

Sclerosteosis: neurosurgical experience with 14 cases

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✓ Fourteen cases of sclerosteosis seen during a 14-year period have been reviewed. Sclerosteosis is a craniotubular bone modeling disorder and presents with cranial nerve palsies and raised intracranial pressure; sudden death may occur without neurosurgical intervention. Experience gained from the management of these patients suggests that bifrontal decompressive craniotomy followed by a posterior fossa decompression should be performed in all cases, preferably in the second decade of life. Cerebrospinal fluid diversion procedures to reduce elevated intracranial pressure are unsatisfactory. Patients should be followed closely since the regrowth of bone may necessitate repeated decompressive procedures. Technical problems related to the surgical management are discussed.

KEY WORDS • cranial decompression • bone disorder • intracranial pressure • sclerosteosis

SCLEROSTEOSIS is a disorder of craniotubular bone modeling. This group of diseases is characterized by cranial nerve palsies and raised intracranial pressure (ICP) resulting from bone overgrowth of the skull.¹³ Sclerosteosis is an autosomal recessive condition with a minimum prevalence of one in 75,000 in the Afrikaner community of South Africa.^{2,4} At least one in 140 Afrikaners is a carrier of the gene; there are approximately 10,000 clinically normal heterozygotes in South Africa.⁸ The condition usually presents in infancy with attacks of acute facial nerve palsy, from which recovery is only partial. Conductive hearing loss develops in childhood and is followed by a perceptible component in the third decade of life. The ICP rises after puberty, and sudden death from impaction of the medulla oblongata in the foramen magnum is a serious risk.⁶

Sclerosteosis differs in many aspects from the more common group of diseases known as osteopetrosis or Albers-Schönberg disease.¹² Unlike osteopetrosis, which is attributed to abnormal osteoclastic function, sclerosteosis appears to be primarily a disorder of osteoblastic hyperactivity.¹³ The term "osteopetrosis" should be reserved for the condition in which generalized skeletal sclerosis predominates in the absence of bone overgrowth.⁷

The bone changes in sclerosteosis are most marked in the calvaria and mandible.⁴ The bone is very hard, thick (3 to 4 cm), and avascular. Microscopically, the cortices and trabeculae are thicker than normal.⁹ The spine is relatively spared. There is no undue liability to

osteomyelitis or dyshemopoiesis, as occurs in some forms of osteopetrosis.^{5,10} The most common clinical manifestations include facial nerve palsy and complete or partial deafness at an early age, visual impairment, and syndactyly of the third and fourth fingers; patients usually exhibit a normal intellect. The facial changes and a tall stature give sclerosteosis a superficial resemblance to acromegaly.⁶ The long-term sequelae of the disease are not well documented, as many patients died in the past from medullary compression in the second and third decades of life. Although survival into old age without neurosurgical intervention is unusual, a few cases have been reported.¹

Very few reports regarding the neurosurgical management of sclerosteosis have been published to date.^{11,13} The purpose of this paper is to describe our experience with the management of 14 patients seen over a 14-year period and to provide additional information regarding potential pitfalls in the surgical management of these patients.

Clinical Material and Methods

Patient Population

The 14 patients included in this study were seen at the H. F. Verwoerd Hospital in Pretoria, South Africa, between 1977 and 1991; only one patient (Case 1) was lost to follow-up review. Two of these patients were related. No history of involvement of ancestors could be obtained in any patient, and it is assumed that all were parented by asymptomatic heterozygous carriers.

Surgical management of sclerosteosis

TABLE 1
Summary of 14 cases of sclerosteosis*

| Case No. | Sex | Age (yrs) | | Surgical Procedure (mo/yr) | Complications | Clinical Outcome |
|----------|-----|-----------|----------------|---|---|---|
| | | At 1st Op | At Last Review | | | |
| 1 | F | 18 | | ant fossa dec (6/77) post fossa dec (8/77) L5-S1 laminectomy (4/82) L1-15 laminectomy (2/86) | | lost to review |
| 2 | M | 14 | 27 | post fossa dec (2/78) ant fossa dec (11/78) LP shunt (4/84) bifrontotemp dec (4/91) | subcutaneous CSF; postural headaches | LP shunt removed; asymptomatic after last op |
| 3 | F | 15 | | ant fossa dec (6/81) post fossa dec (6/82) LP shunt (6/82) post fossa dec (8/91) | pseudomeningocele; postural headaches | shunt removed (8/82); sudden death (8/91) at age 25 yrs, 1 day postop |
| 4 | M | 18 | 27 | post fossa dec (7/82) ant fossa dec (11/86) LP shunt (12/86) | postural headaches | shunt removed; good |
| 5 | M | 9 | 18 | post fossa dec (7/82) ant fossa dec (11/86) LP shunt (12/86) bifrontotemp dec (12/91) | subcutaneous CSF; postural headaches | good |
| 6 | F | 16 | | post fossa dec (7/82) | cardiac arrest 1 day postop | died |
| 7 | F | 19 | 27 | ant fossa dec (4/83) post fossa dec (7/83) LP shunt (1/84) | subcutaneous CSF; postural headaches; shunt removed | good |
| 8 | M | 22 | 29 | post fossa dec (3/84) ant fossa dec (10/84) bifrontotemp dec (11/91) post fossa dec (12/91) | | good |
| 9 | F | 21 | 27 | ant fossa dec (2/85) post fossa dec (7/85) bifrontotemp dec (8/91) post fossa dec (2/91) | subcutaneous CSF | emotional disturbances |
| 10 | M | 19 | 25 | post fossa dec (6/85) LP shunt (5/86) | CSF leak | good |
| 11 | F | 20 | 23 | ant fossa dec (7/88) post fossa dec (11/88) bifrontotemp dec (7/91) | osteitis; difficult laryngeal intubation | prosthesis removed good |
| 12 | F | 33 | 37 | ant fossa dec (2/88) post fossa dec cisternoatrial shunt (4/88) | persistent headaches | intellect subnormal |
| 13 | F | 47 | 48 | ant fossa dec (9/91) | sagittal sinus laceration; bacterial meningitis; intracerebral hematoma | blind & deaf |
| 14 | M | 16 | 17 | ant fossa dec (4/91) post fossa dec (5/91) | | good |

* Ant fossa dec = anterior fossa decompressive craniotomy and placement of acrylic prosthesis (bifrontal decompressive craniotomy); post fossa dec = posterior fossa decompressive craniectomy; LP = lumboperitoneal; bifrontotemp dec = bifrontotemporal decompressive craniotomy; CSF = cerebrospinal fluid.

There were eight females and six males, with a mean age of 20.5 years (range 9 to 47 years) at the time of their first neurosurgical procedure (Table 1). All cases were referred by surgeons in the Ear, Nose, and Throat Department who had evaluated the patients for unilateral or bilateral peripheral facial nerve palsy and hearing loss. Examination revealed cutaneous or bone syndactyly of the second and third fingers in 10 patients and

some form of nail dystrophy of the fingers or toes in all 14 patients. Changes in the optic discs included papilledema in five cases, optic atrophy in one, and the presence of optociliary shunt veins in five. Only one patient (Case 13) had severe impairment of visual acuity (light perception only); this was also the oldest patient in the series. All patients were taller than 1.8 m, had generalized bone overgrowth (mainly of the calvaria



FIG. 1. Follow-up photograph of the patient in Case 2 (aged 27 years). Note the bilateral facial palsy and the overgrowth of bone in the calvaria and mandible.

and mandible), with proptosis, malalignment of the teeth, and craniomegaly (Fig. 1).

Radiological Findings

Computerized tomography (CT) of the brain revealed an increase in skull thickness, and bone windows demonstrated an increase in bone density. The ventricles and basal cisterns appeared compressed and the cerebrospinal fluid (CSF) spaces over the convexities were obliterated (Fig. 2 *left*). The prominent crista frontalis and bone overgrowth in the posterior fossa are common features in the radiological diagnosis (Fig. 3). Regrowth of bone after frontal decompressive surgery is shown in Fig. 4.

Surgical Treatment

All patients underwent surgery as summarized in Table 1. A total of 42 neurosurgical procedures were performed on these 14 patients, consisting of 12 bifrontal (anterior fossa) and five bifrontotemporal decompressive craniotomies, 16 posterior fossa decompressive craniectomies, two lumbar decompressive laminectomies, six lumboperitoneal shunt procedures, and one cisternoatrial shunt placement.

Results

Decompressive Craniotomy

Eight patients had a bifrontal decompressive craniotomy as the initial procedure, while a posterior fossa decompressive craniectomy through the posterior rim of the foramen magnum and a C-1 laminectomy was performed as the first procedure in six patients. The posterior fossa dura mater was routinely opened in all cases. Opening and grafting of the supratentorial dura

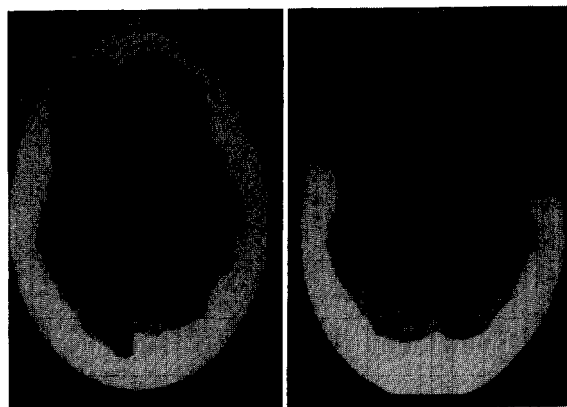


FIG. 2. Computerized tomography scans in Case 5. *Left*: Appearance before surgical decompression showing the thick, dense, irregular bone and ventricular compression. *Right*: Appearance 2 days after bifrontotemporal decompression without opening of the dura. Trapped air bubbles are seen above and below the acrylic prosthesis. The brain has expanded into the newly created cavity.

was abandoned in the five most recently treated patients due to problems experienced with subcutaneous CSF collections in earlier cases. The average duration of the decompressive surgical procedure was $5\frac{1}{2}$ hours. Blood loss from the bone was not a problem as the bone was relatively avascular.

Patients who developed raised ICP after previous decompressive surgery invariably showed signs of bone regrowth under the frontal prosthesis and in the posterior fossa (Figs. 3 *left* and 4). In five such patients, the newly formed bone was removed from under the prosthesis at craniotomy, bitemporal craniectomies were performed, and the same prosthesis was replaced. It took between 3 and 13 years (average 6.6 years) for bone to regrow sufficiently to cause new symptoms of raised ICP after previous decompressive surgery.

Complications

Two patients in the group receiving posterior fossa decompression died postoperatively: one patient (Case 3) underwent a bifrontal decompression procedure, after which ICP again increased; in the other (Case 6) the posterior fossa decompression was performed before supratentorial decompressive surgery. Both of these patients died within 24 hours postoperatively. Both were alert and neurologically intact after surgery; they developed sudden cardiovascular collapse after endotracheal suctioning in the intensive care unit prior to extubation. Postmortem examination confirmed downward migration of the brain stem and signs of pressure necrosis in the lower medulla oblongata.

One patient (Case 10) developed a CSF fistula through the posterior fossa incision, which resolved after a lumboperitoneal shunt was inserted. It was necessary to relieve raised ICP in the supratentorial compartment before posterior fossa decompressive surgery could be performed safely. The four patients with their first operation after 1985 underwent bifrontal decompression as the initial procedure.

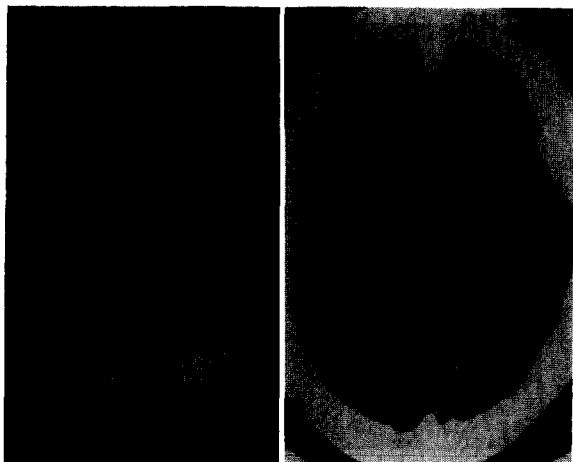


FIG. 3. Computerized tomography scans in Case 8. *Left:* Scan with bone windows of the skull base obtained 7 years after posterior fossa decompression showing bone overgrowth and narrowing of the foramen magnum. *Right:* Scan showing thick, irregular bone and prominence of the crista frontalis.

Symptoms of progressively increasing ICP after bifrontal and posterior fossa decompressive surgery due to continuous bone overgrowth have occurred in five of the 14 patients. Early in the series, we attempted to alleviate these symptoms by CSF diversionary procedures. Ventriculoperitoneal shunting could not be employed due to the small size of the ventricles (Fig. 2). Six patients underwent lumboperitoneal shunt placement and one received a cisternal shunt. In four, the shunts were removed due to complaints of postural headaches; it is uncertain whether the shunts in the three asymptomatic patients are still functional. No CSF diversionary procedures were performed after 1986.

The two patients over 30 years of age (Cases 12 and 13) had an unfavorable outcome. Both were operated on at a late age (33 and 47 years) due to late referral for surgery. One patient (Case 12) has a subnormal intellect, while one (Case 13) is blind and deaf.

Intraoperative complications were related to the thickness and density of the bone. The oldest patient (Case 13) had the thickest bone (38 mm); she came to surgery at the age of 47 years. She developed numerous complications including laceration of a venous lacuna that necessitated blood transfusion, postoperative meningitis that was treated successfully, and a postdecompression intracerebral hematoma that was managed conservatively.

Discussion

Natural History of Sclerosteosis

There is no currently available medical treatment for sclerosteosis. Surgical decompression of the facial nerves has produced alleviation of symptoms in some patients.⁶ Middle-ear surgery to relieve conductive deafness is indicated and partially successful. The major cause of morbidity and mortality is a steadily rising ICP. Elevated ICP in these patients is a cause of intel-

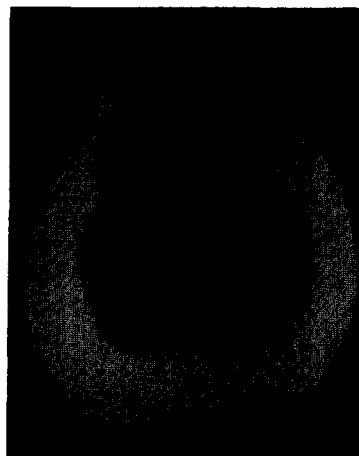


FIG. 4. Case 11. Computerized tomography scan obtained 3 years after bifrontal decompression. New bone has formed under the frontal prosthesis, and the subarachnoid space over the convexity is compressed.

lectual impairment and deterioration of vision, and may lead to sudden death at a young age.⁵ This is caused by medullary compression in the foramen magnum during periods of acute ICP increase, as seen during episodes of coughing or straining.⁵

These patients are born with a normal intelligence, and adequate cranial decompressive procedures improve their quality of life and length of survival. The incidence of osteoblastic hyperactivity tends to decrease after the third decade of life.^{3,5} If a normal ICP is maintained until that time, these individuals might lead a relatively normal life afterward.

Visual Impairment

Only one patient in the series (Case 13) had severe visual impairment. She was the oldest in the series and had long-standing severely raised ICP. It is uncertain to what extent optic foraminal stenosis attributed to the condition. A more common cause of decreased visual acuity is raised ICP that responds well to cranial decompressive surgery. None of the patients in this series who underwent surgery when younger than 30 years of age had evidence of symptomatic optic foraminal stenosis. As this group of patients is expected to survive longer than their predecessors, more cases with cranial nerve involvement other than the seventh and eighth nerves may be seen in the future.

Surgical Management

A generous bifrontal decompressive craniotomy has yielded the best results as the initial form of treatment to control progressively rising ICP due to bone overgrowth of the calvaria and skull base. The frontal sinus is preferably not included in the flap and the large bone defect is covered with a thin acrylic prosthesis. It is suggested that this procedure be routinely performed in all cases, soon after puberty if possible. Early surgical intervention has many advantages: the bone is softer and has not yet reached maximum thickness. Also, the

dura mater has not yet undergone pressure atrophy and the chances are better of maintaining the dura intact, thus preventing underlying brain injury and subcutaneous CSF accumulation. Early surgery results in a shorter operating time and therefore a decreased risk of sepsis. The risks of postdecompression brain swelling and intracerebral hemorrhage are also decreased by early surgical intervention.

The use of standard neurosurgical power instruments is not satisfactory. The thickness of the bone is such that ordinary drill bits are too short to reach the dura. Similar problems are experienced with standard craniotomes. The density of the bone also tends to damage power-driven instruments. We therefore use hand instruments for the craniotomy. Two large burr holes adjacent to one another with no intervening bone make a less acute angle through which the saw guide has to pass, and decreases the chance of dural laceration and harmful compression of the underlying brain. The crista frontalis is often large and space-occupying (Fig. 3 right); this is drilled, but care is taken not to enter the paranasal sinuses that were fortunately not prominent in any patient. Dural hitching stitches are no longer used, as these stitches have been associated with the collection of subcutaneous CSF. The dura mater is invariably very adherent to the bone, due to the raised ICP, and postoperative extradural hematoma was not seen after this practice was abandoned.

A posterior fossa decompressive craniectomy, combined with a C-1 laminectomy and opening of the dura mater, was performed after the supratentorial compartment had been adequately decompressed. This decompressive craniectomy was performed prophylactically, even in asymptomatic patients. The two postoperative deaths suggest that this procedure may be fatal in patients with raised ICP in the supratentorial compartment, due to downward herniation of the lower brain stem after posterior fossa decompression. If the foramen magnum reossifies after surgery, as was seen in three cases in this series (Cases 3, 8, and 9), it should be ascertained that there is no association with raised ICP in the supratentorial compartment before reoperation in the posterior fossa is performed. This was the cause of death in one patient (Case 3). An adequate supratentorial decompression also prevents the development of a CSF fistula or pseudomeningocele after posterior fossa surgery.

Management of Recurrence of Raised Intracranial Pressure

When increased ICP recurs after previous decompressive surgery, the supra- and infratentorial compartments are again decompressed. The new bone tends to be thinner and somewhat softer than the surrounding bone. The frontal prosthesis is easily separated from the underlying new bone and can be used again after the newly formed bone has been removed. Bitemporal craniectomies can be included in the procedure to create extra space. Unilateral temporal decompression may lead to midline shift. Cerebrospinal fluid diversion-

ary procedures in patients with raised ICP caused by the regrowth of bone are ineffective and potentially harmful.

Anesthetic Management

The anesthetic management of these patients poses unique problems. Overgrowth of the mandible and an inability for the patient to open the mouth wide make laryngeal intubation difficult. A fiberoptic laryngoscope has been used successfully in the five most recently treated patients. The heavy, broadened ribs, scapulas, and clavicles cause decreased pulmonary compliance, and ventilator adjustments are necessary to ensure satisfactory blood gas levels intraoperatively.¹⁴ The patients also have difficulty in hearing without their hearing aids; if this is not kept in mind, it may cause concern when the patients' level of consciousness is assessed postoperatively in the recovery room.

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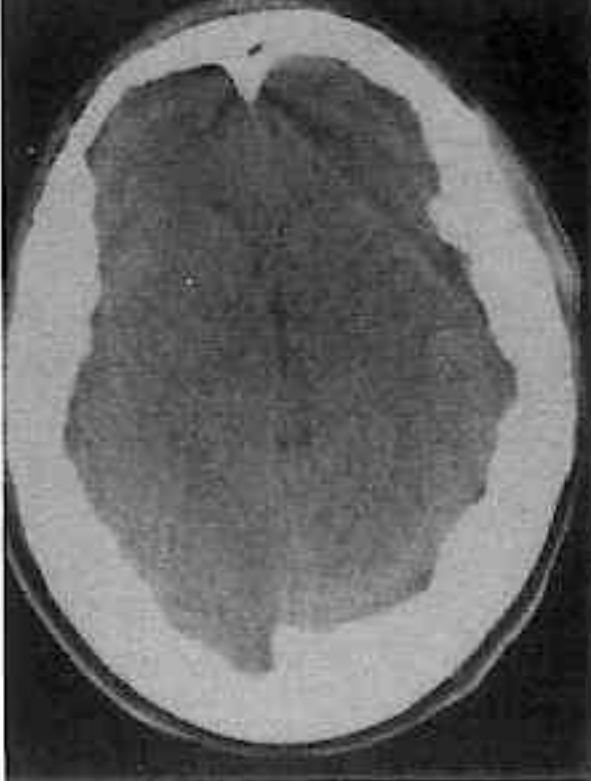
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