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Case Report



# Congenital Coloboma of the Optic Nervehead

**Hu yuzhang*, Liu pengfei and Yan huajing**

*Chengdu Aidi Eye Hospital, Chengdu city, Sichuan Province, China*

***Corresponding author:** Hu yuzhang, Chengdu Aidi Eye Hospital, Chengdu city, Sichuan Province,45 West 2nd Section, 2nd Ring Road, Chengdu city, China.

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## Case Report

A 12-year-old male patient complained of poor eyesight in both eyes since childhood. Physical examination: VOD: 0.25+0.75DS→0.3; VOS: 0.5-1.0DS→1.0. Intraocular pressure was normal. The cornea, lens and vitreous were transparent. The boundaries of the optic nerve papillae were clear, and the arteries and veins ran out from the edge of the optic nerve papillae. No optic cups were observed, only a huge depression of the same size as the optic papillae were observed, and no normal tissue structure was observed in the optic nerve papillae. In the right eye, there was a myopic arc on the nasal side of the papilla, with serous retinal detachment in the macular area. OCT in the macula of the right eye showed superficial detachment of the retinal neuroepithelial layer in the right eye, with a large amount of gaps in the retinal layers. OCT in the macula of the left eye was normal. B-scan of both eyes showed deep depressions of optic nerve papillae. Visual field examination revealed a slightly enlarged physiological blind spot in the right eye, accompanied by a central dark spot. Visual field of the left eye was basically normal (Figures 1-4).

**Figure 1:** Fundus image: the boundary of the optic papillae in both eyes was clear, and the vessels run out along the edge of the optic papillae. The shape, number,distribution and direction of vessels were normal. There was a serous retinal detachment in the macular area (white arrow).

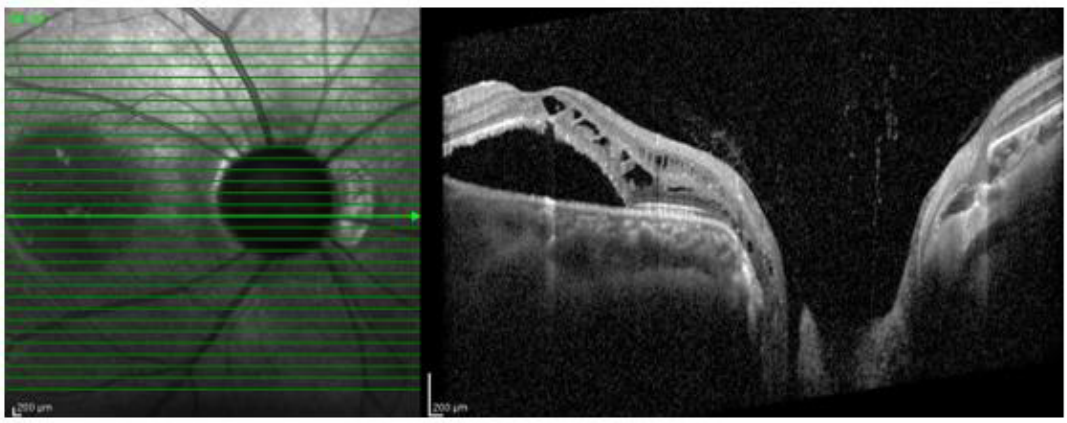

**Figure 2:** OCT: Superficial detachment of the retinal neuroepithelial layer in the macular of the right eye, with a large amount of gaps in the retinal layers.

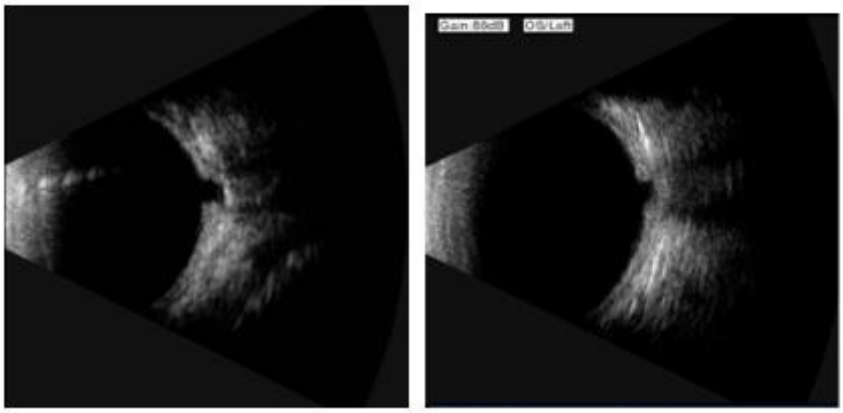

**Figure 3:** B ultrasonography showed clear and deep depressions in the optic papillae of both eyes.

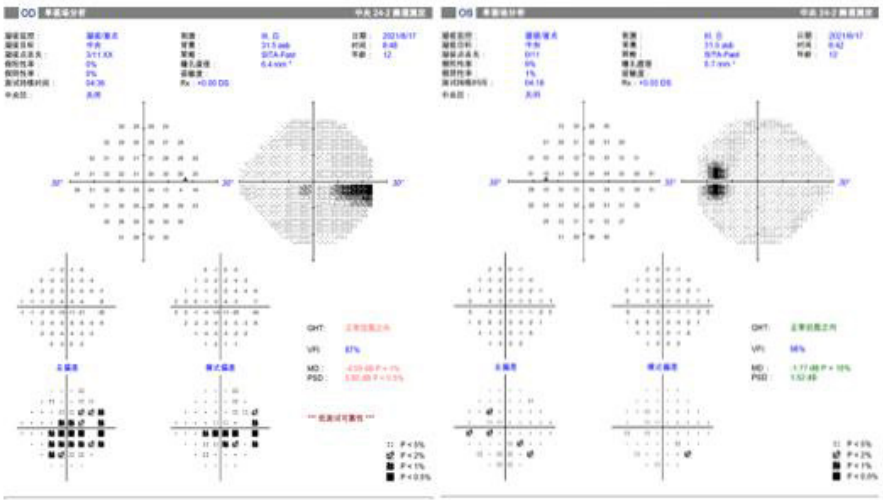

**Figure 4:** Visual field examination revealed a slightly enlarged physiological blind spot in the right eye, accompanied by a central dark spot. Visual field of the left eye was basically normal.

## Discussion

Coloboma of the optic nervehead is a rare, nonprogressive, congenital optic nerve disorder with an incidence of about 0.075%. Congenital defects of the optic nerve and other eye tissues are associated with incomplete or abnormal proximal position of the optic fissure. In a normal eye, the fetus begins to close at 4-5 weeks of development. The defect may be complete or partial. Defects can occur in one or both eyes. It may occur alone or in conjunction with other ocular defects, including lens defects, iris defects and retinal choroid defects. The optic cup is a large and deep depression, irregular funnel-shaped, retinal vessels appear from the edge of the defect. The degree of visual impairment depends on itself or other complications [1-3]. Optic nerve papilla defect is generally divided into two types, one is combined with choroid or other parts of the defect, accounting for most cases; The second is a simple optic nerve papillary defect limited to the optic papilla. The latter is extremely rare, with an incidence of only 0.025% [4,5]. In this case, the defect was limited to the optic papilla, and no obvious abnormality was found in the surrounding retinal tissue, macula and blood vessels. Therefore, the child had better corrected vision. The vision was relatively poor due to serous retinal detachment in the macular area of the right eye. The reason of the detachment may be consistent with the serous detachment of macular from the congenital fovea of the optic papilla.

The disease should be distinguished from chronic simple glaucoma and morning glory syndrome. The former is more common in young patients, and the cup to plate ratio gradually increase, reaching 1.0 in severe cases. The vessels of the optic nerve are seen coming out from the nasal side in a bent fashion. The patient had no abnormal sensation other than occasional discomfort caused by elevated intraocular pressure. However, the visual field appeared centripetal narrowing, gradually developed to tubular visual field defect, and eventually blindness. The patient was 12 years old with normal corneal diameter and normal intraocular pressure. Although the patient has poor vision since childhood, he could move freely. The patient were followed up for more than half a year, and their visual field and intraocular pressure were normal. Therefore, chronic and developmental glaucoma can be ruled out. Morning glory syndrome is also a type of optic papilla defect. However, the abnormal optic papilla is 2-6 times more than the normal ones, There are atrophic rings and pigments at the edge of the optic papilla, and gray and white fibrous tissue hyperplasia in the center of the optic papilla. The blood vessels from the optic papilla can reach more than ten, and it is impossible to distinguish arteries and veins. Coloboma of the optic nervehead belongs to congenital abnormal development and there is no special treatment. Ametropia can be corrected early to prevent amblyopia, and surgery may be required for concurrent retinal detachment. Changes in intraocular pressure and visual field should also be closely monitored. In this study, the patient suffered from serous retinal detachment in the macular area of the right eye. Laser or vitreous surgery may be considered for treatment.

## Acknowledgement

None.

## Conflict of Interest

No Conflict of Interest.

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