Bilateral macular colobomas in Leber’s congenital amaurosis

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Accepted 18 April 1989

Key words: electroretinogram, Leber’s congenital amaurosis, macular coloboma, pattern VECP, sibling

Abstract. Two siblings with Leber’s congenital amaurosis had bilateral macular colobomas, nystagmus, extinguished ERGs, and degenerative salt and pepper like changes in the fundus. They had non-recordable or non-meaningful visually evoked cortical potentials in response to both flash and pattern stimuli. The ophthalmic conditions were thought to be inherited as an autosomal recessive trait.

Abbreviations: ERG: electroretinogram; VECP: visually evoked cortical potential

Introduction

Leber’s congenital amaurosis was first described as a congenital blindness or profound visual disorder demonstrated in the first year of life associated with wandering nystagmus, absent or reduced pupillary reaction, and variable ophthalmoscopic changes [1]. After the report by Franceschetti et al. [2], characterizing the electroretinographic response (ERG) in Leber’s congenital amaurosis as being extinguished or markedly reduced, the ERG became very useful in the early diagnosis of this disease. Frequently associated findings with the disease were keratoconus, keratoglobus, cataract, neurologic deficits, skeletal abnormality, and renal dysplasia [3–9]. The association of macular coloboma and Leber’s congenital amaurosis was rare. We describe two affected siblings with macular colobomas, and similar ophthalmologic conditions, probably due to an autosomal recessive trait.
Fig. 1. Case 1: *Top*, The peripheral retina of the right eye is sharply demarcated from the posterior pole area. *Middle*, Note narrowed retinal artery and salt and pepper changes in the right eye. *Bottom*, Pigmented macular coloboma with hyperpigmented rim in the left eye.