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Peters-Plus Syndrome: What Outcome in the Absence of Genetic Confirmation? A Case Report

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Abstract

Anterior segment dysgenesis is a group of non-acquired ocular anomalies whose cause is multifactorial; many genes are involved. It is characterized by developmental anomalies of the tissues of the anterior segment, of which Peters-Plus syndrome is included. Our aim is to describe the different ophthalmological and systemic aspects of Peters-Plus syndrome in order to improve the quality of diagnosis of this syndrome even in the absence of genetic confirmation, especially in low-income countries or when genetic studies are not available. In this observation, we report the case of a newborn with Peters-Plus syndrome admitted to the neonatology unit. The diagnosis was made on the basis of clinical-radiological criteria, and treatment consisted of caring for the baby and the parents, given the particular psychological context often associated with the birth of a baby with polymalformative syndrome. From this study, Peters-Plus syndrome should be borne in mind in a fetus with typical ocular anomalies, unusual facial appearance and long tubular bone insufficiency, especially in the presence of a positive family history. In such cases, prenatal diagnosis could be an option for the couples. A genetic study should be undertaken to confirm the clinical diagnosis and provide appropriate genetic counseling and prenatal diagnostic options.

Keywords

Peters-Plus Syndrome, Genetic Study, Prenatal Diagnosis

1. Introduction

Peters syndrome is a rare congenital disorder with autosomal recessive inherit-

ance. Peters anomaly is a developmental anomaly of the tissues of the anterior segment of the eye. It is named after Dr. Alfred Peters, a German ophthalmologist. It is characterized by a central corneal opacity of variable size, with a corresponding defect in the posterior stroma, Descemet's membrane and endothelium. The peripheral cornea is relatively clear, but a variable degree of cloudiness might be associated with the central opacity. In Peters' anomaly, iridocorneal adhesions usually arise from the collarbone and attach to the edge of the corneal opacity [1].

Peters' anomaly is currently classified into two types: Type 1 is characterized by central corneal opacity and iridocorneal adhesions. Type 2 is further characterized by lens opacity and lenticocorneal adhesions [2].

Peters-Plus syndrome is defined by the presence of multiple anomalies including: Peters anomaly of the eye (anterior segment dysgenesis), short stature, shortened limbs with brachydactyly, facial dysmorphia, developmental delay, craniofacial anomalies (including cleft lip/palate and enlarged forehead) and other variable anomalies [3]. Although bilateral Peters anomaly is the most common ophthalmological anomaly in Peters-Plus syndrome, unilateral involvement or a different anterior chamber defect can also be observed [4].

2. Case of Study

A male newborn from a pregnancy estimated at 36 weeks of amenorrhea according to the date of the last menstrual period. No parental consanguinity, vaginal delivery. The newborn was admitted with a malformation syndrome and unilateral corneal opacity on the left side. Clinical examination revealed symmetrical intrauterine growth retardation with a height of 46 cm (5 percentile), weight 2340 g (5 percentile), head circumference 33 cm (25 percentile). Facial dysmorphia: wide forehead, low-set ears, with cleft palate. An ophthalmological examination of the left eye revealed hypertelorism associated with unilateral buphthalmia with lagophthalmos and corneal opacity (**Figure 1**). Slit-lamp examination of the right eye was unremarkable, with ocular tone at 2 mmHg. The left eye showed a totally opaque megalocornea (14.5 mm × 14 mm) with superficial neovascularization surrounded by a bluish zone of scleral extension, and a nonindividualized anterior chamber, the posterior segment could not be visualized. The ocular tone was 16 mmHg.

B-mode ocular ultrasound revealed no papillary abnormalities. Ultrasound Bio-Microscopy (UBM) revealed a thinned cornea with a lens attached posteriorly, a rudimentary hypoplastic iris and a nonindividualized iridocorneal angle (**Figure 1**). Examination of the external genitalia revealed unilateral testicular ectopia on the right, associated with hypospadias. All other clinical findings were normal. Cerebro-orbital MRI showed asymmetric eyeball size with an axial length of 18.5 mm on the left, without individualization of the anterior chamber of the left eyeball and without associated cerebral anomaly (**Figure 1**).

In the context of polymalformative syndrome, ToRCH serologies were nega-

tive. Abdominal ultrasound, transfontanel ultrasound and echocardiography were normal. The karyotype did not detect any chromosomal abnormalities and the genetic study was not performed by lack of means.

Medical measures being considered to regulate intraocular pressure include the use of topical hypotonizing eye drops and artificial tears to prevent exposure to keratitis. In addition, penetrating keratoplasty surgery is planned to correct ocular anomalies but the ophthalmologists plan to perform it at the age of 3 months because the newborn is underweight. Other measures included cleft palate repair, parental education, psychological care and support for both parents. Written consent for the publication of photographs has been obtained from the family.

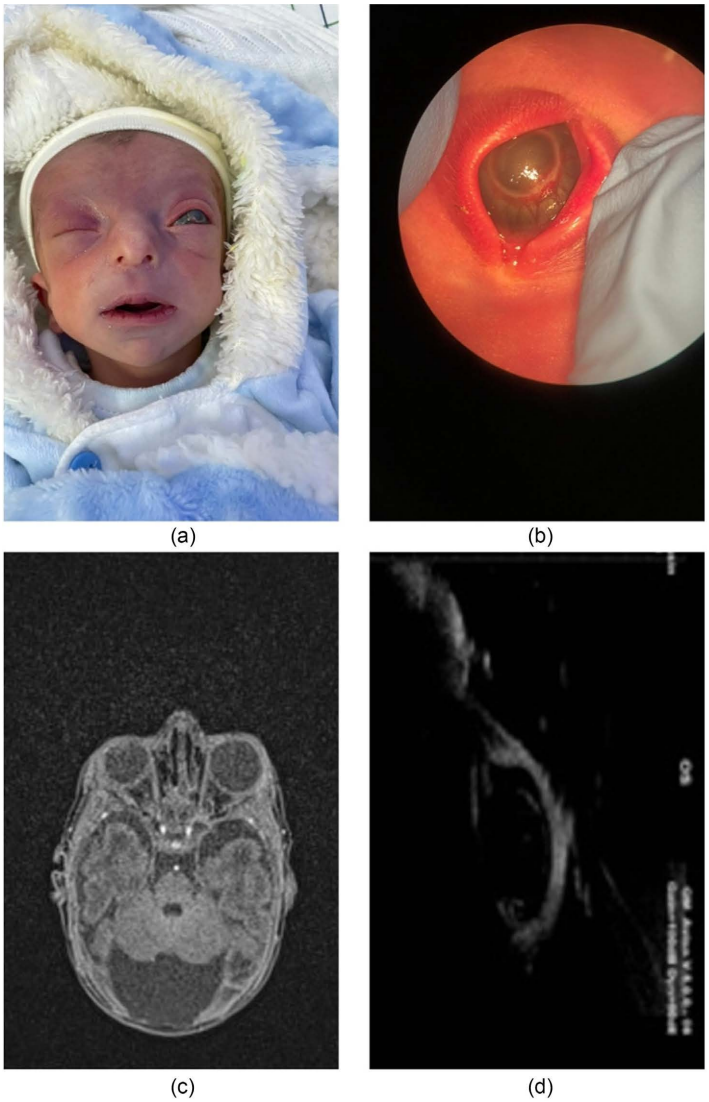

Figure 1. (a) Sign of lagophthalmos. (b) Totally opaque cornea with superficial neovascularization. (c) Axial T1-sequence cerebro-orbital MRI: enlarged left eyeball with absence of invidualization of lens and anterior chamber. (d) Axial-section UBM: dislocated crystalline lens abutting posterior corneal surface (blue arrow), hypoplastic rudimentary iris (orange arrow).

3. Discussion

Corneo-irido-gonio-dysgenesis glaucoma is a generic term for several disorders characterized by anatomical abnormalities of the anterior segment of the eye, including Peters anomaly, Peters-Plus syndrome, congenital sclerocornea and anterior segment staphyloma [5].

Peters-Plus syndrome is due to a biallelic mutation in the B3GLCT gene, which associates the Peters anomaly (anterior segment anomaly) with systemic anomalies such as rhizomelic shortening with large hands and brachydactyly, short stature, auricular malformations, congenital heart defects, genitourinary anomalies, structural brain defects, and facial anomalies such as the round face, prominent forehead, hypertelorism, long philtrum, micrognathia, Cupid's bow upper lips, cleft lip or palate [1] [6]. This syndrome should be borne in mind in any newborn presenting the ocular anomalies associated with an unusual facial appearance. However, other etiologies of anterior segment dysgenesis, notably staphyloma and sclerocornea, may be evoked in the presence of a similar picture. Unlike congenital sclerocornea, where there is non-progressive scleralization of the cornea [5], staphyloma is characterized by an enlarged, vascularized, opaque, ectatic cornea with thinning and ectasia of adjacent structures. Staphyloma is a severe form in which the damage may be unilateral or bilateral. In most cases, these two anomalies are not associated with systemic abnormalities, unlike Peters-Plus syndrome.

In situations where access to genetic confirmation is not possible, the diagnosis of such pathologies remains challenging. This was the case with this newborn, whose diagnosis of Peters-Plus syndrome was based on clinical and radiological criteria:

• Ophthalmological criteria: corneal opacity of the left eye with exophthalmos associated with unilateral glaucoma.

• Systemic anomalies: short, disproportionate stature, craniofacial anomalies and urogenital anomalies.

• UBM exploration: the presence of a thinned cornea with a lens attached to the posterior surface, a rudimentary hypoplastic iris and a non-individualized iridocorneal angle.

Prenatal diagnosis could be an option for couples with a family history, but establishing a diagnosis based solely on ultrasound findings can be difficult due to the presence of variable and non-specific results. A few sonographic dysmorphic features such as hydrocephalus, agenesis of the corpus callosum, microphthalmia and cleft lip and palate, as well as other structural anomalies in a fetus with growth retardation, may point towards a probable diagnosis of fetal plus Peters syndrome. Nevertheless, a diagnosis confirmed by molecular methods is the most reassuring. It also offers the possibility of early prenatal diagnosis for subsequent pregnancies, well before the appearance of non-specific ultrasound anomalies [7].

4. Conclusion

The prognosis for visual acuity in congenital corneal opacities of the corneo-irido-

gonio-dysgenesis type is poor, as it is difficult to restore visual function in the long term, and surgical interventions have a high risk of complications.

Genetic analysis is recommended to validate the clinical diagnosis and offer appropriate genetic counseling and prenatal screening.

Conflicts of Interest

The authors declare no conflicts of interest regarding the publication of this paper.

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