FAMILIAL BILATERAL MACULAR COLOBOMA

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Familial congenital bilateral macular coloboma is a rare case. The mechanisms of macular colobma are still controversial. Two mechanisms had been proposed: 1) congenital infection -- congenital toxoplasmosis 2) congenital developmental anomaly – hereditary or sporadic. We reported a sibling —10 year-old sister and 7 year-old younger brother. They presented with nystagmus or exo-deviation. Their fundi all showed oval shape, 4 to 18 disc size, atrophic lesions in bilateral macular area with pigment clumps around their margin. Review the family history, no other family member had the same finding. Toxoplasma IgG antibody was negative. Electroretinogram showed decreased amplitude in both a and b wave. Concluded from above results, we thought the causes of familiar bilateral macular coloboma in this case being due to congenital developmental anomaly.

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