Net Letter

Delleman syndrome: Report of a case in an adolescent boy

Sir,

Delleman syndrome (DS) or oculocerebrocutaneous syndrome is a rare condition first described by Delleman and Oorthuys in 1981.[1] Since then, approximately 35 cases have been reported in the literature, mainly in ophthalmological journals and Indian Pediatric and Ophthalmologic journals.[1,2] The cutaneous hallmarks of DS, skin tags and alopecic patches among others are frequently seen in our everyday practice, which account for the unawareness in the diagnosis by general dermatologists.

A 14-year-old male was referred to the Dermatology Department with lesions in his left upper eyelid. His personal medical history was unremarkable, but his mother suffered from rubella during the second trimester of the pregnancy.

Physical examination revealed a flesh-colored, asymptomatic papule of 3 mm in the nasal aspect of the left upper eyelid, consistent with a skin tag, and a small coloboma at the inner third of the same eyelid. An alopecic frontal plaque with frontal bossing could also be seen [Figure 1].

A dermoid cyst measuring 5 mm was found over the iris and pupil, resulting in a sclerocornea and blindness of the left eye [Figure 2]. Another blue-gray epibulbar cyst was found in the right eye [Figure 3]. The cystic nature of both the lesions was confirmed by ultrasound biomicroscopy and optical coherent tomography (AS-OCT Visante, Carl Zeiss Meditec Inc, Dublin, CA, USA.) Neurological examination and the laboratory work-up including TORCH profile were normal.

Central nervous system (CNS) computed tomography revealed asymmetric dilated ventricles and two large arachnoid cysts, one located in the parietal area and the other causing thinning of the inner table of the frontal bone [Figure 4].

DS was diagnosed and the patient continued his follow-up with the Neurology and the Ophthalmology Departments.

DS is a rare genetic disorder characterized by ocular, cutaneous and CNS abnormalities.[1] A wide phenotypical variability can be seen. Neurological abnormalities include ventricular and posterior fossa malformations, agenesis of corpus callosum, arachnoid cysts and hydrocephalus. Patients can present with seizures, mental retardation and developmental delay or electroencephalographic abnormalities, all of which account as prognostic factors for this syndrome. Such

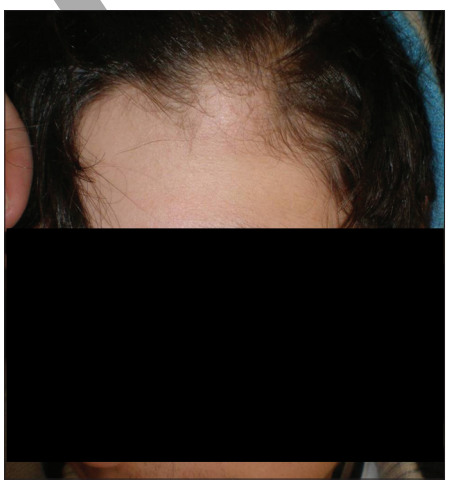

Figure 1: Frontal alopecic area with frontal bossing

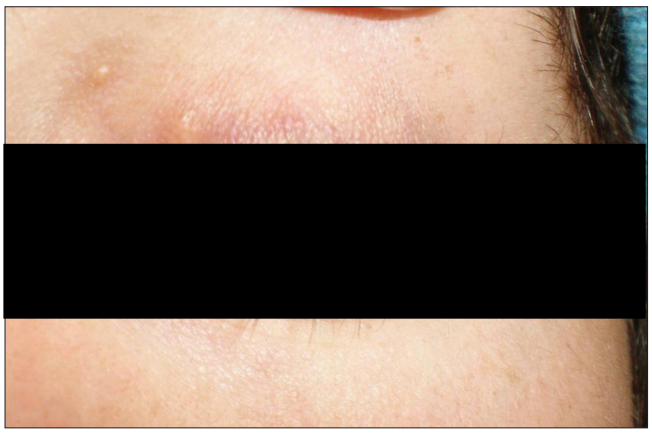

Figure 2: Small palpebral coloboma with a flesh-colored skin tag adjacent to it. Dermoid cyst over the iris impairing the patient´s vision

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