resulting from this mechanism often develop in early adulthood, and several patients have died suddenly from impaction of the medulla oblongata in the foramen magnum. Decompression of the transverse sigmoid sinus and jugular bulb may be lifesaving, combined with a posterior, and if necessary, an anterior, craniectomy for decompression. Early decompression of the internal acoustic meatus and fallopian canal may help in the preservation of cochlear and facial nerve function. (Key words: bone dysplasia; congenital anomalies; sclerosteosis; van Buchem's disease.)

DEFINITION

Sclerosteosis is a rare, autosomal recessive, potentially lethal skeletal disorder in which a poorly understood abnormality of bone remodelling results in generalized sclerosis and

hyperostosis of the cranium and cortex of the tubular bones. The condition usually manifests at birth as syndactyly, usually bilateral, but asymmetrical; cutaneous, rarely osseous; regularly involving the second and third fingers, often the third and fourth fingers, and occasionally also the first and second fingers. One or more fingers may be foreshortened and deformed, and they often have a radial deviation of the terminal phalanges. The terminal phalanges may be kinked and show various forms of dystrophy of the fingernails. There may be partial cutaneous webbing of the second and third toes of the feet. Accelerated skeletal growth begins in early childhood, often leading to gigantism and

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to characteristic enlargement and distortion of the calvaria, mandible, clavicles, ribs, and long bones. Sclerosis and hyperostosis are most prominent in the skull and tubular bones. The steep, high forehead, the ocular hypertelorism, the broad and flat root of the nose, the relative midfacial hypoplasia, and the square and prognathic mandible, frequently accompanied by bilateral facial weakness, are apparent by the age of 10, occasionally earlier, and create an unmistakable facial expression. The complications arise from narrowing of the foramina for the cranial nerves, the canals for the cranial blood vessels, and from increased intracranial pressure. Progressive losses of hearing and facial nerve function, which generally appear in childhood, and impairment of vision are common complications. Death may result from acute compression of the medulla oblongata.

The clinical and radiologic features permit early recognition of the disorder and differentiation from similar bony dysplasias associated with generalized sclerosis and hyperostosis.

Sclerosteosis is very similar to the autosomal recessive form of endosteal hyperostosis (van Buchem's disease). Since many of the afrikaners of South Africa had their origins in Holland, where van Buchem's patients were studied, it seems possible that there might be some fundamental link between the two disorders.

HISTORY

A familial disorder with generalized sclerosis and hyperostosis involving the calvaria, mandible, and clavicles, associated with syndactyly and dysphalangy, has been recognized for more than five decades. It was probably first described by Hirsch.¹ The condition was initially considered a manifestation of osteopetrosis. Kretzmar and Roberts² discussed such a case of Albers-Schönberg disease. Falkoner and Ryrie³ reported a similar example of two siblings and consanguineous parents. Additional cases with identical symptoms and signs were described by Higginbotham and Alexander,⁴ Kelley and Lawlah,⁵ and Pietruschka.⁶ Truswell⁷ was the first to include syndactyly in the syndrome, but considered the condition still to be a rare morphologic variant of osteopetrosis. It was Hansen⁸ who recognized it as a separate clinical entity, independent of osteopetrosis and who gave it the name of "sclerosteose," or in English, sclerosteosis. Case reports have also been published by Klintworth,⁹ Witkop,¹⁰ and Sugiura and Yasuhara.¹¹ Beighton, Cremin, Davidson, Durr, Epstein, and Hamersma¹²⁻¹⁸ provided the definitive description of the disease.

CLINICAL FEATURES

The characteristics of sclerosteosis were well summarized by Hansen⁸ in 1965. Beighton et al.¹³ reviewed the manifestations in 25 affected afrikaners in 1976. Pertinent data were collected from four deceased affected siblings. Extensive investigations of the relatives of these patients were undertaken for possible identification of clinically normal heterozygous carriers of the gene. An additional 17 patients were discussed by Beighton and Hamersma¹⁵ in 1979. The clinical manifestations and important information are summarized here briefly.

Skeletal Manifestations

In most patients the disease is evident at birth in the presence of a partial or, rarely, total syndactyly. The hands usually have bilateral but asymmetrical cutaneous, occasionally osseous, union. It involves the second and third fingers most often, the third and fourth fingers frequently, and the first and second fingers rarely (Fig. 1). In the majority of instances, the cutaneous webbing extends only part way to the first interphalanageal articulation. It may reach the proximal articulation, and occasionally the distal articulation. Characteristically, one or more digits are foreshortened and deformed, with radial deviation of the terminal phalanges, and different forms of onychodysplasia to the point of total aplasia. Nine of Beighton's 42 patients^{13,15} had no syndactyly. Three of the patients without syndactyly had affected siblings with marked syndactyly.¹⁵ The toes also may show cutaneous webbing.

Except for syndactyly, and occasionally facial paralysis, patients appear normal at birth. Mandibular prognathism and frontal prominence become noticeable by the age of 5 years. The skeletal deformities thereafter progress steadily. Overgrowth of the calvaria, a steeply elevated forehead, ocular hypertelorism, proptosis, a flattened nasal root, relative midfacial hypoplasia, and a square and broadened mandible, together with the bilateral facial paralysis, lead in adulthood to a severe facial deformity marked by a somewhat bizarre expressionless countenance. Children affected with the disease are tall for their age, and affected adults may have gigantism and excessive skeletal mass. Some adults reach a height of 210 cm.¹³⁻¹⁵

Neurologic Manifestations

Impairment of hearing is frequently a presenting symptom. It is often preceded by recur-

rent otitis media. It may manifest in early childhood. As a rule, it is bilateral. In the beginning it is an expression of impairment of sound conduction. Later, it may become associated with a loss of perception. A pure sensorineural hearing loss is an exception. All 16 adults and a third of the children among Beighton's 25 patients¹³ had a bilateral, predominantly conductive type of hearing loss. Associated sensorineural hypacusis was encountered only in the adults. The conductive loss was attributed to ossicular immobilization and jamming of the stapedial footplate, resulting from concentric narrowing of the middle ear cavity and distortion of the vestibular fenestra. The sensorineural loss was thought to have resulted from narrowing of the internal acoustic meatus and possibly from obliteration of the vestibular and cochlear fenestra. A reduction of vestibular responses to caloric and rotatory stimulation in these patients has been reported by Hamersma.¹⁹

Impairment of facial nerve function is another salient feature. It is observed almost as often as hearing loss. As a rule, it manifests initially as a recurrent unilateral paresis that eventually progresses to a permanent, in general partial, bilateral loss of facial nerve function. Unilateral facial paresis was present at birth in four of Beighton's 25 patients and developed during the first five years in an additional 12.¹³ All but one of the 16 adults in that series had permanent facial nerve impairment. In the majority the impairment was a partial paralysis, and in five of the 16 adult patients, it was unilateral. The progressive facial paresis is generally attributed to increasing constriction of the nerve within the fallopian canal.*

Unilateral and bilateral impairment of vision associated with optic nerve atrophy is infrequent. It generally represents a late complication that develops during the third decade of life. However, one instance of bilateral optic nerve atrophy in an adolescent has been reported.¹³

Headaches are common and often begin in adolescence.¹⁵ They probably reflect a rise in intracranial pressure.¹³⁻¹⁵ Intracranial hypertension has been documented by lumbar puncture and papilledema.⁹ Several patients have died suddenly from impaction of the medulla oblongata in the foramen magnum.¹³

Anosmia has been observed in several patients.⁹ One possible explanation is constriction

of the foramina for the transmission of the olfactory nerves in the cribriform plate.

Involvement of the trigeminal nerve may be another manifestation of the disease. Six of Beighton's 25 patients¹³ had hypesthesia in the distributions of V1 and V2, and one had unilateral trigeminal neuralgia involving V2 and V3. Corneal hypesthesia occurs less frequently. Constriction of the foramen rotundum has been documented by multidirectional tomography.¹⁸

So far there has been no report of lower cranial nerve involvement. Constriction of the cervical intervertebral foramina may result in impingement on the respective spinal nerves, leading to paresthesia, pain, and muscular weakness in the upper limb.

Radiologic Manifestations

Beighton et al.¹² originally found no radiographic abnormality other than syndactyly in their three youngest patients, age 9 months, 3½ years, and 5 years. However, some cranial thickening and increased density may be noticeable in infancy, and widespread changes are well advanced by the age of 5 years.¹⁶ Hyperostosis and sclerosis of the skull and tubular bones progress well into the third decade of life, when the disease tends to stabilize. The bones are enlarged in all dimensions (hyperostosis), which accounts for the progressive increases in body length and skeletal mass. Moreover, there is a moderate to marked increase in bony density (sclerosis). In addition, there is a disproportionate thickening of the cortices of many bones. Tubular bones so affected are massive, disproportionately long and wide. They have less diaphyseal constriction than do normal bones and may have some alteration of the external contours. Flat bones involved in the disease are abnormally thick. Narrowing of the medullary cavities may occur in some sites, but it is not a striking feature of the disease; neither is anemia a feature of sclerostosis. Hyperostosis and sclerosis are most prominent in the skull and tubular bones.

In the skull, the changes are greatest in the base of the neurocranium and in the body of the mandible. The base becomes increasingly sclerotic and the cranial nerve foramina and vascular channels become progressively narrowed. The internal auditory and facial canals are constricted and the optic foramina are deformed. The calvaria may be almost as severely affected. The head circumference can be increased, and the width of the vault may be enlarged as much as 3 cm.³ On regular radiograms the external and internal tables often cannot be recognized in adolescents and adults, suggesting obliteration of

* Fallopian is spelled fallopian. It has been misspelled in most anatomical textbooks and atlases but I have Fallopius' original textbook in my collection. In the European, particularly in the German, literature, the word fallopian is routinely spelled with two p's.

the diploë. We believe this is an illusion (Fig. 14). The overgrowth of the calvaria results in a diminution of cranial capacity. The hyperostosis leads to the formation of a steep and elevated forehead, ocular hypertelorism, and prominence of the supraorbital rims, and to constriction of the orbits. The sella turcica was enlarged and rounded in six of 12 adults.¹² The mastoid air cells are obliterated, but the paranasal sinuses remain intact. Enlargement of the body of the mandible, often severe, leads to prognathism and malocclusion. The mandibular rami are less involved. The facial bones are the least affected, which explains the clinically noticeable relative midfacial hypoplasia.

The long bones are massive, with cortical hyperostosis and some alteration of their external appearance. One of the most prominent features of the disease is the changes in the hands. The metacarpals and proximal phalanges are the most severely involved, they lack the usual modelling and usually are gently curved. Since they are less involved, the middle and distal phalanges appear relatively small. However, they may be more deformed. The distal phalanges may show a bizarre radial kinking, most frequent in the index finger. Often the entire finger has a radial deviation. As mentioned above, about 90 per cent of patients have some cutaneous syndactyly, and some have osseous syndactyly as well. The changes involving the metatarsals and the phalanges of the feet are similar, and bilateral partial cutaneous webbing of the second and third toes is not infrequent.^{12,13,16}

The clavicles and the ribs are widened and dense. The scapulae and the pelvis are sclerotic, somewhat thickened, but not expanded. The sclerosis in the pelvis tends to spare the iliac wings, which are somewhat narrow.¹²

The vertebrae tend to have normal proportions. Beighton et al.¹² found partial sparing of the vertebral bodies, with more sclerosis of the pedicles and laminae.

Endocrine Aspects

In addition to the gigantism and increased skeletal mass, lateral radiographs of the skull demonstrated an enlarged sella in six of 12 adults in Beighton's series.¹³ Pituitary function studies, including measurements of serum T₄ and T₃, resin uptake, growth hormone, and cortisol, have shown no abnormality in three patients, and growth hormone and cortisol levels during insulin tolerance tests were normal.¹⁸ A pituitary tumor has not been reported, and histologic examination of the hypophysis of one patient revealed no abnormality (Witkop CJ, per-

sonal communication). Alkaline phosphatase usually is elevated, but serum calcium, 24-hour urinary calcium, and serum phosphorus are normal.^{13,18}

Age Incidence and Sex Distribution

The skeletal changes become apparent in early childhood, steadily increasing in severity until progression ceases towards the end of the third decade of life. Of the 16 adults, eight were female, and of the nine children, five were female, eliminating a predilection for one sex.¹⁴

GENETICS

All of Beighton, Durr, and Hamersma's 25 affected individuals were of the afrikaner community and were descendants of settlers from Holland, who came to South Africa during the 17th century.¹³ Analysis of pedigrees of these families led to the conclusion that sclerosteosis in South Africa is inherited as an autosomal recessive trait with a minimum prevalence of 1 in 75,000.¹³ One in 140 afrikaners is a carrier of the gene. Witkop's analysis¹⁰ of the cases of Kelley and Lawlah⁵ also indicated autosomal recessive inheritance. Parental consanguinity has been reported.^{3,7,12,15} To date no reliable method of detecting heterozygotes has been found. No karyotypic abnormality has been demonstrated.¹⁸

Sclerosteosis is a rare disorder of bone remodelling, so far recognized only in some 40 individuals of the afrikaner community of South Africa,^{12,18} in three separate families in the United States,^{4,5} including that of our patient, and in one Japanese girl¹¹ and one Swiss patient.⁶ The observation of four instances of sudden death and the fact that only two of 16 adult patients were more than 35 years old¹³ indicate that sclerosteosis is potentially lethal. However, at least one affected individual has lived to at least 76 years old.¹⁵

Since impairments of facial nerve function and hearing are two of the salient features of this bony dysplasia, and either may become clinically manifest at birth or in early childhood, the responsibility for recognizing the disease often falls upon the otolaryngologist.

REPORT OF A CASE

The patient, a black girl, was 10 9/12 years old when her parents first sought medical treatment for her condition at our hospital. Both parents are tall but otherwise appear normal. Skull radiographs showed their calvariae to be somewhat thicker than average but within normal

limits. Three of the patient's distant relatives are reported to have sclerosteosis (Witkop CJ, personal communication).

The patient was born with cutaneous syndactyly, involving the left second, third, and fourth fingers, that required surgical correction (Fig. 1). The onset of a subsequent gradual loss of hearing was first suspected at the age of 3 years, when the patient had five aspiration myringotomies for recurrent serous otitis media. An audiogram at the age of 6 years disclosed a bilateral flat conductive hearing impairment with an average pure-tone loss of 35 dB in the speech frequency range. Radiograms of the temporal bone showed marked sclerosis of the mastoid processes and petrous pyramids. An exploratory tympanotomy at that time allegedly revealed a partial fixation of the malleus and stapedial footplate. The patient was subsequently fitted with a hearing aid.

Present Illness. Six months before her initial admission to the Johns Hopkins Hospital at the age of 10 9/12 years, the patient complained of bifrontal headaches and progressive difficulties with vision. On the initial admission, the funduscopic examination disclosed bilateral optic nerve atrophy, and a lumbar puncture revealed a pressure of 480 mm H₂O, with normal cell count, glucose, and protein. The increased intracranial pressure and decreased visual acuity were initially amenable to medical management, but at the age of 11 8/12 years the pressure increase became more severe, and bilateral facial paresis suddenly developed. The patient had significant difficulties chewing, but denied swallowing problems, vertigo, facial pain, or symptoms suggesting an endocrinopathy.

On physical examination at the time of the patient's second admission, a month later (age 11 9/12 years), her large size made her appear to be some years older than her stated age. Her weight was 75 kg and her height 177 cm. Vital signs were normal. The patient's face was ovoid, with a steep and high forehead. She had ocular hypertelorism, slight exophthalmos, broadening of the nasal base, relative hypoplasia of the midfacial area, and a square, prognathic mandible. The bilateral facial nerve palsy added to the characteristic expressionless facial appearance.

The neurologic examination revealed the following: Cranial nerve I, slight hyposmia on forced stimulus testing. Cranial nerve II, visual acuity 5/200 (OD/OS) and bilateral optic nerve atrophy. Cranial nerves III, IV, and VI, full extraocular movements, but slightly sluggish pupillary responses to light. Cranial nerve V, normal facial sensation in response to light touch, pinprick and cold; normal corneal sensation. Cranial nerve VII, bilateral facial paresis

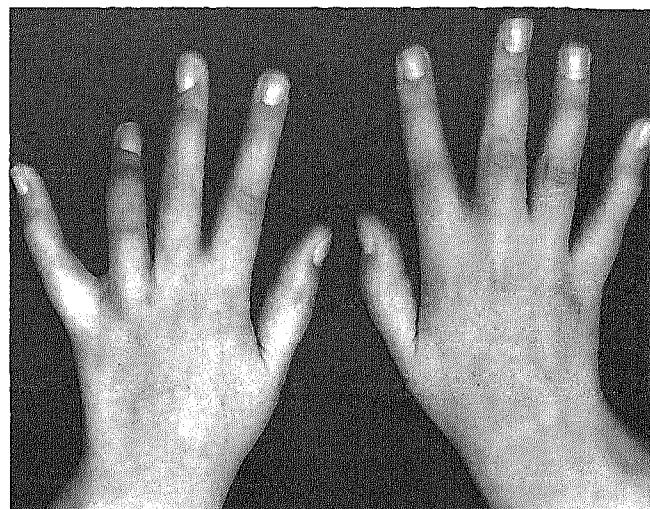


Figure 1. Slight cutaneous syndactyly involving the second, third, and fourth digits, moderate camptodactyly of the (L) fourth digit and slight camptodactyly of other digits, slight onychodysplasia of the (L) third digit, and a tendency toward axial deviation of some digits.

with some residual function in the orbicularis oculi muscles. Facial electromyography revealed bilateral partial denervation of the frontalis, orbicularis oculi, and orbicularis oris branches. The frontalis branch was most affected. Taste was normal to gross testing of all four modalities. The Schirmer test was 12 mm on the right (R), and 15 mm on the left (L). Cranial nerves IX and X, slight paresis of palatal elevation. Cranial nerves XI and XII, normal. The patient's speech was dysarthric. On tandem gait, the patient demonstrated mild ataxia. Muscular strength was normal.

Radiologic examination of the skull showed significant increases in the diameter and density of the calvaria, with apparent obliteration of the diploic space. The sclerosis and thickening were even more marked in the base and temporal bones (Figs. 2, 11, and 12). The body of the mandible showed distinct hyperplasia. The foramina of exit for cranial nerves II, V, VII, VIII, IX, X, XI, and XII had obvious concentric narrowing bilaterally.

The clavicles and ribs were wide and sclerotic (Fig. 3). Sclerosis was more striking in the spine than in most of the reported cases, and only in the thoracic and midcervical regions was there some sparing of the vertebral bodies (Figs. 4 and 5). The pelvis showed a marked increase in bony density, but no thickening was apparent (Fig. 6).

The long bones were enlarged and the cortices were thick and sclerotic. The epiphyseal ossification centers were much less affected than other portions of the bones, so that many epiphyses appeared to be relatively flat (Figs. 7 and 8). In the hand, cutaneous syndactyly was still rec-

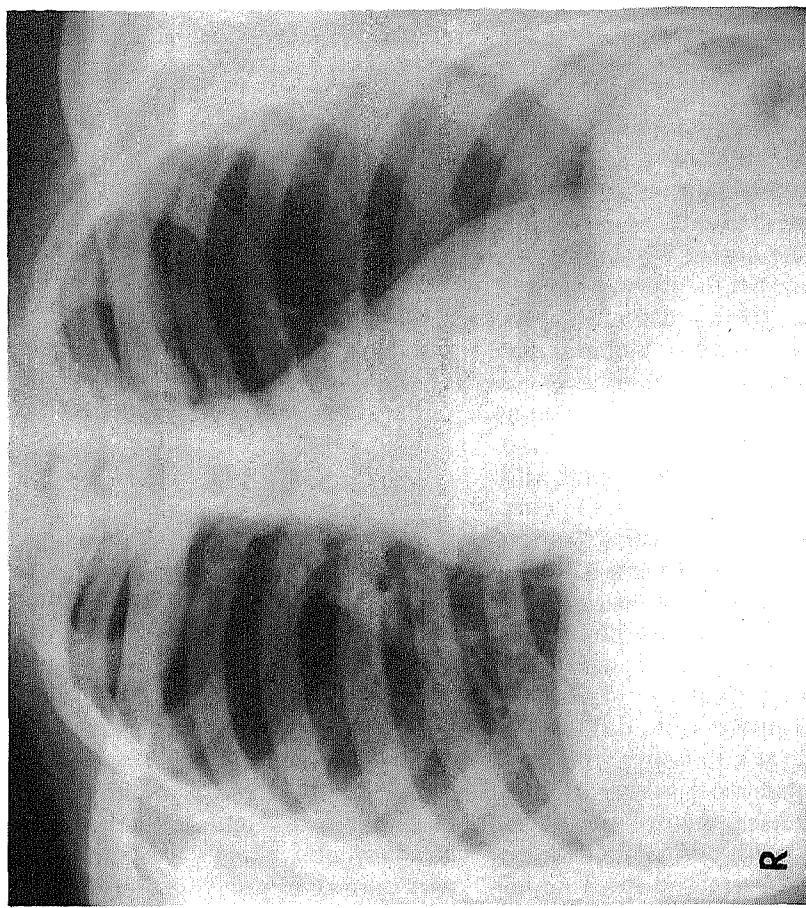
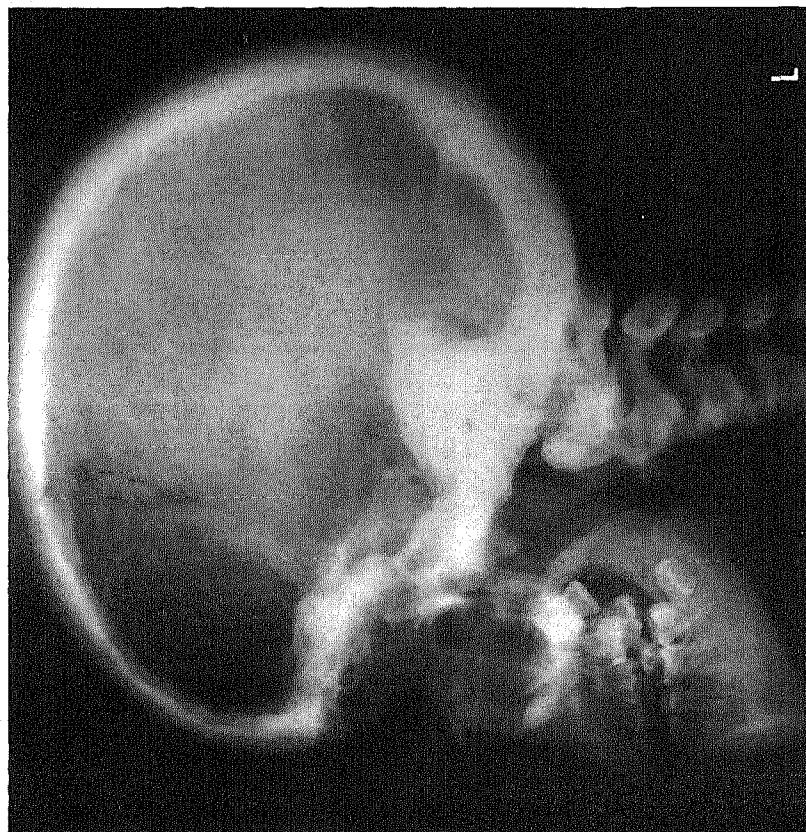


Figure 2 (left). Skull, lateral radiograph. The striking increase in thickness and density of most bones is greatest in the base of the neurocranium, especially in the petrous portions of the temporal bones. The calvaria shows slight involvement; the hyperostosis and sclerosis of the inner and outer tables appear to have obliterated the diploë. The sella turcica is enlarged and the inner cortex is lost along the floor and the dorsum. No mastoid air cells are visible, yet the paranasal sinuses are well pneumatized. The body of the mandible is greatly thickened.

Figure 3 (right). Chest, anteroposterior radiograph. The increased diameter and the density of the clavicles and ribs are clearly visible.



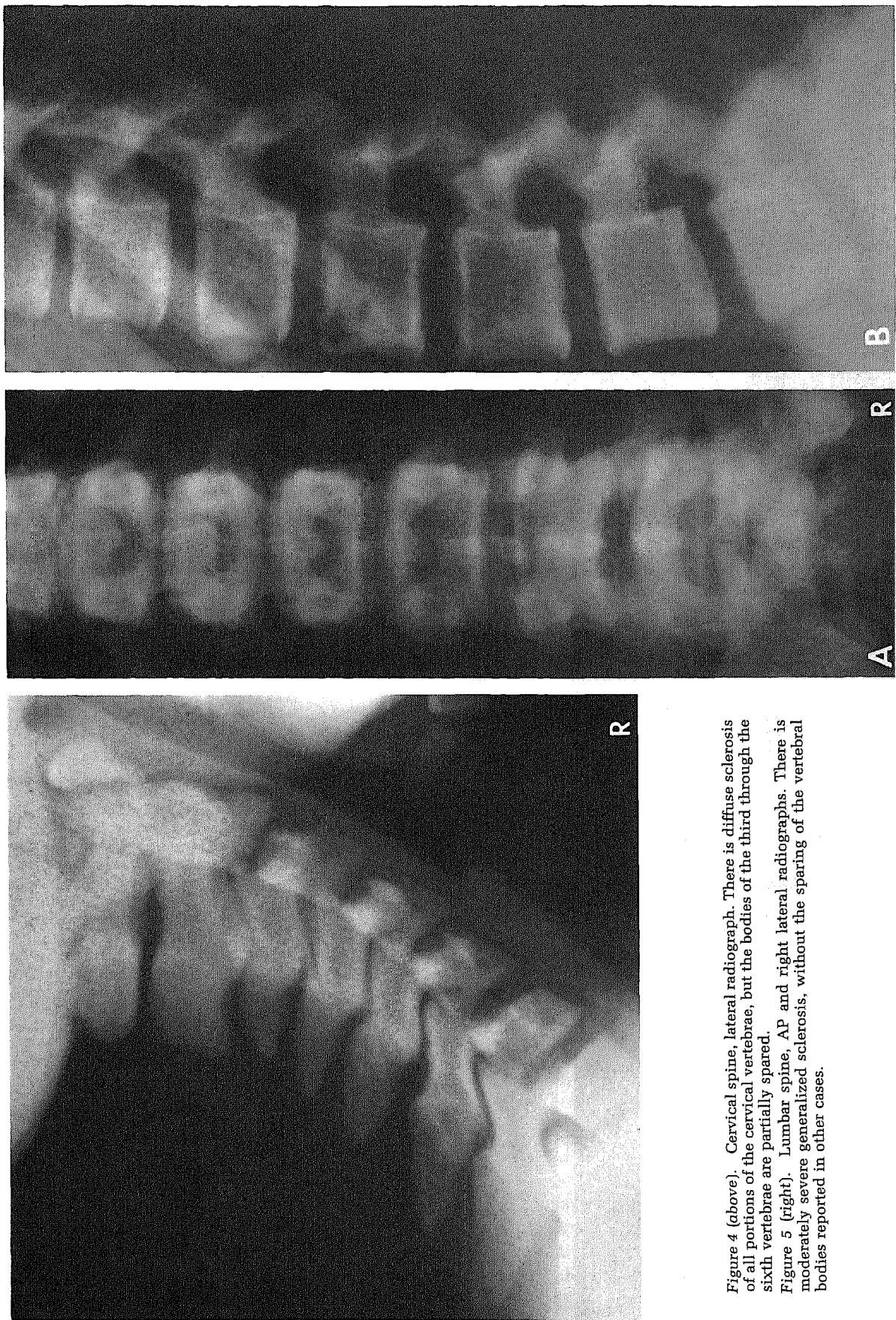


Figure 4 (above). Cervical spine, lateral radiograph. There is diffuse sclerosis of all portions of the cervical vertebrae, but the bodies of the third through the sixth vertebrae are partially spared.
Figure 5 (right). Lumbar spine, AP and right lateral radiographs. There is moderately severe generalized sclerosis, without the sparing of the vertebral bodies reported in other cases.



Figure 6. Pelvis. The generalized sclerosis is quite striking, yet the bones do not appear disproportionately wide. Notice the normal proportion of the ischial and pubic rami. The sclerosis is greatest along the medial portions of the iliac bones and about the acetabular cavities.

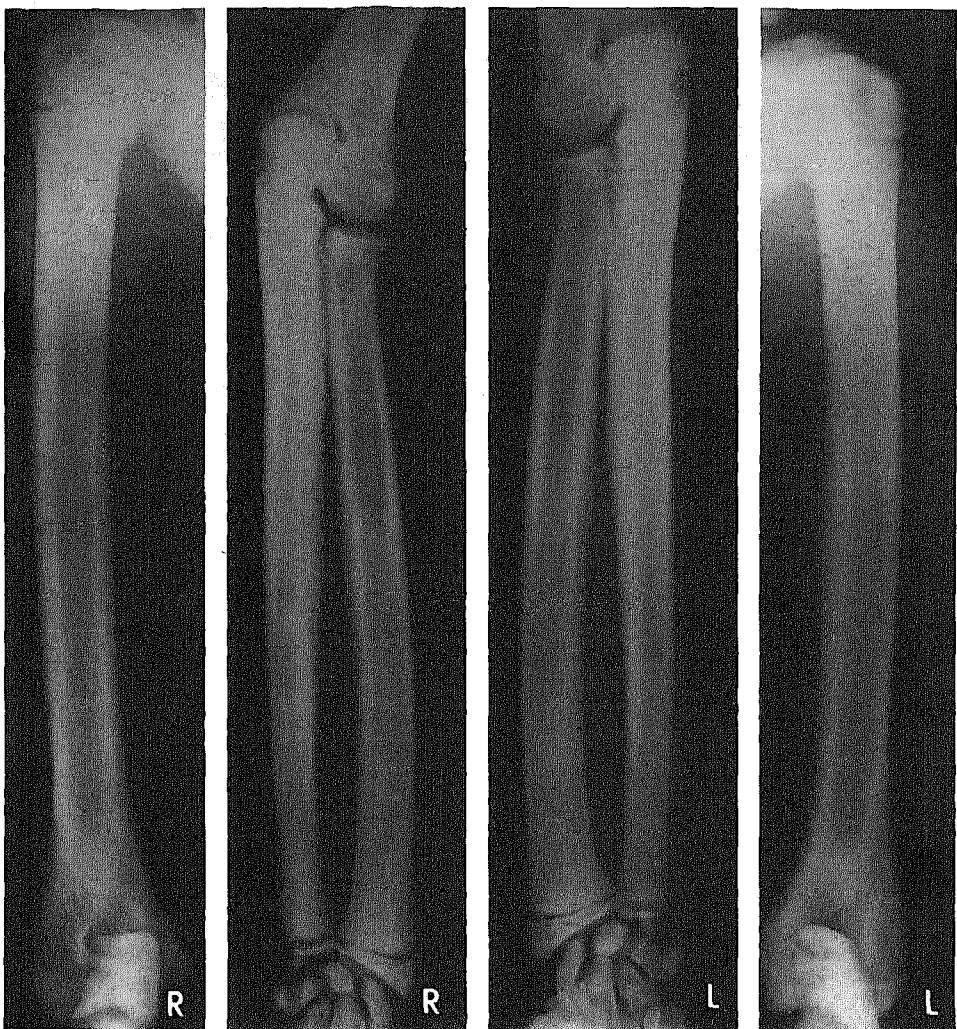


Figure 7. Arms and forearms, AP radiographs. The bones are sclerotic and relatively widened, particularly in the diaphyses, which accounts for the decreased modelling. While the cortices are thickened, the medullary cavities are in most areas only slightly expanded or of normal width. The epiphyseal ossification centers are much less affected than the rest of the bones and appear moderately flattened in the elbow and wrist.

Figure 8. Hands, PA radiograph. The carpal bones and the epiphyseal ossification centers of the distal parts of the radius and ulna are of about normal size. The other bones are strikingly elongated and disproportionately wide. The changes are most marked in the second and third rays. Except in the thumbs, all of the metacarpals and phalanges show a slight radial bowing. The deformity of the (L) ring finger is the result of contracture of the proximal interphalangeal joint and moderate bowing of the middle phalanx.

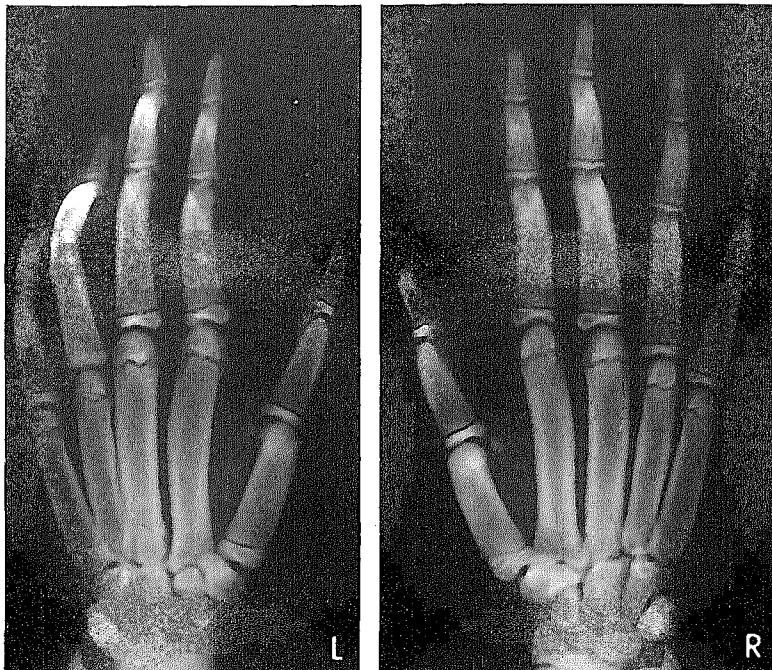


Figure 9. Feet, dorsal-plantar radiograph. The changes in the metatarsals and phalanges are similar to those in the hands except that the phalanges and medial metacarpals are less bowed. The middle and distal phalanges are relatively short, and there is hallux valgus.



ognizable, even though the major portion had been surgically corrected on the left side. The metacarpal and phalangeal bones showed striking changes (Fig. 8). The abnormalities in the feet were as severe as those in the hands (Fig. 9).

Computed tomography of the skull and brain confirmed the calvarial thickening and showed a moderate, bilateral parietal lobe atrophy but no ventricular abnormality.

The **otologic examination** revealed slight con-

centric narrowing of the osseous external auditory canals. Except for a slight retraction of the (R) tympanic membrane, both tympanic membranes were normal. There was no fluid in either middle ear, and promontorial hyperemia was not found. The malleus was firmly fixed bilaterally, and the short process and handle of the malleus were slightly increased in size.

Cochlear function studies showed a bilateral, asymmetric conductive hearing loss, gradually

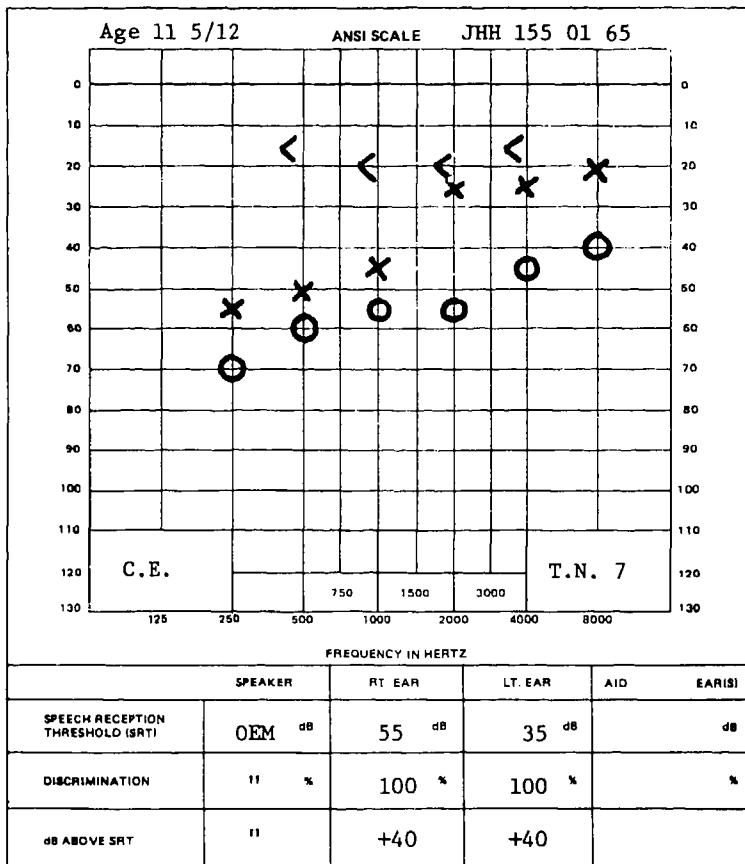


Figure 10. Audiogram obtained from the patient.

decreasing towards the higher frequencies, more marked in the (R) ear (Fig. 10). The speech reception threshold was 55 dB in the (R) ear and 35 dB in the (L) ear; discrimination was 100 per cent bilaterally. The (R) tympanogram revealed a type C pattern and the (L) a type B pattern. The acoustic reflexes were absent bilaterally from 500 to 4,000 Hz at 110 dB sound pressure level.

Vestibular function studies disclosed a vertical nystagmus, which increased on (R) and (L) lateral gaze. Caloric responses to conventional hot and cold stimulation were markedly diminished, with an average slow-phase eye speed of 4.75°/sec. Responses to ice-water stimulation were at 11°/sec. Eye tracking and refixation movements were severely disturbed, but optokinetic nystagmus appeared normal in both horizontal and vertical directions.

Radiograms of the temporal bones in the conventional projections, and anteroposterior and lateral multidirectional tomography of the petrous pyramids and mastoid bones, showed increases in size and striking diffuse sclerosis of all portions of the temporal bones. On each side, the middle ear cavity, but above all the attic, and the aditus and antrum were markedly reduced in size (Fig. 11). There was no single visible air cell. The contours and lumina of the three semicircular canals, vestibule, and cochlea appeared normal, but the internal acoustic meatus had an almost thread-like lumen as a result of concentric narrowing. The remodelling process had caused a slight lateral and anterior displacement of the fallopian canal in reference to the lateral semicircular canal, a minimal but recognizable distortion of the horizontal and vertical segments, and a slight constriction of its lumen (Fig. 12). The malleus and incus showed an overall increase in size (Figs. 11 and 12). This increase in ossicular size and the concentric narrowing of the epitympanic recess had led to impingement of the malleus and incus in several directions (Figs. 11 and 12). The lower portion of the sigmoid groove and the jugular foramen were almost totally obliterated (Fig. 12). The carotid canal, however, had remained unaltered.

The venous phase of the carotid angiogram (Figs. 13 and 14) demonstrated marked narrowing of the (L) transverse sinus and sigmoid sinus (Fig. 13), almost complete obstruction of the lower segment of the (R) sigmoid sinus, and near-total occlusion of the (R) jugular foramen (Fig. 14). The (R) and (L) jugular venograms (Fig. 15) showed a minimal remaining lumen of the (L) sigmoid sinus and jugular bulb and extreme constriction of the (R) jugular bulb.

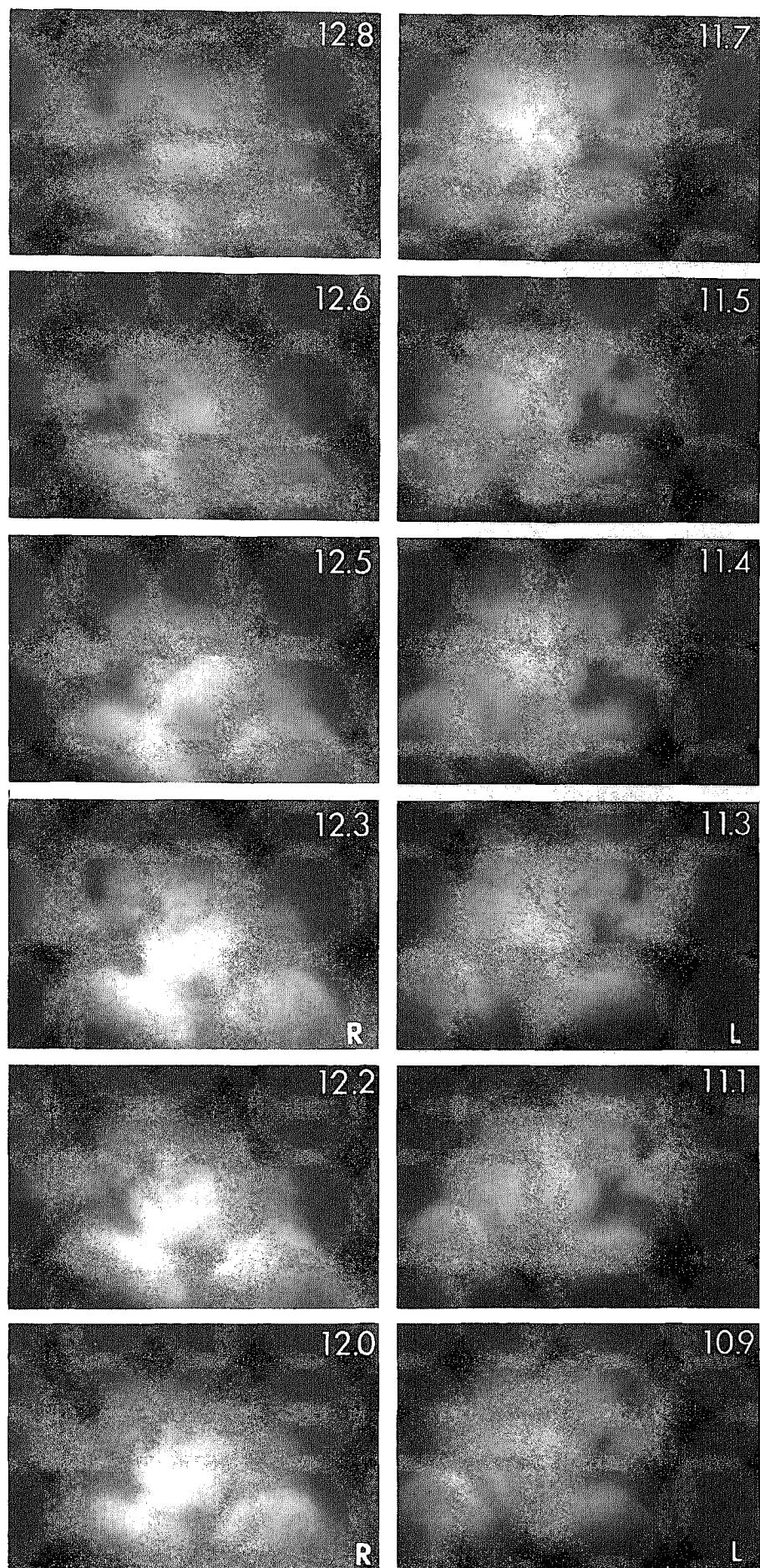


Figure 11. Anteroposterior polytomograms of (R) and (L) temporal bone at corresponding levels. All portions of the temporal bone are massive in size and sclerotic. The hyperostosis resulted in complete obliteration of air cells and marrow spaces, and in severe constriction of the internal auditory canal, middle ear cleft, and external auditory meatus [Sects. 12.5, and 12.3 (R), and 11.5 and 11.4 (L)]. These structural changes eventually led to compression of cranial nerves, immobilization of middle ear ossicles, and distortion of the cochlear and vestibular fenestra.

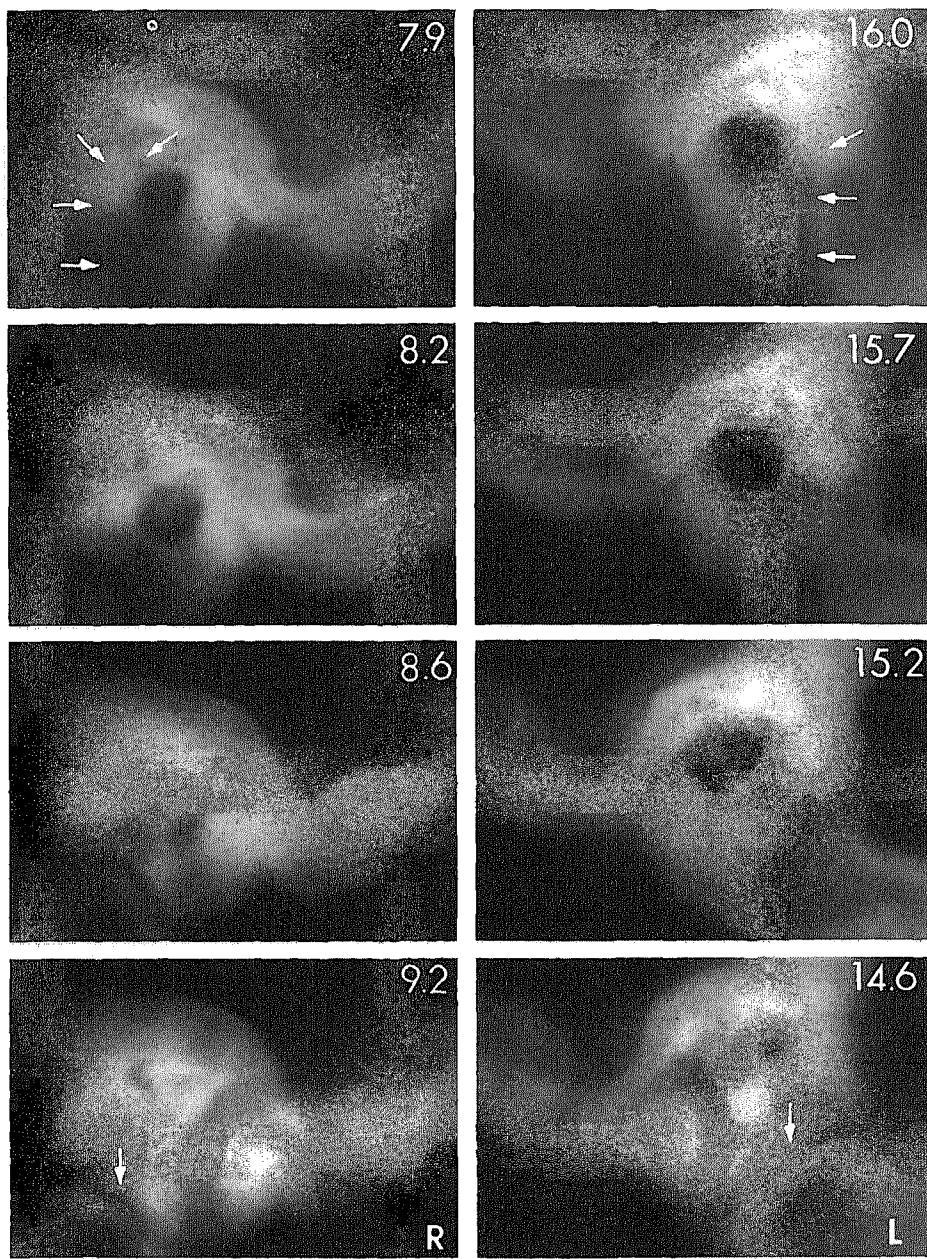


Figure 12. Lateral polytomograms of (R) and (L) temporal bone. The narrowing of the epitympanic recess, the enlargement of the outer middle ear ossicles, and the distortion and constriction of the tympanic and mastoid segments of the fallopian canal (arrows) are recognizable in sections 7.9 (R) and 16.0 (L). The carotid canal appears normal, but the inferior portion of the sigmoid groove and the jugular foramen (arrows) are almost totally obliterated [Sects. 9.2 (R), 14.6 (L)].

The laboratory studies revealed a normal hemogram and electrolytes. Serum calcium, inorganic phosphorus, alkaline phosphatase, parathyroid hormone, and calcitonin were normal. Growth hormone, thyroid function studies, and prolactin were within normal limits.

Operations (Drs. M. Holliday, G. Nager, D. Kennedy, and D. Long). Because of the increased intracranial pressure associated with jugular outflow obstruction and bilateral facial paresis, the (R) sigmoid sinus, (R) jugular bulb, and (R) facial nerve were decompressed. Because of the ivory-hard texture of the bone, complete absence of any pneumatic cells, and extreme narrowing of the antrum, aditus, and attic, the operation was tedious and time-consuming. The lumina of the lower portion of the sigmoid sinus and of the

jugular bulb were extremely constricted. However, by the end of the decompression these venous channels had expanded to their original normal sizes. The antrum, aditus, and attic were markedly constricted by concentric new bone formation. The hyperostotic bone was impinging upon the malleus and incus. It caused distortion, displacement, and immobilization of the ossicular chain, fracture of the stapedial arch at the base, and anterior and inferior dislocation of the stapedial suprastructure. Narrowing and distortion of the oval window margin had created a jamming and fixation of the stapedial footplate. All middle ear ossicles were of normal size and configuration. The middle ear cavity was greatly reduced in size.

The posterior cranial fossa and the foramen

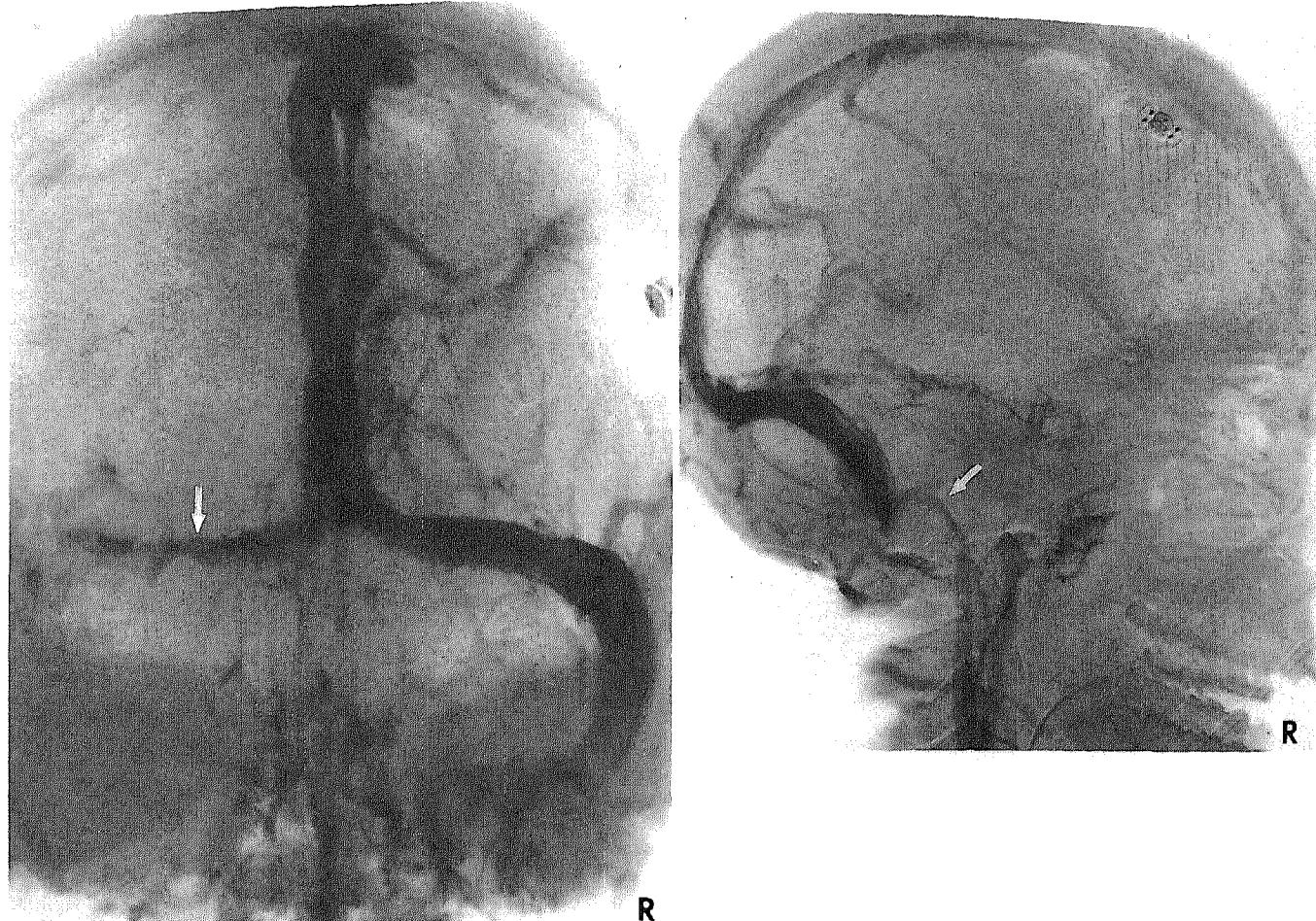


Figure 13 (left). Venous phase of carotid angiogram anteroposterior subtraction study. The severe constriction of the (L) transverse sinus (arrow) with total occlusion at its junction with the (L) sigmoid sinus, and the near total obliteration of the caudad end of the (R) sigmoid sinus are well shown.

Figure 14 (right). Venous phase of carotid angiogram, (R) lateral subtraction study. The near total occlusion of the (R) jugular bulb and the adjacent sigmoid sinus are clearly evident (arrow). The diploë, which appeared to be obliterated on the standard radiographs (Fig. 2) is clearly shown to be preserved and relatively thick on this and the other subtraction studies.

magnum were decompressed in a second stage by the neurosurgeon (Dr. D. Long). The patient made an uneventful recovery, her vision and mental functions improved remarkably, and intracranial pressure returned to normal and has remained so for the past 24 months. Biopsy of the (R) mastoid process revealed conspicuous widening of the bony trabeculae of the spongiosa, leading to progressive obliteration of the marrow spaces and sclerosis (Fig. 16).

DIFFERENTIAL DIAGNOSIS

Sclerosteosis must be differentiated from other diseases that increase bony density. These include osteopetrosis with late manifestation, pyknodysostosis, metaphyseal dysplasia (Pyle's disease), dysosteoclerosis, craniometaphyseal dysplasia, diaphyseal dysplasia (Engelmann's disease), congenital hyperphosphatasia, and

possibly hyperostosis corticalis generalisata (van Buchem's disease). Brief descriptions and excellent illustrations of the pertinent findings in all of these disorders are available in the Atlas by Spranger et al. (1974).²⁰

Of these disorders, van Buchem's disease is the one that most closely mimics sclerosteosis. The principal differences are the gigantism, syndactyly, and curvature of the fingers present in most cases of sclerosteosis. Most authors state that the bony abnormalities tend to be more severe in sclerosteosis, but Cremin¹⁷ concluded that they are identical. In view of the similarity of the two disorders, it is interesting that most cases of van Buchem's disease have come from Holland, while most cases of sclerosteosis have been found in Afrikaners, who had their origin in Holland.¹³ Gorlin²¹ recently reached the conclusion that they represent the same disorder.

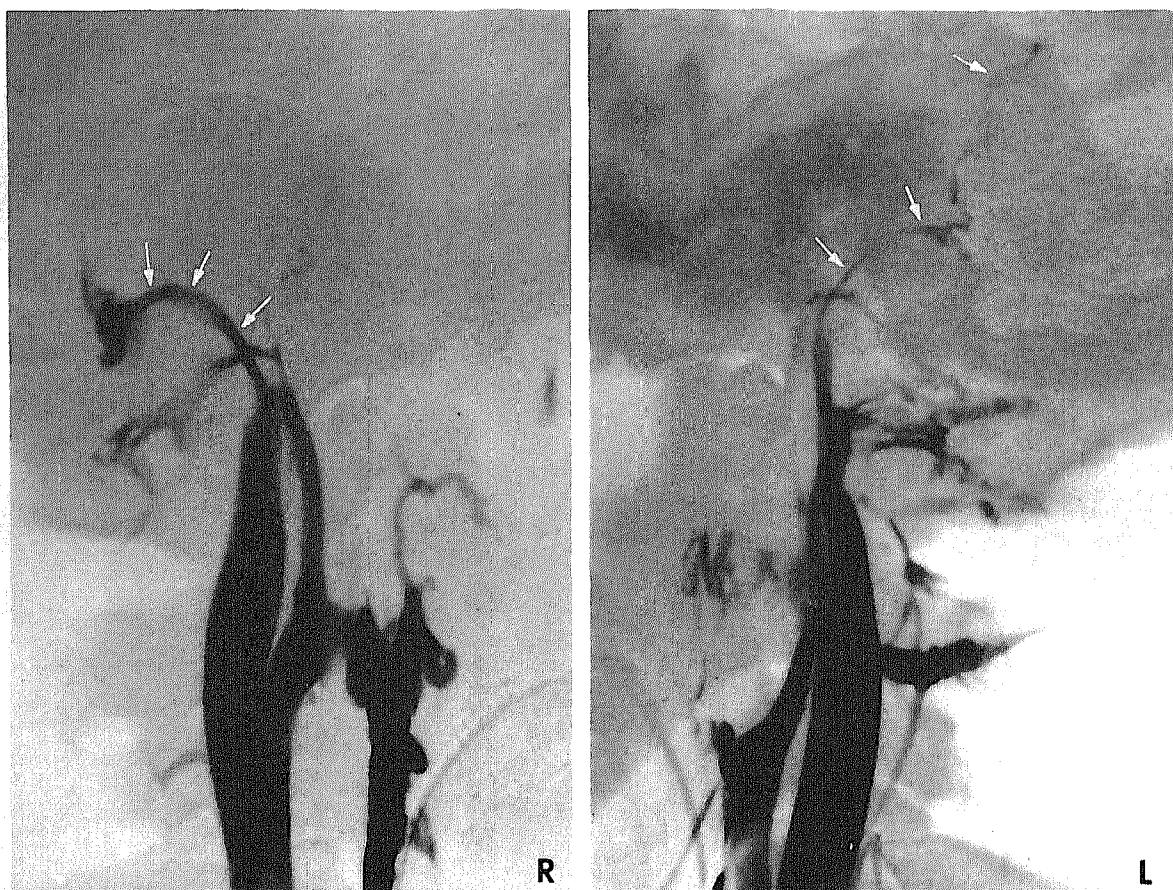


Figure 15. The (R) and (L) retrograde jugular venograms confirm the thread-like lumen of the (L) jugular bulb and sigmoid sinus (arrows) and the extreme narrowing of the (R) jugular bulb (arrows).

DISCUSSION

Some of the disorders of skeletal development and remodelling are of particular interest to the otolaryngologist because of their associations with hearing loss, vestibular dysfunction, and facial weakness. Osteopetrosis, fibrous dysplasia, osteitis deformans (Paget's disease), Pyle's disease, and sclerosteosis may alter the architecture of the external auditory canal and tympanic sulcus, involve and enlarge the middle ear ossicles, produce constriction of the middle ear cleft, and lead to narrowing and distortion of the vestibular and cochlear fenestra. Impingement upon the ossicular chain in the epitympanic recess, jamming, and occasional synostosis of the stapedial footplate will result in ossicular fixation and cause conductive hearing loss. Remodeling of the cochlear and vestibular capsule may give rise to axial and radial distortion of the cochlear modiolus, to fracture and dislocation of the osseous spiral lamina, and to distortion of the cochlear turns.²²⁻²⁴ Apposition

of new bone in the tractus spiralis foraminosus, in the vestibular areas, in the fallopian canal, and in the internal acoustic meatus may lead to constriction of the canals for the respective cranial nerves and their branches. Active bone remodelling involving the endosteal capsule layer may produce secondary morphologic changes of the membranous cochlea and vestibular labyrinth and interfere with the physiologic integrity, metabolism, and oxygen supply of the inner ear and labyrinth. Thus, a variety of disease-related mechanisms may be responsible for the dysfunction of sound perception and equilibrium.

On the other hand, impairment of facial nerve function, as observed in osteopetrosis and sclerosteosis, appears to be caused by a less complicated mechanism, namely direct neural compression interference with the blood supply to the nerve.¹⁹

Since sclerosteosis does manifest at birth or in early childhood with facial weakness and impairment of hearing, in addition to syndactyly and other skeletal features, it becomes the re-

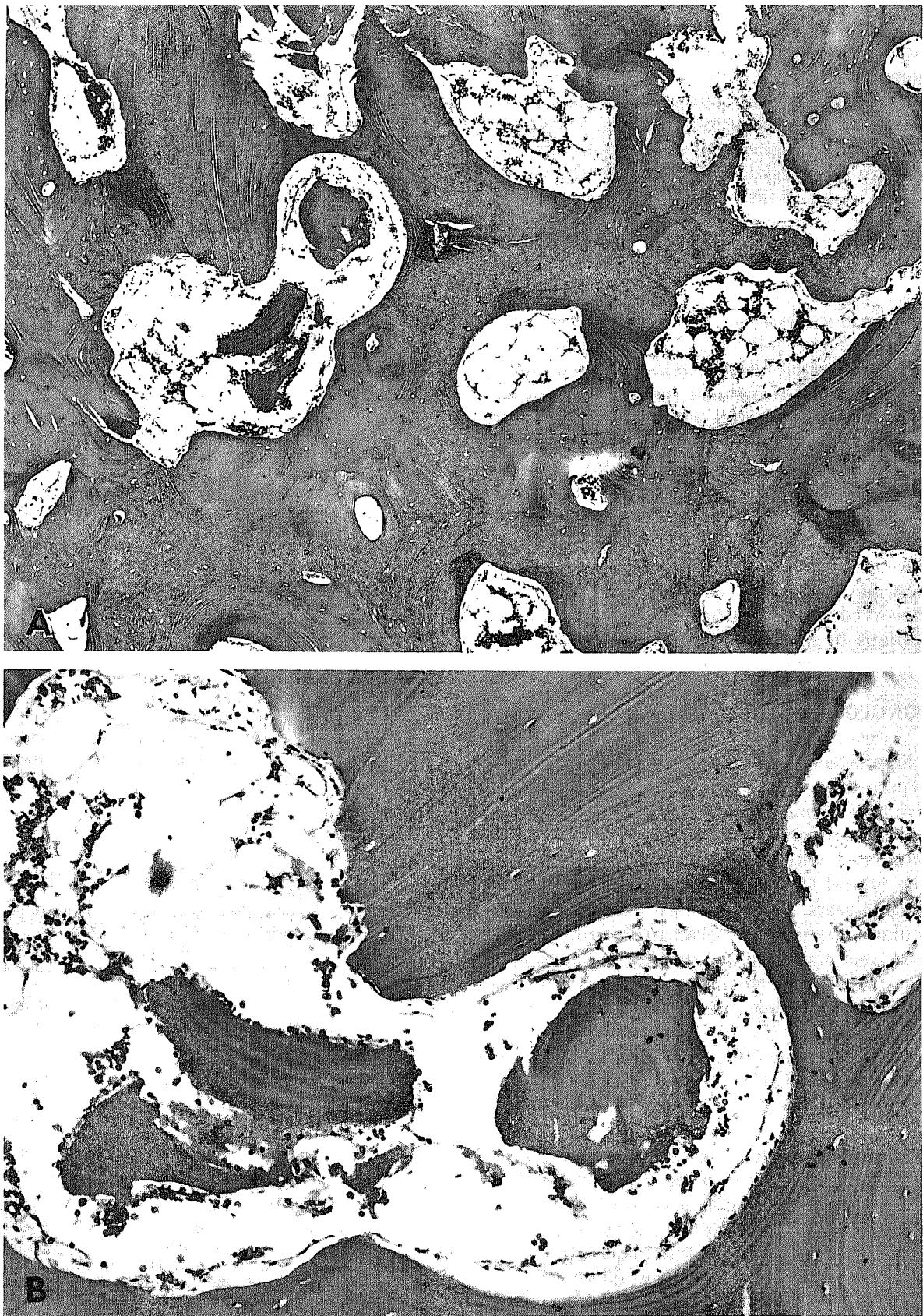


Figure 16. A (above), biopsy from (R) mastoid process; hematoxylin and eosin $\times 65$. Progressive obliteration of the marrow spaces caused by widening of the bony trabeculae and minimal bone remodelling have led to advanced sclerosis of the spongiosa in the tip of the mastoid. B (below), same section, $\times 175$. The osseous lamellae are arranged in parallel and concentric patterns. [Photomicrographs reduced to 95 per cent of size.]

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sponsibility of the otolaryngologist to recognize the condition and to assume his part of the management in a joint effort with the pediatrician, neurologist, and neurosurgeon. The complexity of the disease makes the treatment very challenging. The surgical treatment, when indicated, requires special skill. It has the potential to preserve vision and fifth, seventh, and eighth nerve function; to improve cerebral venous drainage; to reduce intracranial hypertension; and to alleviate compression of cerebellum and brainstem. Successful control of intracranial hypertension and alleviation of pressure upon brainstem and cerebellum by decompressing the posterior cranial fossa have been carried out by Devilliers and Hamersma (personal communication). Decompression of the entire fallopian canal must be given serious consideration in instances of progressive facial nerve involvement. Similarly, decompression of the optic canal, sigmoid sinus, and jugular bulb may be indicated, in combination with anterior and posterior craniotomy. If indeed the patient's life, vision, hearing, and facial nerve function can be preserved for a significant length of time or perhaps in some instances permanently, substantial progress will have been achieved.

CONCLUSION

Sclerosteosis is a rare form of craniotubular hyperostosis, inherited as an autosomal recessive trait, characterized by progressive sclerosis and overgrowth of the skeleton. It is frequently associated with excessive height and weight. The typical facial deformity is apparent by the age of 5 years. It includes a steep, high forehead, ocular hypertelorism, a broad flat root of the nose, and a prognathic, broadened, and square mandible. Syndactyly of the second and third or other fingers, either cutaneous or osseous, is noticeable at birth and helps to differentiate sclerosteosis from other dysplasias in the group. The terminal phalanges have a radial deviation and dystrophy of the fingernails. Constriction of the neural foramina results in cranial nerve involvement. Facial weakness and progressive losses of hearing and vision are the most frequent manifestations of cranial nerve involvement. Facial nerve paralysis may be present at birth or develop soon afterwards. It is frequently unilateral for many years but eventually becomes bilateral. Impairment of hearing usually appears in early infancy. In some instances, anosmia, facial hypesthesia, optic nerve atrophy, conver-

gent strabismus, nystagmus, and exophthalmos are present.

The radiologic changes involving the temporal bone include a marked increase in overall dimensions, extreme sclerosis, and narrowing and constriction of the external meatus, middle ear cleft, internal acoustic meatus, and fallopian canal. There may also be severe narrowing of the neural compartments of cranial nerves IX to XI, and subtotal or complete obliteration of the sigmoid sinus and jugular foramen.

Interference with the cerebral blood flow, resorption of cerebrospinal fluid, reduction of the intracranial volume, and thickening and distortion of the cranial base and foramen magnum may contribute to the gradual increase of intracranial pressure. This may have led in several instances to sudden death from impaction of the brainstem in the foramen magnum.

Early decompression of the neural and vascular channels and extensive decompression of the posterior fossa may help in the preservation of facial and cochlear nerve function and even of life.

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