The Surgical Management of Cranio-orbital Neurofibromatosis

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Experience with the surgical management of cranio-orbital neurofibromatosis in 14 patients is reported (age range, 6–40 years). The skeletal abnormality of the orbit that occurs in a small proportion (less than 1%) of patients with neurofibromatosis is, in essence, the absence of the membranous portions of the sphenoid and the adjacent bone forming the boundaries between the cranium and the orbit. The goal of surgery is tumor resection and reconstruction of the posterior bony defect by bone graft. Two additional procedures are also described that better enhance aesthetically—the mask lift and facial tissue expansion.

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Neurofibromatosis is a hereditary condition occurring in 1 in 3,000 live births. It is transmitted in an autosomal-dominant pattern with varying expressivity. The term neurofibromatosis is applied to two clinical genetic disorders. The first, neurofibromatosis type 1 (NF 1), also known as von Recklinghausen NF [1], is more common. The second, neurofibromatosis type 2 (NF 2), is a rarer condition and is known as bilateral acoustic NF. The two disorders have very distinctive clinical manifestations, with some overlapping features. Mutation of genes on two different chromosomes are at the origin of the two disorders. The gene for NF 1 is located on the long arm of chromosome 17, while the gene responsible for NF 2 is located on the long arm of chromosome 22.

Our paper deals with the cranio-orbital localizations of von Recklinghausen's NF. This localization occurs in only a small number of patients with neurofibromatosis (less than 1%). The clinical features of the disease include orbital and eyelid manifestations. The neurofibroma involves the orbit, the eyelids, and the temporal region to a variable extent.

The first clinical sign is usually a slight swelling of the eyelid that begins in childhood and slowly increases. Progressive and severe involvement of subcutaneous tissues of the eyelid is responsible for varying degrees of lid ptosis, due to infiltration of the levator muscle and increase in weight of the upper lid. At this stage, the eye usually remains functional.

In the most severe forms, there is considerable swelling of the eyebrow and frontoparietal region. The lesion may cover half of the face, with associated gigantism. In this case, there is gross involvement of the eyelids, making it impossible to open the eye. The globe is involved with the neurofibroma and there may be buphthalmos, severely diminished visual acuity, and even blindness. Associated symptoms are irritation, pain in the eye, and moderate-to-severe epiphora (Fig 1A).

A complete medical eye examination and an ophthalmologist are very important in the surgical management of the disease.

Craniofacial skeletal abnormalities are constant in computed tomography (CT) scans.

Partial or complete absence of the greater wing of the sphenoid is responsible for enlargement of the sphenoidal fissure, with a consequent defect in the posterior wall of the orbit. Brain tissue, generally the temporal lobe, may herniate into the orbit (Fig 1C), further increasing exorbitism and causing pulsation of the eye. The orbit is enlarged, with hypoplasia of the supraorbital and infraorbital rims and the zygomatic arch. Occasionally, an associated arachnoid cyst may be found.

Enophthalmos is occasionally encountered (Fig 2). The causative factors seem to be the enlargement of the inferior orbital fissure, which allows the orbital contents to prolapse into the infratemporal fossa, and the increase in size of the orbital cavity.

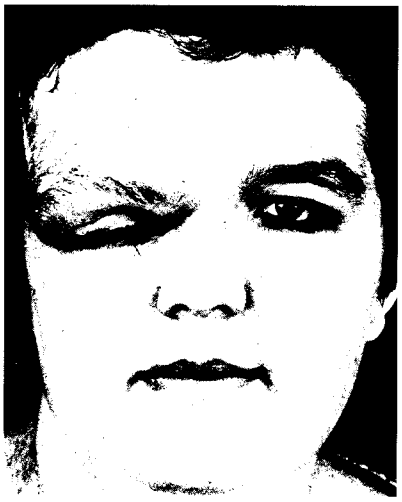

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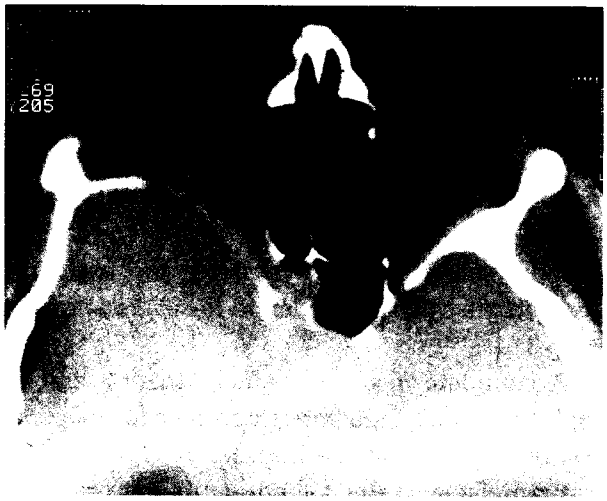
Fig 1. (A) Cranio-orbital neurofibromatosis with pulsatile exophthalmos, ptosis, inferior displacement of the globe, and enlargement of the orbit. Vision is normal. (B) After all the steps of surgery: tumor resection and orbital reconstruction. The result is presented after a mask lift and facial aesthetic sculpturing. We can see the effect on the harmony of the face, which is now less aggressive and more pleasant. (C) Axial CT scan of the patient shows the enlargement of the orbit. We can see the prolapse of the frontal lobe into the orbit. (D) CT scan after reconstruction. No resorption of the bone graft is seen after 6 years of follow-up.



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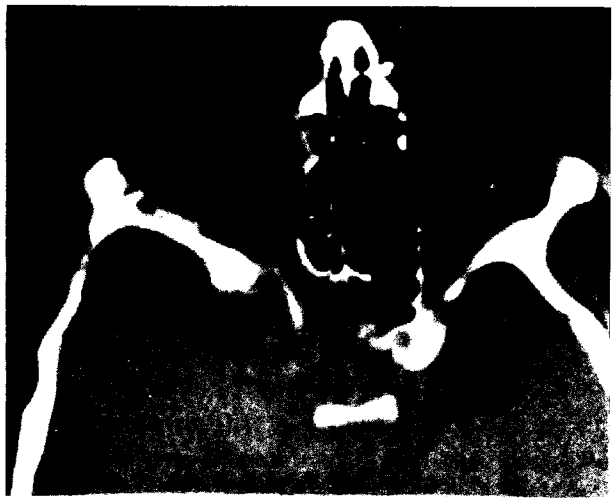

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This paper will report our experience in the surgical management of this disease.

Materials

Fourteen patients (6 men and 8 women, age range 6–40 years) with cranioorbital neurofibromatosis have been operated on since 1987. Positive family history was found in 3 cases. The common clinical signs of the disease were found in 9 patients, including café au lait spots, neuromas, and subcutaneous neuromas. The cranio-orbital localization was characterized by exophthalmos in 8 cases (in 6 cases, it was pulsatile), enophthalmos in 3 cases, and no skeletal involvement in the remaining 2 patients. Tumor infiltration of the upper lid was present in all cases, whereas the lower lid was involved only in 3 cases.

All patients underwent a complete ophthalmologic examination, which revealed severe visual impairment of the affected eye in patients with pulsatile exophthalmos. Facial bone radiographs allowed comparison of orbital shape and size. CT scans and magnetic resonance imaging demonstrated the absence of the greater wing of the sphenoid, found in 11 cases, responsible for enlargement of the orbital fissure and consequent herniation of the temporal lobe into the orbit.

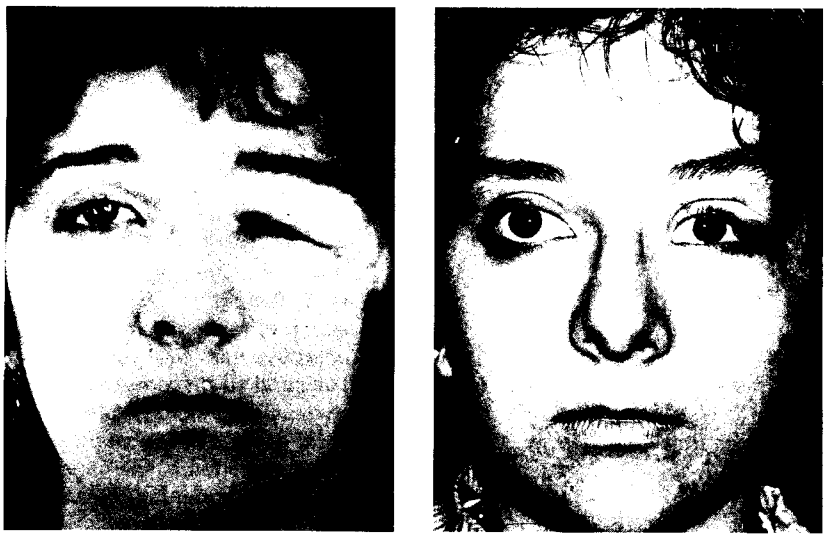

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Fig 2. Orbitotemporal neurofibromatosis with enophthalmos. There is no visual impairment. (A) Preoperative view. (B) Postoperative view at 4-year follow-up. The surgical procedure includes tumor resection by a bicoronal approach and orbital reconstruction by calvarial bone graft. A subperiosteal dissection with facial bone remodeling and bilateral canthopexies was performed 1 year after the first operation.

Three-dimensional imaging played a major role in the assessment of soft-tissue and bone deformities.

In the 3 cases with enophthalmos, a moderate degree of orbital enlargement was observed. The explanation seems to be that the area of sphenoid absence affects the posterior portion of the lateral orbital wall, allowing the escape of orbital contents into the orbital fossa. This, together with the increase in bony orbital volume, results in enophthalmos.

Methods

Neurofibromatosis is often associated with a myriad of cutaneous and skeletal abnormalities that present problems for the plastic surgeon. Surgical treatment must be approached with the understanding that all the cells carry the abnormal gene. The usual goal of total excision cannot be applied to this disorder, because so many cells have the potential for developing tumors.

Surgical procedures can, however, be very beneficial in removing abnormal tissue and reconstructing skeletal defects that interfere with function or cause cosmetic concern. In the literature, it has been suggested that surgical interference might increase the known risk of malignant generation of this disease, but this hypothesis is no longer considered true.

Surgical treatment includes five stages: selective arteriography, tumor resection and orbital reconstruction, eyelid surgery, management of the eyeball, and ancillary procedures.

1. Selective Arteriography

To prevent excessive bleeding during tumor resection, we perform, in some patients, a selective arteriography that can be followed by selective arterial embolization, if necessary. This is performed especially in cases of gigantic tumor.

2. Tumor Resection and Orbital Reconstruction

With the advent of craniofacial surgical techniques, repair of the bony defect using an intracranial approach is a more feasible proposition. Repair of the bony defect in the orbit is, in fact, not a new procedure, having been performed with a cranial bone graft by Dandy (a Baltimore neurosurgeon) in 1927 with a good result [2].

We use a coronal and upper lid approach to resect the tumor from the underlying tissues. We always try to accomplish total resection of the tumor, although the resection is very difficult and bleeding is usually conspicuous. In one case, preoperative embolization of the tumor was carried out to reduce bleeding.

In the case of cerebral herniation into the orbit, an intracranial approach is used. A frontotemporal craniotomy is carried out by the neurosurgical team, with an extradural dissection to separate the dura from the orbital contents and expose the boundaries of the bony defect.

In some cases, to completely separate protrud-

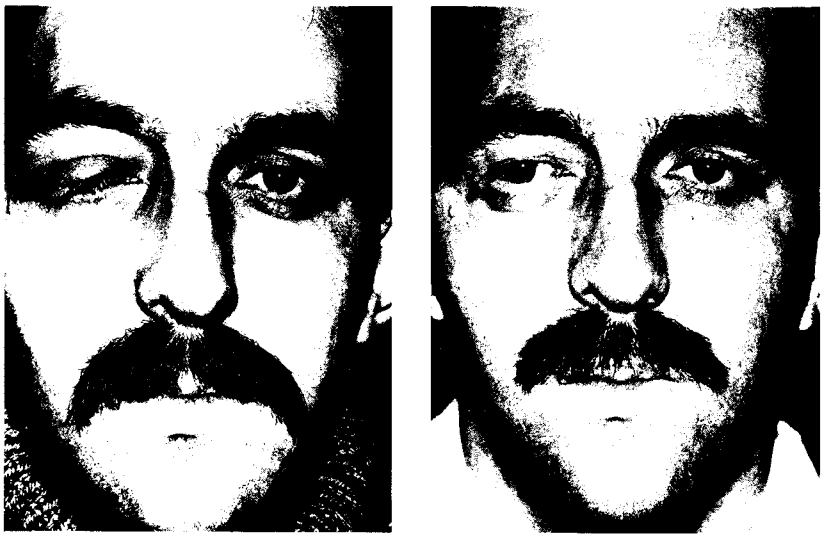

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Fig 3. (A) Patient with eyelid involvement of neurofibromatosis. (B) Postoperative view at 5-year follow-up. The surgical procedure includes tumor resection by a bicoronal and upper lid approach. Six months later, the correction of the ptosis was performed by shortening the levator muscle.

ing brain from the periorbita the optic nerve has to be cut. This procedure was performed in 2 patients with major pulsatile exophthalmos. In the other cases, minimal dissection was done to preserve optic and ocular nerves and, consequently, vision (even if low) and mobility of the globe. The dissection should, however, allow repair of the bony defect.

The dura is plicated where necessary over the temporal lobe and the bone defect is repaired using cranial bone grafts obtained by splitting the craniotomy bone flap. Bone grafts are placed on the orbital floor to elevate, if necessary, to correct globe dystopia. When an intracranial procedure is not necessary, bone grafts are harvested from the parietal region.

3. Eyelid Surgery

Tumor involvement of the upper lid is usually responsible for ptosis (Fig 3). Excess eyelid skin is removed after measuring the difference with respect to the healthy contralateral side. The levator aponeurosis is shortened according to the degree of preoperative ptosis. Levator shortening should not be overdone. It is better to reoperate for insufficient correction, than to have a short lid that leads to corneal exposition.

4. Management of the Eyeball

The general rule is to preserve the globe as long as there is useful sight, as there is always the possibility of loss of vision in the normal eye. If, in time and following several surgical attempts, the globe is proptotic and has no vision or, in the case of buphthalmos or a painful eye with impaired visual acuity, evisceration is justified. Enucleation was performed in 1 patient with buphthalmos. In these instances, remodeling of the orbital framework, resection of residual tumor, and prosthetic replacement is simplified and results in improved cosmetic appearance.

5. Ancillary Procedures

It must be reemphasized that the surgical procedures described cannot eradicate the disease. Neurofibromatosis progresses slowly and irregularly, with possible accelerations during puberty and pregnancy. Nevertheless, carefully planned and staged procedures can improve contour and function. Additional procedures can be done years to decades later to enhance cosmetic results and for the well being of these patients.

A mask lift with facial aesthetic sculpturing is a newcomer to aesthetic surgery and a viable ancillary procedure. A mask lift seeks to normalize, rejuvenate, and embellish the face through a subperiosteal face lift and transformation of the underlying structures. In fact, the typical sagging aspect of the face in this disease is due to drooping of the tissues caused by the weight of the tumor and progressive infiltration of the skin. Through the same bicoronal incision used for tumor resection and the intracranial approach, the subperiosteal dissection is carried out, expos-

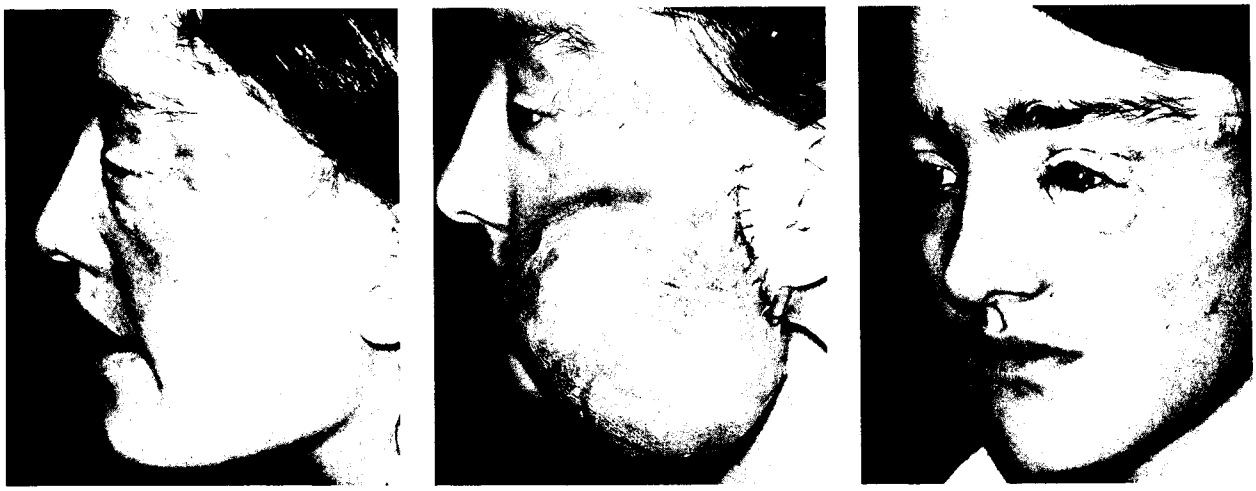

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Fig 4. (A) Orbitotemporal neurofibromatosis with tumoral skin in the parietal region. (B) Skin expander in the cheek. (C) Result after tumoral skin excision at 1-year follow-up.

A B

Fig 5. (A) Orbitotemporal neurofibromatosis with upper and lower eyelid involvement, and no vision impairment. (B) Three years after surgery. One can see the position of the lateral canthi on both sides.

ing the forehead and the orbital region. Onlay bone grafts permit a full-thickness redraping of the soft tissues of the face. Lifting the facial mask then permits optimal harmony of the face, reinforced by bilateral canthopexy, which also corrects globe dystopia.

A second ancillary procedure is tissue expansion to complete the tumor skin resection of the frontoparietal region (Fig 4). An expander is placed under the normal skin in the cheek through a preauricular incision. The expander is inflated every day for 2 weeks, after which it is removed and tumor skin excision is carried out followed by redraping of the expanded skin flap. This procedure has been carried out in 2 patients with a good cosmetic result.

Results

The results of surgery can be satisfactory if the whole tumor is removed. This occurred in patients with tumor infiltration limited to the upper lid and no skeletal involvement. The most unsatisfactory part of the treatment is ptosis correction, which requires repeated procedures. Care must be taken not to overelevate the lid, as this would lead to corneal exposure and consequent complications.

Two patients with pulsating exophthalmos and severe visual impairment underwent complete separation of the brain (temporal lobe) from the periorbita after severing of the optic nerve. The whole orbit was reconstructed with bone grafts (cranial and iliac) and the eyeball was retained and covered with conjunctiva. The prosthesis placed over the eyeball is immobile after surgery.

One patient with pulsatile exophthalmos and buphthalmos had enucleation without section of the optic nerve and, in this case, the prosthesis placed over the eyeball kept some mobility. In the remaining cases with exophthalmos, dissection between brain and periorbital tissues was

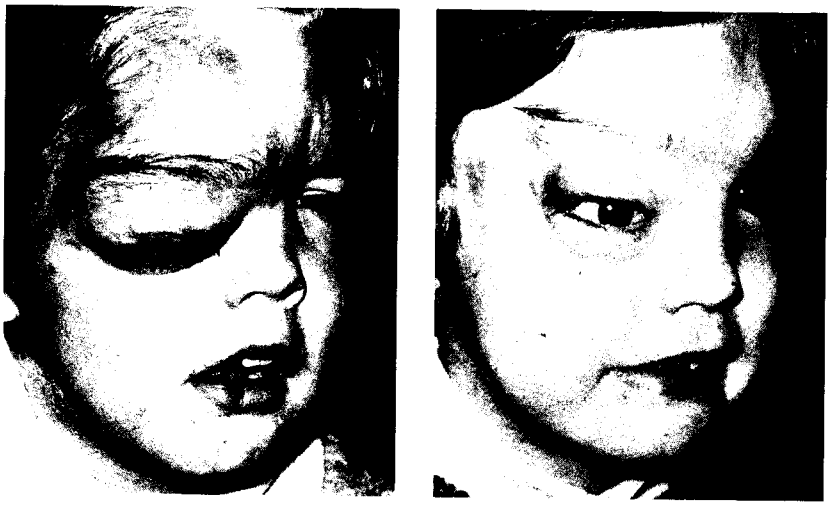

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Fig 6. (A) A 7-year-old patient with orbitotemporal neurofibromatosis, severe exophthalmos, and visual impairment. (B) Postoperative view at 2-year follow-up. The surgical procedure included a bicoronal and upper lid approach with tumor resection and orbital reconstruction (posterior wall and floor of the orbit). We performed eye evisceration 6 months later and an ocular prosthesis was placed with very low mobility. The next operation will be tissue expansion to remove the tumoral skin from the frontoparietal region.

stopped short of the optic nerve, preserving it. These patients showed mild visual impairment before surgery and maintained the same visual acuity after the procedure. Exophthalmos was corrected and no pulsation was seen after surgery. The patients were very happy to be able to retain their eye and visual acuity.

The result was satisfactory in the patient with enophthalmos.

We have seen no recurrence of exophthalmos or enophthalmos within a follow-up of 5 years in our series, although it has to be kept in mind that these patients carry the potential to develop the disease in their cells.

If the surgical procedure is carried out through abnormal tissue, wound healing of skin edges may be delayed, but surgical scars are usually more than acceptable with a minimal tendency to hypertrophy. Tissue expansion can reduce the scars. Bleeding is often persistent and insidious during surgery but, is not usually life threatening. In 3 cases, we have done tumor embolization to prevent excessive bleeding during the operation.

The ancillary procedures described and performed in 11 patients gave them a better cosmetic appearance. No major complications were observed in our series following the intracranial or the extracranial procedures.

Discussion

Surgical management of cranio-orbital neurofibromatosis is difficult to standardize because of its polymorphism and unpredictable evolution. Early surgery is recommended when excessive tumor growth in size and extension occurs. We operate in case of ptosis and pulsating exophthalmos in order to avoid loss of vision and complications due to brain herniation into the orbit. We also operate in cases of severe enophthalmos and extensive tissue infiltration by the tumor without skeletal abnormalities to prevent facial dysmorphism. The goal of surgery is to prevent loss of vision and to give these patients a better aesthetic appearance (Fig 5).

Because the lesions are not defined and total resection results in major orbitopalpebral mutilations, many authors recommend partial resection [3, 4], as only well-defined tumors can be totally removed. Van der Meulen and colleagues [5], and Jackson and associates [6], have recommended orbital tumor resection with preservation of part of the eyelids and sockets. The main problem is, actually, whether or not the eye can be preserved [7–9].

Recently, Jackson and colleagues [10] proposed a three-group classification of orbitotemporal neurofibromatosis. Group 1 is orbital soft-tissue involvement with a seeing eye. Group 2 is orbital soft-tissue and significant bony involvement with a seeing eye. Group 3 is orbital soft-tissue and significant bony involvement with a blind or absent eye. The authors recommend eye conservation in the first two groups whenever possible and radical treatment in the last group.

In 2 patients, the optic nerve was cut and a

major orbital bony reconstruction was carried out. This resulted in sacrifice of the eye and an immobile prosthesis. In the other cases with pulsating exophthalmos, the optic nerve was preserved and the exophthalmos was treated by keeping a large orbit and performing a partial tumor resection.

Preservation of a functional eye is of capital importance, except in severe orbitotemporal deformities requiring radical dissection. In these cases, the adhesions between the brain and periorbita necessitate sectioning of the optic nerve. However, the eye is kept in place to serve as a support for the future prosthesis (Fig 6).

Surgical attitude toward the eyeball is variable. Regardless of its functional value, preservation of its anatomic integrity is advisable, because this will allow some orbital volume to be conserved, thus facilitating prosthesis adaptation. We feel that only those eyes that are nonfunctional and infiltrated by the tumor should be enucleated.

Resection of an orbital tumor behind a functional eye is controversial. In addition to the fact that it is difficult to individualize, it usually involves the whole of the orbital contents. In such cases, the risk of visual sequelae is very high. We feel it is wiser to correct the exophthalmos and pulsation with minimal dissection of the brain from the periorbita.

When faced with a tumoral exophthalmos that threatens the eyeball, we think it is safer to increase the orbital volume by orbital osteotomies [7, 11] to reduce the exophthalmos and save the eye.

The ancillary procedures, mask lift and facial aesthetic sculpturing, were performed in almost all patients 1 or 2 years after the primary surgery. These procedures are usually carried out to embellish and harmonize the face of patients who have a sad, sagging look [12].

We have to keep in mind that these ameliorations can be temporary, because we are faced with two problems: tumor recurrence and bone resorption, especially in the posterior wall of the orbit. Consequently, the brain may herniate again, causing the recurrence of the exophthalmos.

Correction of ptosis can be done primarily if it is the only problem. It is better to delay its correction if there are other procedures to be done. Palpebral adjustments will be more precisely carried out after the treatment of orbital abnormalities or the fitting of a prosthesis.

Conclusion

Every patient with soft-tissue or skeletal involvement requires individual consideration. The surgeon and patient must accept the fact that several operations may be necessary and that the disease cannot be completely eradicated by surgery. Successful treatment by surgical means requires a complete evaluation to determine what lesions exist and their extension. Partial excisions are the procedure of choice, while radical excisions have specific indications. Everything must be done to preserve the eyeball, since this will help in maintaining orbital volume and a better-fitting prosthesis. An improved cosmetic appearance can be obtained by ancillary procedures in addition to the classical ones.

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