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A case of PAX6 gene mutation with bilateral Peters anomaly

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Abstract

The patient in this case can walk alone and take care of himself. Therefore through surgical separation and removal of the cloudy lens, the establishment of a visual pathway can help improve the children's vision and improve their quality of life.

A case of PAX6 gene mutation with bilateral Peters anomaly

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Abstract

Rationale: Peters anomaly as a kind of anterior segment diseases was rarely reported, we reported a case of PAX6 gene mutation with bilateral Peters anomaly treated with cataract surgery.

Patient concerns: A 5-year-old male was referred to our ophthalmologic hospital, because of bilateral corneal opacity since birth. Anterior segment photograph showed the central corneal opacity with neovascularization in the right eye, which peripheral cornea was clear and peripheral pupillary membrane could be found. The obscured opacified lens adhesion to the porcelain white cornea could be found in the left eye. A heterozygous PAX6 gene mutation c.357+1G>C leads to splicing mutations in amino acids.

Diagnosis: Peters anomaly Type 2

treatment: Cataract extraction combined with anterior vitrectomy in the double eyes in different time.

Lessons: The patient in this case was in stable condition after surgery, can walk alone and take care of himself. The patient and family members are satisfied with the treatment scheme. Therefore through surgical separation and removal of the cloudy lens, the establishment of a visual pathway can help improve the children's vision and improve their quality of life.

Keywords: Peters anomaly; PAX6 gene;

Key Clinical Message

A 5-year-old male was referred to our ophthalmologic hospital, A heterozygous PAX6 gene mutation c.357+1G>C leads to splicing mutations in amino acids. Peters anomaly Type 2 was made. Cataract extraction combined with anterior vitrectomy surgery was mede in the double eyes in different time.

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Peters anomaly(PA) is a form of anterior segment malformation, characterized by corneal opacity and keratolenticular adhesion with associated defects in the posterior layers of the cornea. PAX6 gene mutation may be related to anterior segment abnormity such as PA[1]. Congenital corneal opacity as a serious vision-threatening disease, leads to severe visual impairment. The PA has been classified to Type 1, Type 2 and peters anomaly syndrome, according to different clinical manifestation. There are different surgical approaches have been taken to treat different PA. The clinical diagnosis and management of Peters anomaly type II in a Chinese child has been reported in our presentation.

A 5-year-old male was referred to our ophthalmologic hospital, because of bilateral corneal opacity since birth. Written informed consent was obtained from the patient's parents to create this report. He was delivered normally by the mother and there was no the experience of oxygen uptake. Both parents are in good health. There was no congenital abnormities nor similar ocular disorders was revealed in family members. The patient underwent fundamental ophthalmic examinations. His vision was hand movement in double eyes, so he can not walk independently. The intraocular pressure measured with I-care tonometer (Finland Oy, Helsinki, Finland) was respectively 21.3 mmHg(1mmHg= 0.1333224kpa) and 32 mmHg in the right and left eye. Anterior segment photograph(Haag-Streit, Bern, Switzerland) (Figure 1A) showed the central corneal opacity with neovascularization in the right eye, which peripheral cornea was clear and peripheral pupillary membrane could be found. The obscured opacified lens adhesion to the porcelain white cornea could be found in the left eye (Figure 1B). The retina and optic disc could not be seen. Binocular nystagmus. Axis oculi measured with ophthalmic A - scan ultrasonography was 21.83mm and 23.44mm in the right and left eye. B-scan ultrasonography(Figure 2) showed a normal posterior segment in the double eyes. Anterior segment OCT(Figure 3) showed lens echo enhanced and adhere to the cornea. Genital, skeletal and cardiac abnormalities didn't present through physical examinations. We did the genetic testing for the patient and his parents, by extracting the peripheral blood for genetic testing and found mutations in the PAX6 gene. Individual exons of the PAX6 gene was amplified by polymerase chain reaction(PCR). A heterozygous PAX6 gene mutation c.357+1G>C leads to splicing mutations in amino acids(Figure 4). However, PAX6 gene of his parents was normal, so it was spontaneous mutation. The diagnosis of Peters anomaly Type 2 was made. Immediate cataract extraction combined with anterior vitrectomy in the right eye was advised, and the patient's parents consented our surgical scheme. Under general anesthesia, a conjunctival peritomy was created at the area of maximally clear peripheral cornea. A limbal incision was fashioned directly at corneal limbus. Viscoelastic was injected to the anterior chamber to create space between the cornea and lens. Then Inserted anterior chamber perfusion. During the operation, we found that the lens was not fully developed and optoaxial lens was opacity. 23G vitrectomy removed the lens and anterior vitreum(Figure 5A). The corneal incision was then closed with 10–0 Vicryl suture. The first day after surgery, the intraocular pressure measured with I-care tonometer was 16.5 and 35mmHg in the right and left eye. The viusal acuity was FC/40cm. The right eye was treated with anti-inflammation and mydriasis therapy. 1 week later, the same operation was performed to the left eye. Because the cornea was porcelain, 23G vitrectomy removed the lens with the assistance of 23G cold light illuminator (Figure 5B). During the operation, we found that the lens was not fully developed and optoaxial lens was opacity, indistinctly. Separation of adhesive opacified lens and anterior vitreum was performed. The first day after surgery of left eye, there was no obvious abnormality in slit-lamp examination(Figure 6). Anterior segment OCT(Figure 7) and B-scan ultrasonography(Figure 8) showed a normal anterior and posterior segment in the double eyes. Viusal acuity was 0.01 and hand movement in the right eye and left eye. The intraocular pressure measured with I-care tonometer was 19.5mmHg and 18.2mmHg in the right and left eye. Topical antibiotics and corticosteroid drops were applied after the procedure and were gradually reduced.

PA as a rare congenital dysgenesis of the anterior segment diseases, was first described in 1906 by Dr Alfred Peters[2] PA is a polygenic hereditary disease caused by abnormal neural crest cell migrate to cornea during development. This abnormal migration has been proved to be related to mutations in pax6, pitx2, foxe3 and cyplbl genes. PAX6 mutation also has been proved as a Genetic Factor in one Peters'anomaly patient of Tetsuyuki Yasuda research[3]. Alough mutation of the PAX6 gene maybe related to Peters'anomaly, Calvas et al. investigated the PAX6 coding region in four patients with Peters' anomaly but without any

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mutations[4]. We found exon 6 mutations in pax6 gene that were previously confirmed to be aniridia related[5]. The etiology of PA mainly includes intrauterine infection, incomplete separation of lens vesicles from the epidermal embryoblast and developmental disorder of nerve cells[6]. Therefore, the environment and genes maybe both as pathogenic factors[7]. The clinical sign of Type II PA is keratoleukoma with cataract or adhesion of corneal and lens[8]. Clinical ophthalmic characteristic led us to classify our patient as Type II PA. The management of PA depends on the corneal status and accompanied anomalies. Haruna Yoshikawa considers it is likely that in eyes with the less-severe type of PA without glaucoma, corneal opacity can be expected to decrease during the first several years from birth. However, patients with cataract and glaucoma should be given surgical intervention to prevent sensory amblyopia and irreversible glaucoma damages. In this case presentation, we successfully removed the cataract. Because the corneal of left eye was porcelain, 23G vitrectomy removed the lens with the assistance of 23G cold light illuminator to overcome the difficulty of the lack of transparent cornea. Due to abnormal ocular tissue development and visual impairment, patients with Peters abnormality have greater difficulty in refractive correction in the evaluation of therapeutic effects and postoperative refractive parameter measurement. The patient in this case was in stable condition after surgery, can walk alone and take care of himself. The patient and family members are satisfied with the treatment scheme. Therefore through surgical separation and removal of the cloudy lens, the establishment of a visual pathway can help improve the children's vision and improve their quality of life.

Figure 1A:Anterior segment photograph showed the central corneal opacity with neovascularization in the right eye. Figure 1B: The obscured opacified lens adhesion to the porcelain white cornea could be found in the left eye.

Figure2: B-scan ultrasonography showed normal posterior segment in the double eyes

Figure3 Anterior segment OCT showed lens echo enhanced and adhere to the cornea

Figure4: A heterozygous PAX6 gene mutation c.357+1G>C leads to splicing mutations in amino acids, PAX6 gene of his parents was normal.

Figure5A: 23G vitrectomy removed the lens and anterior vitreum in the right eye.

Figure5B: 23G vitrectomy removed the lens with the assistance of 23G cold light illuminator in the left eye.

Figure6: Anterior OCT showed the normal anterior segment in the double eyes

Figure7: B-scan ultrasonography showed a normal posterior segment in the double eyes

There is no conflicts between the authors

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