Intraocular Choristoma, Anterior Staphyloma With Ipsilateral Nevus Sebaceus, and Congenital Giant Hairy Nevus: A Case Report

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A 5-year-old girl presented with choristoma of the eye along with nevus sebaceus and congenital giant hairy nevus over the face. Anterior staphyloma also was present. Although choristomas have been seen occasionally occurring with nevus sebaceus, an associated ipsilateral, regional, congenital giant hairy nevus is rare.

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The eye and skin share a common embryologic origin and are derived from both surface ectoderm and mesoderm primordial sources. Choristoma is a rare, benign, ectopic mass of tissue that is histologically normal for an organ or part of the body other than the site at which it is located.[1] Nevus sebaceus is a verrucous hamartoma of predominantly sebaceous origin presenting as yellowish orange, hairless, plaquelike lesions, usually present at birth and most commonly occurring in the scalp or midfacial area.[2] Giant hairy nevus is a congenital nevus of nevocytic cell origin that is mostly seen over the trunk; it may undergo malignant transformation.[3] Staphyloma is a localized protrusion of any part of the globe of the eye. Staphylomas may be anterior, posterior, or equatorial. Choristoma of the middle ear and eye have been reported.[1,4-6] Salivary gland choristoma (heterotopic salivary gland tissue) is a rare condition that occurs at various locations within the head and neck. Choristomas of the eye, though rare, constitute approximately 33% of all epibulbar tumors in children.[7]

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The authors report no conflict of interest.

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Case Report

A 5-year-old girl demonstrated blackish discoloration over the right half of the face; an asymptomatic, flesh-colored, verrucous bald patch over the scalp; complete loss of vision; and a painful mass in the right eye, all present since birth. The hyperpigmented patch over the right side of the face gradually increased in size and darkened with hair growth. There was no history of pain and redness over the lesion.

The patient's father revealed a history of convulsions/epileptic seizures on the right side of the body with unconsciousness 2 years prior, which was treated by a local doctor. The child was a full-term pregnancy without any complications. She has 2 brothers, both in good health.

The general examination showed the patient as conscious, oriented, and afebrile. The pulse rate was 90 beats per minute, which was within reference range. There was no pallor, cyanosis, clubbing, edema, or regional lymphadenopathy. Systemic examination including neurologic, cardiac, pulmonary, and abdominal examination revealed normal findings. Ultrasonography of the abdomen was normal. Radiograph of the chest was normal; however, radiograph of both the lower limbs showed osteopenia, widening of metaphyses, fraying, and capping of metaphyses as seen in rickets.

Local Examination—A hyperpigmented blackish patch was observed. It involved the right side of the face over the cheek and upper eyelid and slightly crossed over the nose and upper lip area. It measured 8.5 cm in length from the right eyebrow to the angle

of the mouth and 7.5 cm from the middle of the nose to the right ear (Figure 1). Hair was present over the hyperpigmented patch, more on the cheek and upper lip. Hypertrichosis of the right eyebrow also was present. A small nodule was present over the lateral side of the right eye.

The 2 lesions over the right lateral aspect of the scalp were flesh colored, hyperkeratotic, and verrucous, and measured 6 cm and 4 cm in length (Figure 2). Alopecia was present over the lesions; the rest of the skin and mucosa was normal.

Both upper and lower eyelids were hyperpigmented with hypertrichosis. A nodular mass, which was 3 mm in size, firm to hard in consistency, and nontender, arose from the lateral canthus. There was no perception of light in the right eye, while vision in the left eye was normal.

Right eye examination findings revealed mild proptosis. The conjunctiva was congested and hypertrophied. The cornea was not visible and an ulcerated mass was present in the corneal area. The sclera, cornea, lens, anterior chamber, and other details could not be made out due to a mass present in the corneal area (Figure 1). The fundus details could not be seen due to total opacity of media. Ophthalmic sonography showed ectasia of the outer coat and the anterior chamber of the right eye was occupied by echogenic iris tissue, thus producing features of anterior staphyloma. The lens and the size of the eyeball were normal. No retinal detachment was observed.

Enucleation of the right eye with removal of the epicanthal nodule and skin biopsy was done; the tissues were submitted for histopathologic examination.

Histopathologic Examination—Results of the biopsy from the skin of the face showed nests of nevus cells containing plenty of melanin pigment, mainly in the upper papillary dermis. Junctional activity was observed at few places. Plenty of hair follicles were present in the dermis. Collagenization of subepithelial tissue was seen.

A section from the tissue of the nodule at the lateral canthus showed features of junction nevus.

A histopathology section from the anterior portion of the eyeball showed increased melanocytes and pigmentation of the conjunctiva, sclera, and uveal tract. A section from the staphyloma portion showed presence of sebaceous glands and sweat glands. Histopathology of the eyeball showed presence of cartilage along with adipose and neural tissue. The cartilage was present in the whole cornea above the Descemet membrane and extended into the sclera on both sides just beyond the ciliary processes. A diagnosis of complex choristoma was made.

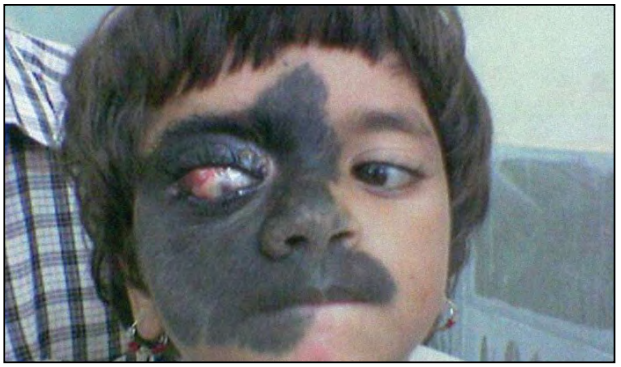

Figure 1. Large melanocytic nevus over the face and eye lesion.

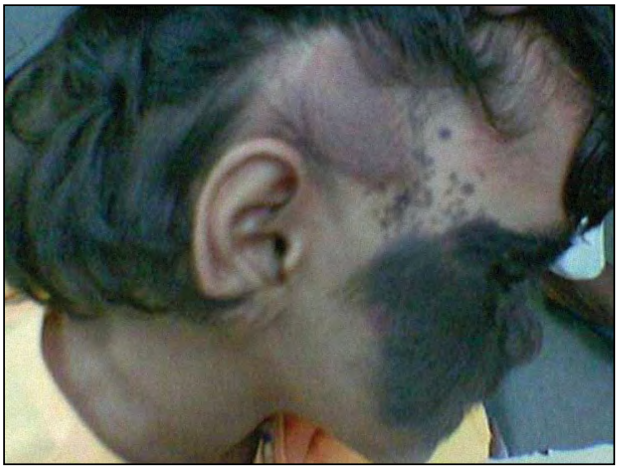

Figure 2. Ipsilateral nevus sebaceus and melanocytic nevus.

A histopathology section from the skin of the scalp showed papillomatosis of the epidermis, few sebaceous glands in the upper dermis, and lack of mature hair follicles (Figure 3).

A final diagnosis of nevus sebaceus with congenital giant hairy nevus of the face as well as anterior staphyloma with complex choristoma of the eye was made.

Comment

Choristomas of the eye have been reported in the orbit, cornea, conjunctiva, uvea, retina, and optic nerve. Many reports have described the choristoma mainly in the epibulbar area,[7,8] but little is known of the choristoma that occurs intraocularly.[5] Osseous choristoma, one subtype of this entity that occurs within the orbit, has rarely been reported in the world literature.[6] Phacomatous choristoma is a rare congenital hamartoma of lens tissue.[9] It presents in newborns or young infants as a subcutaneous mass in the medial lower eyelid. A case of encephalocraniocutaneous lipomatosis, a congenital neurocutaneous syndrome characterized by lipomatous craniofacial hamartomas, has been reported.[10]

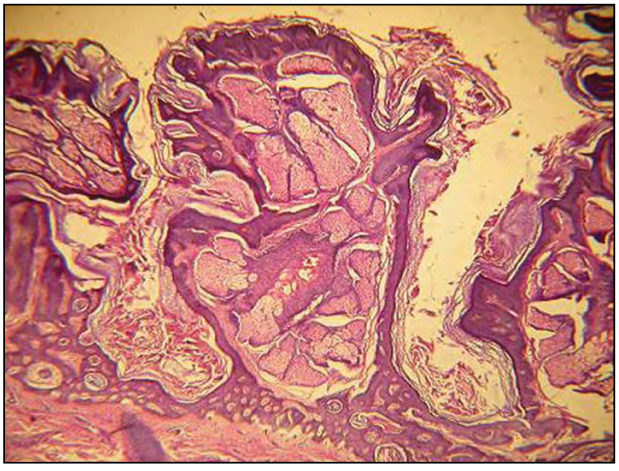

Figure 3. Results of a biopsy from the skin of the scalp showed papillomatosis and hyperkeratosis of the epidermis along with presence of excessive sebaceous glands in the upper dermis (H&E, original magnification ×400).

The most common ocular manifestation is epibulbar choristoma, but additional eye anomalies may be present. Anterior scleral staphyloma has been reported to be associated with neurofibromatosis.[11]

Linear epidermal nevus syndrome and linear nevus sebaceus syndrome are rare neurocutaneous syndromes characterized by epidermal nevi, epilepsy, and mental retardation. Ocular abnormalities described in these patients are microphthalmia, colobomas of the eyelids, large optic nerve heads, and congenital teratomas or dermoids of the conjunctiva and cornea. Although rare, complex limbal choristomas can occur in the setting of linear nevus sebaceus syndrome and can be associated with multiple ocular and systemic abnormalities. Linear nevus sebaceus syndrome has been observed with the ectopic lacrimal gland tissue at the limbus, which further confirmed the association of complex limbal choristomas in linear nevus sebaceus syndrome.[12] Choristoma has been reported to rarely occur in association with staphyloma.[13]

Nevus sebaceus occurs in 0.3% of newborns.[14] Hyperpigmentation, hyperkeratosis, and neurologic complications such as seizures and mental deficiency may be present with nevus sebaceus. Other associated disorders may include esotropia, conjunctival lipodermoid lesions, corneal opacity, colobomas of the eyelids and choroid, hydrocephalus, cerebral cortical lesions, alopecia, coarctation of the aorta, ventricular septal defects, and nephroblastomas. Nevus sebaceus rarely occurs as part of a syndrome consisting of central nervous system and ophthalmologic abnormalities. Complex choristomas rarely have been observed in association with nevus sebaceus.[15] Although our patient experienced epileptic seizures 2 years prior, there was no mental retardation.

Wilkes et al[16] reported ocular malformation in the form of corneoscleral limbal mass in association with ipsilateral facial nevus sebaceus, but congenital giant hairy nevus of the face with ipsilateral nevus sebaceus with choristoma and anterior staphyloma of the eye is rare.

We believe a defect occurred at the time of tissue formation during the development and evolution of the fetus, which resulted in the clinical findings reported in this interesting case.

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