Optic nerve decompression in osteopetrosis

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Osteopetrosis is a rare disorder characterized by generalized increased skeletal density with abnormalities of bone modeling. The skull base is usually involved. Loss of vision with optic nerve atrophy is the most common neurological finding and is traditionally believed to be the result of optic nerve compression within the compromised optic canal. However, retinal degeneration has recently been described and is hypothesized to be the etiology of the visual loss (thus challenging the value of surgical decompression).

The authors report their experience with six children suffering from osteopetrosis and severe visual loss. All six patients underwent bilateral microsurgical optic nerve decompression through a supraorbital craniotomy. Improvement in visual acuity occurred postoperatively in five patients, and none had complications. Two technical points are emphasized: 1) optic nerve decompression should be wide and include not only unroofing of the canal but also drilling along both sides of the optic nerve, and 2) the thick, irregular, and highly domed orbital roof should be smoothed down by high-speed drilling to facilitate surgical exposure with minimal retraction of the frontal lobe.

KEY WORDS • osteopetrosis • optic nerve decompression • supraorbital craniotomy

Osteopetrosis is a group of disorders caused by defective osteoclastic function. It is characterized by a generalized increase in skeletal density and by abnormalities of bone modeling, with a proclivity for involvement of the skull base. Cranial nerve deficits, particularly associated with the second, seventh, and eighth nerves, are known to occur in these patients. Visual loss with optic atrophy is the most common neurological finding and is traditionally believed to be the result of optic nerve compression within the narrowed optic canal. This hypothesis has been supported radiologically, as well as by autopsy findings. Successful optic nerve decompression with resulting visual stabilization or improvement is reported rarely. Recently, retinal degeneration has been described in cases of osteopetrosis and is hypothesized to be the etiology of the visual loss. These reports thus challenge the theory of optic nerve compression and the value of surgical decompression. We are reporting our experience with six patients suffering from osteopetrosis and severe loss of vision.

Summary of Cases

Among 10 children with osteopetrosis evaluated at the King Faisal Specialist Hospital and Research Center, eight had severe loss of vision. Six of these patients underwent bilateral optic nerve decompression in 1985. Table 1 depicts the visual acuity of these patients on preoperative, postoperative, and follow-up examinations. Only one patient showed visual deterioration on follow-up review after initial postoperative improvement.

During the preoperative evaluation, particular attention was given to the possibility of sepsis and bone-marrow depression. Optic canal encroachment was documented radiologically by computerized tomography (CT). Caution was exercised during the positioning of the patient on the operating table in order to avoid injury to the craniovertebral junction, which is usually involved.

The supraorbital approach described by Jane, et al., was utilized. The right side was selected for bilateral optic nerve decompression in all six patients. To minimize frontal lobe retraction, the irregular, highly domed orbital roof (Fig. 1) was completely smoothed down by drilling prior to opening the dura. When the dura was opened, cerebrospinal fluid was released and the optic nerves were identified. The perioptic cisterns were usually dilated. The dura over the optic canals was coagulated and dissected away. The thick but fragile bone of
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FIG. 2. Case 1. The facies typical of a patient with malignant osteopetrosis.

Representative Cases

Case 1

This 7-year-old boy was the son of parents who were first cousins. He was admitted on September 1, 1985, with a history of progressive visual and hearing loss. His parents reported that he had had normal vision in early childhood. In September, 1984, his visual acuity was 20/100 in both eyes.

At this admission his visual acuity was limited to counting fingers from a distance of 1 ft in the right eye and was 20/70 in the left. He had bilateral optic atrophy, a peculiar facies (Fig. 2), and hepatosplenomegaly. The rest of the general and neurological examination findings were within normal limits. Auditory brain-stem evoked response testing revealed delay of all waves.

<table>
<thead>
<tr>
<th>Case No.</th>
<th>Age (yrs), Sex</th>
<th>Preop Visual Acuity</th>
<th>Postop Visual Acuity</th>
<th>Follow-up Visual Acuity</th>
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</thead>
<tbody>
<tr>
<td></td>
<td>Age, Sex</td>
<td>Date, OD, OS</td>
<td>Date, OD, OS</td>
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<tr>
<td>1</td>
<td>7, M</td>
<td>9/85, CF at 1 ft, 20/70</td>
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<tr>
<td>2</td>
<td>8, F</td>
<td>9/85, 20/300, 20/200</td>
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<tr>
<td>3</td>
<td>17, F</td>
<td>12/85, 20/200, 20/200</td>
<td></td>
<td></td>
</tr>
<tr>
<td>4</td>
<td>14, M</td>
<td>5/85, cannot follow objects or walk alone</td>
<td></td>
<td></td>
</tr>
<tr>
<td>5</td>
<td>7, M</td>
<td>10/85, 20/400, 20/400</td>
<td></td>
<td></td>
</tr>
<tr>
<td>6</td>
<td>6, M</td>
<td>12/85, CF at 1 ft, CF at 3 ft</td>
<td></td>
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</tr>
</tbody>
</table>

* OD = right eye; OS = left eye; CF = counting fingers.
especially on the right. A bone survey showed a generalized increase in density consistent with the diagnosis of osteopetrosis. A CT scan of the head showed osteopetrosis at the base of the skull with severe narrowing of the optic canal and internal auditory meati bilaterally (Fig. 3). On September 9, the patient underwent bilateral optic nerve decompression. A week later his visual acuity improved to 20/40 in both eyes, which was maintained at follow-up evaluation in January, 1987.

Case 2

This 8-year-old girl was the sister of Case 5 (Table 1). She was first evaluated at the age of 3 years 8 months and was found to have osteopetrosis, renal tubular acidosis, and brain calcification. Since then she had suffered repeated fractures and dental caries, and exhibited retardation of growth and mental development. In 1979, she had no report of visual loss and had a normal fundus examination.

On reevaluation in February, 1985, the patient had visual acuity of 20/300 in the right eye and 20/200 in the left, with bilateral optic atrophy and nystagmus. A CT scan showed severe optic canal narrowing and extensive brain calcification (Figs. 4 and 5). She underwent bilateral optic nerve decompression on February 17; postoperatively, there was subjective improvement with disappearance of her nystagmus. On September 29, 1985, she had visual acuity of 20/200 in the right eye and 20/70 in the left, this remained stable on follow-up evaluation in September, 1986.

Discussion

In 1904 the German radiologist, Albers-Schönberg, described a 26-year-old man with generalized skeletal sclerosis and multiple fractures. He introduced the term "marble bone disease." In 1926 Karshner coined the term "osteopetrosis." Osteopetrosis is an uncommon genetic disorder with subtypes characterized by increased skeletal density and abnormalities of bone modeling. It is believed to be mainly the result of defective osteoclastic function with resulting impairment of bone resorption. In the classical manifestation, osteopetrosis occurs in either a malignant recessive form or a benign dominant form. The benign dominant form of osteopetrosis occurs in adolescents and adults. The diagnosis is often made by roentgenographic study alone. Normal longevity may be expected. Since the dominant form is most consistent with the description by Albers-Schönberg, Beighton, et al., suggested that the designation "Albers-Schönberg disease" be restricted to the dominant type of osteopetrosis. The recessive form, often reported with consanguinity, has a high association with neurological and hematological involvement, leading to a shortened life span. Ohlsson, et al., described the recessive type in association with renal tubular acidosis and intracranial calcification in three patients, two of whom are included in our series. Carbonic anhydrase II deficiency was recently reported in association with the above subtype of osteopetrosis.

Neurological complications frequently occur, particularly in the recessive malignant type. These include optic atrophy with blindness, nystagmus, macrocephaly, strabismus, facial paralysis, deafness, hydrocephalus, intracranial hemorrhage, mental retardation, convulsions, and trigeminal nerve involvement. Successful results have been reported following decompression of the facial nerve and the acoustic nerve.

A frequent finding in osteopetrosis is visual loss with optic atrophy. It is generally attributed to the mechanical encroachment upon the optic nerve within the narrowed optic canal. This hypothesis has been supported by radiological and postmortem examinations. However, other hypotheses have been pro-
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FIG. 4. Case 2. Axial computerized tomography scan (lower) and perpendicular reconstruction across the right optic canal (upper) showing the severe narrowing of the optic canal almost to the point of complete obliteration (arrow).

posed, including primary dysmyelination of the optic nerve, atrophy secondary to papilledema from compression of the retinal vein, primary increased intracranial pressure, and hydrocephalus. Keith has shown pathological evidence of retinal degeneration. Subsequently, Hoyt and Billson described three patients in whom an electroretinography study indicated diffuse retinal degeneration. One of their three patients underwent optic nerve decompression and continued to have a progressive loss of vision after surgery. Thus, they questioned the value of optic nerve decompression in their osteopetrosis patients.

Failure to improve or stabilize vision by decompression has resulted in some reservation and disappointment. Optic nerve decompression via craniotomy has been reported to stabilize vision in only four patients, and we could find improvement in only one patient. Another patient's vision was stabilized after optic nerve decompression was carried out through the transethmoidal approach. On the other hand, Guyer, et al., reported on 11 patients with optic nerve decompression for various lesions (none with osteopetrosis); 90% of these cases improved or stabilized immediately following surgery. Visual stabilization or improvement has also been reported with optic nerve decompression in cases of fibrous dysplasia. The visual improvement in five of our six consecutive patients is, in our opinion, due to the more extensive decompression carried out under the operative microscope. The nystagmus is believed to be of amblyopic etiology. The disappearance of nystagmus in two of our patients after improvement of vision supports this theory.

Optic foramen views might not be very reliable in children, especially in patients with osteopetrosis. Optic canal tomography has been accurate in documenting both enlargement and narrowing of the optic canal. This radiological investigation is currently obtained with the aid of CT scanning. Coronal and sagittal reconstruction will enhance the radiological demonstration of optic canal encroachment.

Conclusions

In patients with osteopetrosis, extensive microsurgical optic nerve decompression is a safe and effective method of preserving or improving vision. The supraorbital approach is recommended, and both optic nerves can be decompressed at the same procedure.

Acknowledgment

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