Bilateral Cataracts in Tuberous Sclerosis

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An 11-year-old girl and a known case of tuberous sclerosis was referred for ophthalmic evaluation. The parents had noted multiple pigmented lesions on her face, scalp and neck since the age of 3 years. There were no neurological symptoms at the time of presentation. General physical examination revealed adenoma sebaceum on her face (Figure 1A), neck (Figure 1C) and scalp; a shagreen patch was detected on the left arm and leg (Figure 1B). Ash leaf spots were present over her left hip and leg, and subungual fibromas were also present (Figure 1D) on her finger and toe nail beds. Coronal T2 weighted magnetic resonance image of the brain showed cortical tubers and subependymal nodules (Figure 1E). Ophthalmic examination revealed unaided visual acuity of 20/20 in her right eye and 20/25 in the left one which improved to 20/20 with -0.5D correction. Fundus examination showed astrocytic hamartoma with calcification located adjacent to the optic disc in the right eye (Figure 2A). Fundus examination in her left eye showed a smooth semi-transparent hamartoma inferior to the optic disc (Figure 2B). In addition, the peripheral fundus in both eyes had retinal hamartomas (Figures 2A, 2B). Bilateral punctate cataracts were detected on slit lamp biomicroscopy (Figures 2C, 2D).

DISCUSSION

Tuberous sclerosis was described by Bourneville in 1880.[1] The condition has autosomal dominant inheritance but the majority of cases occur as new mutations. It involves multiple systems in the body including the skin, central nervous system, eyes, heart and kidneys. Diagnosis is dependent on careful clinical observation. Signs may be manifest at birth, but the diagnosis is usually made in the first or second decades of life. The classic clinical manifestations of epilepsy, adenoma sebaceum and mental deficiency may occur only in 30% of patients. The "ash leaf" sign, a hypopigmented skin spot seen under ultraviolet light, is believed to be pathognomonic and may occur in 80% to 90% of patients. Intracranial calcified hamartomas seen radiologically are diagnostic and 60% of these patients exhibit mental deficiency. A wide range of other lesions occurs including subungual fibromas, cardiac rhabdomyomas, and hamartomas of the kidney, liver, thyroid, and gastrointestinal tract.[1,2]

Significant ocular findings occur in 66% of patients. The classic ocular lesion is a peripapillary astrocytic hamartoma or "mulberry lesion" which may be single or multiple. These may undergo cystic degeneration and rarely mimic necrotizing retinochoroiditis. Loss of vision is unusual. Strabismus, hypopigmented iris and fundus lesions, retinal angiomas and poliosis have also been reported. In a study on 100 patients with tuberous sclerosis, Rowley et al found a high prevalence of retinal hamartomas (44%). Punched out areas of retinal depigmentation (39%), myopia (27%), hyperopia (22%), astigmatism (27%), eyelid angiofibromas (39%), non-paralytic strabismus (5%), and colobomas (3%) were also noted.[2]

In addition to retinal lesions in patients with tuberous sclerosis, unusual findings such as band keratopathy and keratoconus have been reported.[3] Cataracts associated with tuberous sclerosis have been described in very few case series.[3-4] In our case, age of the patient, cataract morphology and bilaterality of the lenticular opacities were compatible with a diagnosis of congenital cataracts. Although the occurrence

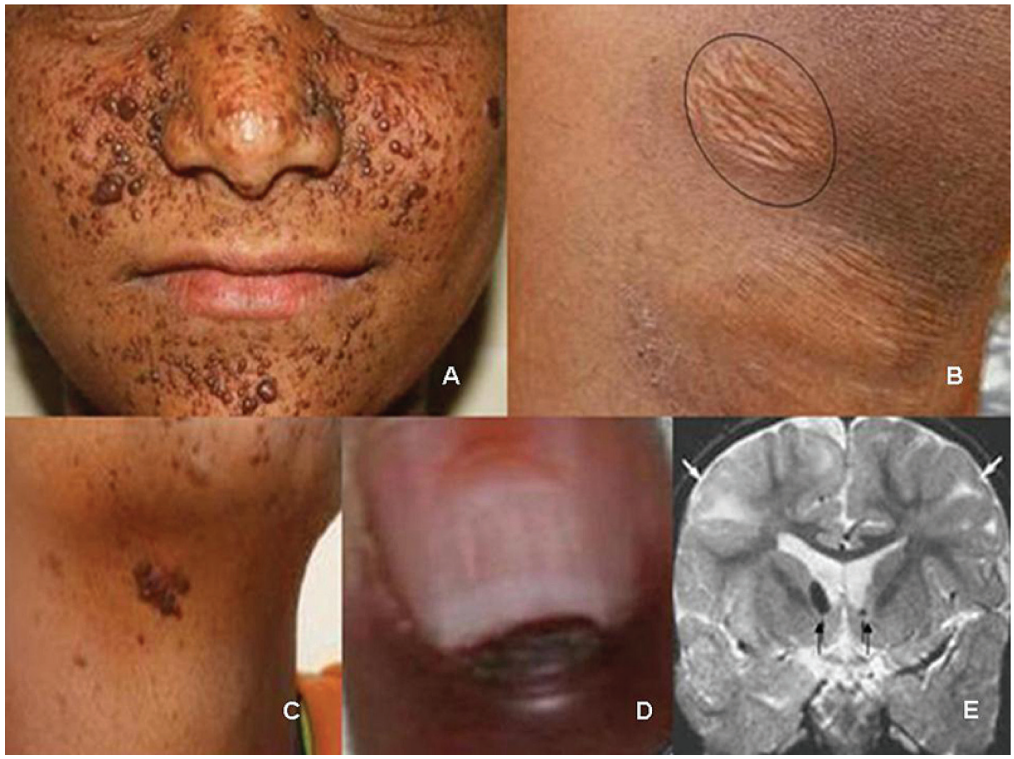

Figure 1. Clinical photographs of the patient show multiple angiofibromas of the face (A) and neck (C), a shagreen patch over her leg (1B, oval), and subungual fibroma (1D). Coronal T2-weighted magnetic resonance image shows cortical tubers (white arrows) and subependymal nodules (black arrows).

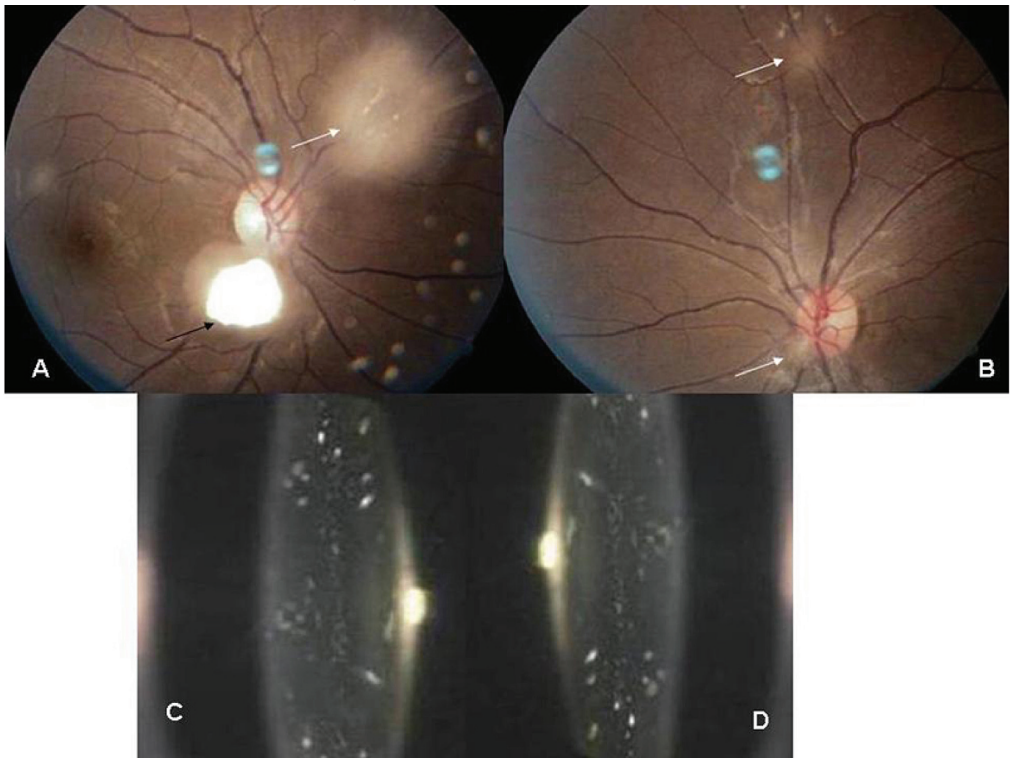

Figure 2. Fundus photographs demonstrate retinal astrocytoma ("mulberry lesion") with calcification adjacent to the optic disc (2A, black arrow) and semitransparent, oval lesions in the midperipheral fundus (2A, white arrow); similar lesions were seen in the left eye (2B, white arrows). Bilateral punctate cataracts were observed on slit lamp examination (2C, 2D).

of cataract may be coincidental, we recommend that young patients with tuberous sclerosis be screened for lenticular opacities and followed for the development of visually significant cataracts.

Conflicts of Interest

None.

REFERENCES

1. Bourneville DM. Sclerose tubereuse der circonvolutions cerebrales: idiotie et epilepsie hemiplegique. *Arch Neurol (Paris)* 1880;1:81–91.
2. Rowley SA, O'Callaghan FJ, Osborne JP. Ophthalmic manifestations of tuberous sclerosis: a population based study. *Br J Ophthalmol* 2001;85:420-423.
3. Ross AT, Dickerson WW. Tuberous sclerosis. *Arch Neurol Psychiatry* 1943;50:233-257.
4. Warner JE, Lessell S. Cataract in tuberous sclerosis. *J Neuroophthalmol* 1997;17:33-35.