

Two Sets of Sisters in Two Families with Bilateral Choroidal Osteoma

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Abstract

Background: To describe the clinical characteristics of two sets of sisters in two families with bilateral choroidal.

Methods: The ocular clinical examinations of all the families were performed.

Results: (1) Thirteen immediate family members from three generations were included and screened in two families. Bilateral choroidal osteoma was found in two sets of two sisters in the second generation of both families. (2) All of the four patients were young women aged 22 years, 26 years, 26 years and 30 years respectively. (3) Choroidal osteoma lesions located at the posterior to the optic disc and showed a relatively large area. Fundus findings, fluorescein angiography and computed tomography scan results showed typical characteristics of choroidal osteoma.

Conclusions: This is the first report of such cases in China. The clinical characteristics of bilateral choroidal osteoma are a larger lesion around the optic disc and early onset. Eyesight of the patient is damaged with the time lapse.

Keywords: Choroidal osteoma; Choroidal tumor; Familial

Introduction

Choroidal osteoma is a rare and benign tumor located in the choroid, which has a basic pathological tissue structure of mature bone tissue. Choroidal osteoma was officially named by Gass in 1978. This disease frequently occurs in young women and is usually unilateral. Its etiology and pathogenesis are unknown [1]. Previous pedigree case reports suggested that choroidal osteoma showed a genetic predisposition and when it ran in families, it was often bilateral, and the disease was observed to span between mother and daughter, father and daughters, siblings and twins [2-6]. But the disease occurred between sisters haven't been reported in China. Our current report demonstrated that both of the families studied had two sisters who developed bilateral choroidal osteoma. We performed clinical examinations of the families, and long-term follow-up observations of the patients. Following the medical history provided by the first diagnosed patient, we examined the bilateral visual function, the anterior segment by slit-lamp biomicroscopy, and the fundus by ophthalmoscopy. In addition, we selectively performed bilateral fundus fluorescein angiography (FFA, Zeiss FF450), and double orbit computed tomography (CT,

Philips Brilliance 16 slice CT). Concurrently, we also performed eye examination on the parents and siblings as well as their children of the patients to screen them for the disease.

Case Reports

Family 1

Seven immediate family members, spanning three generations, were screened for the disease, which revealed bilateral choroidal osteoma in two sisters from the second generation.

The first diagnosed patient was a 26-year-old female with a blurry vision in both of her eyes for five years. Her vision was 0.4 in the right eye and 0.25 in the left eye, and these impairments were uncorrectable. Her intraocular pressure was 13 mmHg in the right eye and 14 mmHg in the left eye. Examination by slit-lamp biomicroscopy revealed that the anterior segments of both eyes were normal. The area around the optic disc and macula in the posterior right fundus exhibited a large area of retinochoroidal atrophy that extended outward to form an annulus-shaped yellow-white flat protrusion with a rough surface and some mild pigment deposition (Figure 1A). The early phase of FFA revealed a high fluorescence staining in a mottled pattern that had already shown an osteoma lesion, and the fluorescence intensity gradually increased and became further enhanced during the late phase of FFA (Figure 1B). Similarly, the area around the optic disc and macula in the posterior left fundus also showed a large area of yellow-white flat protrusion representing the choroidal osteoma lesions (Figure 1C), and also exhibited high fluorescence intensity during the late phase of FFA (Figure 1D). Bilateral orbital CT scanning revealed arc-shaped high density areas in the posterior of both eye rings (Figure 1E).

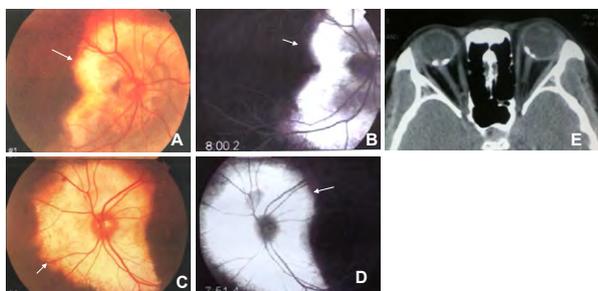


Figure 1: Family 1, the first diagnosed patient was a 26-year-old female. A. Color image of the right fundus revealed the osteoma lesion at the posterior area (arrow). B. Right eye FFA revealed the osteoma lesion, which exhibited a strong fluorescence intensity (arrow). C. Color image of the left fundus revealed the osteoma lesion at the posterior area (arrow). D. Left eye FFA revealed the osteoma lesion, which exhibited a strong fluorescence intensity (arrow). E. Bilateral orbital CT scanning revealed high density areas of osteoma lesion in the posteriors of both eye rings.

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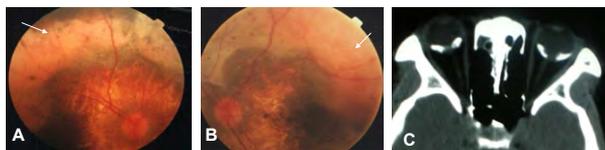


Figure 2: Family 1, the older sister of the patient was 30 years old. A. Color image of the right fundus revealed rear atrophy and the osteoma lesions (arrow). B. Color image of the left fundus revealed rear atrophy and the osteoma lesions (arrow). C. Bilateral orbital CT scanning revealed high density areas of osteoma lesion in the posteriors of both eye rings.

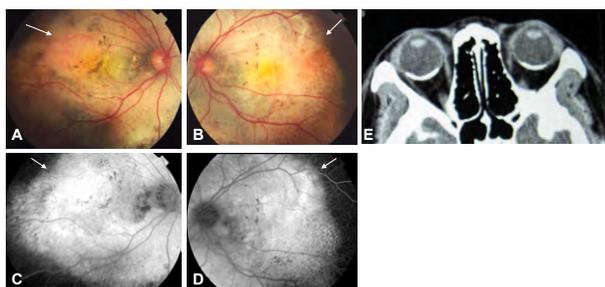


Figure 3: Family 2, the first diagnosed patient was a 22-year-old female. A. Color image of the right fundus revealed the osteoma lesion at the posterior area (arrow). B. Color image of the left fundus revealed the osteoma lesion at the posterior area (arrow). C. Right eye FFA revealed the osteoma lesion, which exhibited a strong fluorescence intensity (arrow). D. Left eye FFA revealed the osteoma lesion, which exhibited a strong fluorescence intensity (arrow). E. Bilateral orbital CT scanning revealed high density areas of osteoma lesion in the posteriors of both eye rings.

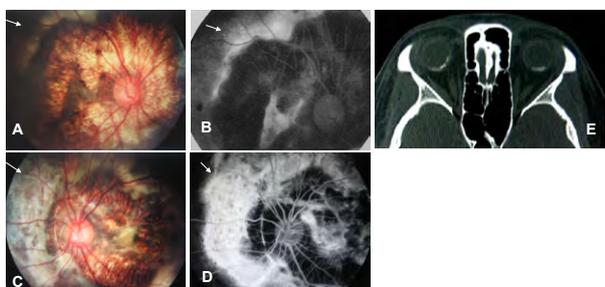


Figure 4: Family 2, the older sister of the patient was 26 years old. A. Color image of the right fundus revealed rear atrophy and the osteoma lesions (arrow). B. Right eye FFA revealed the osteoma lesion, which exhibited a strong fluorescence intensity (arrow). C. Color image of the left fundus revealed the osteoma lesion at the posterior area (arrow). D. Left eye FFA revealed the osteoma lesion, which exhibited a strong fluorescence intensity (arrow). E. Bilateral orbital CT scanning revealed high density areas of osteoma lesion in the posteriors of both eye rings.

The elder sister of the patient was 30 years old and complained of a gradually decreased visual acuity in both eyes for about 12 years. Her vision was 0.08 in the right eye and 0.06 in the left eye, and the impairments were uncorrectable. Her intraocular pressure was 16 mmHg in the right eye and 15 mmHg in the left eye. Examination by slit-lamp biomicroscopy revealed that the anterior segments of both eyes were normal. The areas around the optic disc and macula in both fundi exhibited large areas of retinochoroidal atrophy that extended outward to form annulus-shaped yellow-white flat protrusions with a rough surface and some mild pigment deposition (Figures 2A and 2B). This patient did not have FFA examination because she is allergic to

fluorescein sodium. Bilateral orbital CT scanning revealed arc-shaped high density areas in the posteriors of both eye rings (Figures 2C).

The parent and younger brother of the patients were healthy and had normal visual acuity. Examinations of the anterior segment and fundus did not reveal any abnormalities. The elder sister had a 5-year old son and a 7-year old daughter. The examinations of the anterior segment and fundus of their eyes were all normal. Two patients were followed up for 7 years. Neither the bilateral fundus nor the visual acuity exhibited any significant changes.

Family 2

Six immediate family members, spanning three generations, were screened for the disease, and two sisters from the second generation presented bilateral choroidal osteoma.

The first diagnosed patient was a 22-year-old female with vision in both of her eyes decreased slowly for approximately 7 years. Her vision was 0.04 in the right eye and 0.12 in the left eye. Her intraocular pressure was 12 mmHg in the right eye and 13 mmHg in the left eye. Examination by slit-lamp biomicroscopy revealed that the anterior segments of both eyes were normal. Lesions in the bilateral fundus had surrounded the optic disc and invaded the retinal equator (Figures 3A and 3B). FFA revealed strong fluorescence staining in the lesion areas (Figures 3C and 3D). Bilateral orbital CT scanning revealed arc-shaped high density areas in the posteriors of both eye rings (Figure 3E).

The elder sister of this patient was 26 years old and presented with an approximately 9-year history of the decreased vision. Her vision was 0.04 in the right eye and 0.08 in the left eye. Her intraocular pressure was 16 mmHg in the right eye and 17 mmHg in the left eye. Examination by slit-lamp biomicroscopy revealed that the anterior segments of both eyes were normal. Large areas of retinochoroidal atrophy were observed in the posteriors of both fundi, and the lesions extended outward and invaded the retinal equator, forming annulus-shaped yellow-white flat protrusions with a rough surface and some pigment deposition (Figures 4A and 4C). FFA revealed that most of the areas of the lesions around the optic disc did not show fluorescence, whereas the other areas, which extended outward and formed a wide ring zone, exhibited strong fluorescence staining (Figures 4B and 4D). Bilateral orbital CT scanning revealed arc-shaped high density areas in the posteriors of both eye rings (Figure 4E).

The parent and younger brother of the patients in family 2 were healthy and had normal vision. Examination of the anterior segment and fundus did not reveal any abnormalities. The elder sister of the first diagnosed patient had a 2-year-old daughter, whose eye examination was normal for both eyes. Two patients were followed up for 4 years. Neither the bilateral fundus nor the visual activity exhibited any significant changes.

Discussion

Choroidal osteoma frequently occurs in one or both eyes of otherwise healthy young women and progresses slowly. The symptoms of this disease include decreased vision, visual distortion, and visual field defects. The etiology and pathogenesis of choroidal osteoma remain unknown. It was previously believed that osteoma might be caused by a heterotopic ossification due to trauma and inflammation and that osteoma might also associate with the calcification caused by the long-term use of certain drugs. It was also suggested that choroidal osteoma could develop from the congenital residual original mesodermal tissue in choroid. Choroidal osteoma has also been called

choristoma [1]. Since the clinical confirmation of the disease, there have been many reports describing obvious genetic predispositions among the families of patients [2-6]. We reported that two sets of sisters in two families developed the disease, indicating a genetic predisposition, and this is the first report of such cases in China. The times of onset of the disease in the patients are unknown. Clinical diagnosis was based on the fundal appearance accompanied by FFA examination and on the specific changes of osteoid high density in the posteriors of the eye rings that were revealed by eye CT scanning. The degree to which choroidal osteoma affects vision depends on the tumor location and on the secondary retinal damage caused by the disease. Long-term progressive retinal degeneration can lead to a gradual vision loss. Choroidal osteoma is not rarely seen in clinical practice. Effective treatment is still lacking. We believe that an investigation of the etiology and pathogenesis will be a key factor for finding a treatment. Studies have demonstrated that genetic factors should not be excluded. Further study should focus on genetic examination and its association with the disease.

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