CT of Infantile Myofibromatosis of the Orbit with Intracranial Involvement: A Case Report

Curtis C. Stautz

Infantile myofibromatosis is the most common fibrous tumor of infancy. The prognosis depends on the number and location of the lesions. An unusual case report and short review of the literature are presented.

Case Report

An infant boy presented at birth with a large mass protruding from the left orbit that significantly displaced the left globe laterally and inferiorly. Biopsy of the mass was performed the next day, yielding the diagnosis of infantile myofibromatosis. Therapy was considered but the parents opted for no treatment, although they agreed to yearly CT follow-up examinations.

The CT study at the time of birth showed an enhancing 5.2 x 5.4 cm mass centered over the left orbit (Fig. 1A). The left globe was deformed and displaced inferiorly and laterally by the mass. The mass entered the ethmoid region through the lamina papyracea, the sellar region through the optic foramen, and the middle cranial fossa through the greater wing of the sphenoid. The mass also showed intracranial extension into the suprasellar region and anterior cranial fossa (Fig. 1B).

CT examinations performed at yearly intervals showed significant regression of the mass. The study at the age of 2 years revealed residual enhancing tumor surrounding the left globe but predominantly contained within the left orbit (Fig. 2). The globe had regained a normal position. By the age of 4 years the tumor showed minimal remaining mass with only a thin rim of enhancement (Fig. 3). Review on bone windows revealed interval regrowth of a portion of the posterior and medial orbital walls with residual defects in the greater sphenoid wing and ethmoid. Sclerosis and cortical thickening of the ethmoid had an appearance mimicking meningioma.

Discussion

Infantile myofibromatosis, first described by Stout in 1954, is a relatively rare entity yet represents the most common fibrous tumor of infancy. Since its description, infantile myofibromatosis has progressed through various stages and categorizations. Its origin remains unknown and the possibility of genetic transmission continues to be investigated. This benign tumor is best classified by the number of lesions and by the presence or absence of visceral involvement [1].

Approximately half of the patients will have solitary lesions involving the skin, muscle, or subcutaneous tissue [1, 2]; 25% of these children will present after the neonatal period [1]. The lesions in these patients are usually detected incidently by palpation or through visible swelling. There are few symptoms except occasional pain due to nerve compression.

The other half of the patients will present with multiple lesions numbering 2-100, which are again most common in the subcutaneous tissues. Other sites of involvement include bone, lung, myocardium, and the gastrointestinal tract [3]. Renal and CNS lesions have been reported; however, lymph nodes have never been found to be involved. These lesions are also often asymptomatic. When symptoms are present, they are usually pulmonary in nature or secondary to obstruction of the gastrointestinal, renal, or cardiovascular systems.

The diagnosis of infantile myofibromatosis is made by biopsy of the firm, rubbery, usually superficial lesion with histologic evidence of a distinct zoning pattern and staining characteristics of myoblastic and fibroblastic cells [2].

The prognosis is related to the number of lesions and the presence or absence of visceral involvement [4]. The patients with single lesions show a uniformly benign and self-limited course with spontaneous regression of the lesion being the norm. Biopsy of regressing lesions has shown areas of necrosis and calcification. Tumor necrosis can be so severe that only a thin rim of viable tissue remains.

As in our case, a small number will have their lesion in a critical location, causing local symptoms or significant deformity. Some patients will require surgical intervention under these circumstances, and subsequently 7% will have local recurrence [5].

Of the patients with multiple lesions, two-thirds will have their lesions confined to bone and soft tissues, and these patients will usually do well. Multicentric lesions that involve the viscera herald a 75% mortality. Death is usually caused by cardiopulmonary or gastrointestinal complications and often occurs shortly after birth.

Our case is unique in that few lesions are as large at presentation, reside in such a critical location, or provide such close follow-up without medical intervention. This case did
Fig. 1.—A, Axial contrast-enhanced CT scan on day of birth at level of orbit reveals large, enhancing soft-tissue mass with marked displacement of left globe. B, Axial contrast-enhanced CT scan on same day at a slightly higher level reveals intracranial extension.

Fig. 2.—Enhanced CT study at 2 years of age reveals regression of the tumor. There is residual tumoral enhancement surrounding left globe, which has returned to a normal location.

Fig. 3.—Enhanced CT study at 4 years of age shows little evidence of tumor.

show the expected regression of the imposing tumor mass present at the time of birth. Residual porencephaly and focal atrophy is all that remained in the region of previous tumoral involvement. The globe has returned to a normal position and configuration. The regularly decreasing tumor now shows only a thin rim of enhancement within the orbit. This is the expected course of events for a single lesion, even with presentation as large and in such a critical location as in our case.

Little has been described previously regarding enhancement of the tumor with contrast material. This lesion enhanced markedly with contrast both at the time of presentation and during regression of the lesion. This would suggest that the use of contrast material with MR imaging might provide improved lesion detection. MR imaging is already being used with success in the detection and evaluation of infantile myofibromatosis, although without the use of contrast enhancement [1].

The literature is clear and this case is a striking example of how single lesions can be followed expectantly. This would seem to be best accomplished with contrast-enhanced CT or MR imaging. There are few other cases in which a tumor can be followed with as much confidence in both expected regression and ultimate favorable outcome as is possible in this disease process.

REFERENCES