Bony Hamartoma of the Inferior Orbital Rim in a Patient With Tuberous Sclerosis

Named by Bourneville, one of its earliest discoverers, tuberous sclerosis is a heritable neurocutaneous syndrome that is classically manifested by the Vogt triad of mental deficiency, epilepsy, and adenoma sebaceum.1 Despite the classic triad of findings, tuberous sclerosis is a protean disorder characterized by the presence of hamartomas (benign neoplasms composed of cellular elements normally present in tissue) in multiple organ systems, including the brain, kidneys, heart, spleen, lungs, and eye.2,3 Osseous involvement is known to occur in the skull, long bones, pelvis, and metacarpal and metatarsal bones. We report a case of tuberous sclerosis that manifested as a bony mass arising from the inferior (maxillary bone) orbital rim. Histopathologic examination of the excised lesion revealed a hamartoma composed of mature bone. Based on our MEDLINE review of the English-language literature, this is the first reported case of a bony hamartoma arising from this location of the facial skeleton in a patient with tuberous sclerosis.

Report of Case. A 6-year-old girl with known tuberous sclerosis had a mass involving the right inferior orbital rim. The lesion was first noticed by her parents at 1 year of age, and it slowly enlarged during the next 5 years. The patient was diagnosed as having tuberous sclerosis at the age of 6 months based on the characteristic symptoms and signs of a seizure disorder, developmental delay, and classic skin lesions, including adenoma sebaceum along the bridge of the nose and an ash leaf spot on the back of the right calf. There was no other significant medical or family history. The patient’s fraternal twin was unaffected. Although the mass did not cause any functional symptoms, the patient’s parents were concerned about the progressive increase in size and the obvious facial deformity.

On initial examination, a 1.5 × 1.0-cm firm, nontender, immobile mass was identified along the right inferior orbital rim and cheek area (Figure 1). In addition, a 1.0 × 0.5-cm hyperpigmented, slightly elevated skin lesion was noted overlying the inferior orbital rim mass. There were also several small skin lesions involving the midface and nose, which appeared to be adenoma sebaceum, and an ash leaf

Figure 1. Patient with a large elevated mass along the right inferior orbital rim and cheek area measuring 1.5 × 1.0 cm.
spot on the back of the right calf. Visual acuity without correction was 20/25 OD and 20/30 OS. The remainder of the ophthalmic examination findings were unremarkable. A computed tomographic scan of the head and orbits showed a bony lesion arising from the upper portion of the right maxillary bone just below the orbital rim (Figure 2). The lesion was sessile in appearance and measured 1.0 cm along the base and approximately 1.5 cm in the anteroposterior projection. There was no evidence of bony erosion or excavation, and the mass was described as a bony exostosis. Additional radiologic findings included a dense 4-mm calcification in the subependymal space lining the left frontal horn, as well as multiple areas of low-density attenuation consistent with cortical tubers within the cortex of both cerebral hemispheres. The right maxillary sinus was noted to be nearly completely filled with what appeared to be a retention cyst associated with chronic sinusitis. Because of the prominent location of the lesion and associated facial distortion, the decision was made to proceed with surgical excision of the bony mass and the overlying skin lesion.

The overlying skin lesion was removed first via an elliptical skin excision. Dissection then proceeded through the layer of orbicularis muscle, and the bony lesion was identified (Figure 3). The lesion was whitish and firm and had a bony consistency on palpation. The periosteum surrounding the mass, which was normal in appearance, was incised and reflected. An angled oscillating saw was used to remove the lesion flush with the anterior plane of the maxilla, and a 5-mm cutting burr was used to smooth the edges. In the same setting, an ear, nose, and throat specialist performed a right endoscopic maxillary antrostomy for treatment of the right-sided chronic maxillary sinusitis noted on computed tomography.

The bony lesion consisted of medullary bone surrounded by dense fibrosis. Numerous interconnecting bony trabeculae were present within normal bone marrow. Osteoblasts were present on the surface of the trabeculae (Figure 4).
The skin lesion showed a localized area of fibrosis and vascular proliferation consistent with an angiofibroma.

Comment. Tuberous sclerosis, otherwise known as Bourneville disease, is an autosomal dominant syndrome with an incidence of 1 case per 6000 to 29900 people per year.4 Despite its capability of following classic Mendelian patterns, approximately two thirds of cases are sporadic and seem to arise from new somatic mutations. Genetic research has implicated mutations at 2 gene loci, 9q34 (TSC1) and 16p13 (TSC2), in the pathogenesis of tuberous sclerosis.5 These genes are thought to be tumor suppressors. Historically, the clinical criteria for diagnosing tuberous sclerosis include facial adenoma sebaceum, mental deficiency, seizures, and retinal astrocytomas (hamartomas), although not all of the criteria need to be met to make the diagnosis.2,3 In addition to skin and retina, hamartomas have been identified throughout the body in locations such as the iris, brain, bones, kidneys, lungs, heart, and tongue.2,3,6,7

Bony abnormalities in patients with tuberous sclerosis have been previously described in the skull, long bones, pelvis, and metacarpal and metatarsal bones, with the hands and feet the most commonly involved. These lesions can be local or diffuse; tend to be round, oval, or flame shaped; and range in size from a few millimeters to several centimeters. These bony lesions typically manifest as small cortical cysts or areas of dense periosteal sclerosis, the result of fibrous replacement of the normal trabecular bone pattern, and can produce an undulation in the bony contour.2,8,9 Bony abnormalities can be found in the cranium in up to 45% of patients with tuberous sclerosis.2 Radiographs of the skull may reveal sclerotic areas of dense bone formation, often due to hyperostosis of the diploe trabecular of 1 or both tables of the cranial vault. The calvaria is thinned, marrow spaces are replaced by fat, and the periosteum is thickened.2,8,9

After an extensive review of the peer-reviewed English-language literature using MEDLINE, we found only 1 report describing an osseous abnormality of the facial skeleton and orbit in a patient with tuberous sclerosis and none with histopathologic confirmation of a bony hamartoma in this location. Breningstall et al10 reported a case of a sclerotic lesion of the sphenoid bone in a 9-year-old girl with tuberous sclerosis who had left-sided proptosis. Skull radiographs and computed tomography revealed marked sclerosis and thickening of the left wing of the sphenoid bone, which was attributed to fibrous dysplasia. However, no histopathologic analysis was provided.

To our knowledge, this case of a large bony hamartoma arising from the inferior orbital rim in a patient with tuberous sclerosis provides the first clinicopathologic confirmation of a bony hamartomatous lesion involving the facial skeleton in the setting of tuberous sclerosis. Physicians treating patients with tuberous sclerosis should be aware of this presentation.

Ari Abel, MD
David T. Brockbank, MS
Martha Farber, MD
Dale R. Meyer, MD
Albany, NY

The authors have no relevant financial interest in this article.

Corresponding author: Dale R. Meyer, MD, Lions Eye Institute of Albany Medical Center, 35 Hackett Blvd, Albany, NY 12208.


Total Opacification of Intraocular Lens Implant After Uncomplicated Cataract Surgery: A Case Series

Opacification of intraocular lenses (IOLs) in various forms has been reported in all 4 of the varieties of lens materials available. Herein we report total opacification (optic and haptic) of a single-piece acrylic hydrophilic IOL in 5 cases, where the AquaSense IOL (Ophthamal Innovations, Inc, Ontario, Calif) was used. In 2 of the cases, the initial diagnosis was posterior capsular opacity, and in 1 patient who had diabetes it was also thought to be nonresolving vitreous hemorrhage.

Report of Cases. Case 1. A 76-year-old man had uneventful left eye phacoemulsification with posterior chamber IOL implantation in November 2000. Best-corrected visual acuity (BCVA) preoperatively was 6/18 OS. Postoperative BCVA in January 2001 was 6/9 OS with a quiet eye. The patient was discharged.

This patient was referred again 16 months later in March 2002 with blurred vision of about 6 weeks’ duration. The BCVA was 6/18. Anterior and posterior capsular opacification was diagnosed. The patient underwent Yag laser capsulotomy in July with little success. In August 2002, the patient’s condition was reevaluated by the ophthalmic consultant with a view to repeating the Yag laser treatment when opacification of the IOL was suspected. The patient was scheduled for IOL exchange.

This patient underwent successful IOL exchange with anterior chamber IOL and anterior vitrectomy in October 2002. The IOL was fibrosed to the capsular bag and hence had to be removed en masse (Figure 1). On his last visit in late October, BCVA was 6/18 OS with a quiet eye. The patient was pleased with the results and discharged from our care.