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Peters Syndrome: A diagnostic approach, clinical evolution and review

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ABSTRACT

Introduction: Peters syndrome (or Peters anomaly, or anterior segment mesenchymal dysgenesis) is a rare and congenital genetic condition, described by Arthur G. H. Peters in 1906, which affects the anterior ocular mesenchymal segment, presenting varying degrees of corneal opacity, iridocorneal synechiae, presence of anterior chamber narrowing and Descemet's membrane defect. Case Presentation: A 6-year-old boy, born in Goiânia, state of Goiás, with a term birth and no history of previous health problems, was referred to our service by a general practitioner due to reduced visual acuity (VA) problems in both eyes and strabismus. The mother, who accompanied him, reported that the symptoms of strabismus began approximately a year and a half ago. She also mentioned that she was properly vaccinated during pregnancy against infections that could cause birth defects and denied any complications during pregnancy or childbirth. There is no relevant family history of eye diseases.

Discussion: Because it is a rare anomaly, epidemiological data are scarce in the world literature, the only constant data is that the bilateral presentation is present in most cases. In this summary, we will address the main aspects of Peters syndrome, including its pathophysiology, classification, clinical characteristics, diagnosis and treatment. It is a syndrome that requires multidisciplinary evaluation to treat systemic complications, in some cases requiring a surgical approach such as penetrating keratoplasty and glaucoma treatment. Conclusion: The treatment varies according to the presentation of the condition and the genetic and multidisciplinary evaluation is of paramount importance in these patients.

Keywords: Peters anomaly, Amblyopia, Glaucoma, Congenital, Anterior segment dysgenesis.

1 INTRODUCTION

Peters' anomaly is a fairly common congenital corneal opacity, which occurs due to a defect in the migration of neural crest cells, resulting in malformation of the anterior segment of the eye. The anomaly was named after the German physician Albert Peters, who described it clinically and histologically in the early twentieth century.

The exact cause of congenital corneal opacities is still unknown, but there are suspicions that they may be genetic, infectious, traumatic, or toxic. It is known, however, that this etiological factor affects fetal development between the 6th and 16th week of gestation, a period in which differentiation of the anterior chamber occurs.

Although the occurrence of Peters' anomaly is usually sporadic, there are reports that it may be associated with mutations in the PAX 6 gene, which may have recessive or dominant expression with variable penetrance, and which also appears in other types of anterior segment malformations.

The main features of Peters' anomaly include central corneal opacity and synechiae of the iris and/or lens with the leucoma region. Corneal opacity is the most obvious symptom of the disease and can vary in density and size. Leukoma, which is surrounded by a transparent cornea, is caused by edema of the posterior stroma and the absence or thinning of Descemet's membrane and endothelium. In addition, there is disorganization of the epithelium and loss of the Bowman's layer. In more severe cases, leucoma can be vascularized and protruding. The atrophic iris adheres to the posterior aspect of the leucoma, which can lead to anterior synechiae and disorganization of the anterior chamber. Although the peripheral cornea is transparent, sclerization of the limbus may occur.

Peters' anomaly can occur alone or in conjunction with other ocular anomalies. In more severe cases, cataracts and glaucoma may develop (in 50% of cases). Other possible ocular changes include microcornea, microphthalmos, flat cornea, sclerocornea, irian coloboma, angle and iris dysgenesis, ptosis, and optic or foveal nerve hypoplasia. In addition, mental retardation, congenital heart disease, congenital nephropathy, cleft lip, craniofacial dysplasia, and bone malformations may occur, which, when present together with at least one of these systemic findings, make up Peters-plus syndrome.

2 CASE PRESENTATION

A 6-year-old boy, born in Goiânia, state of Goiás, with a full-term birth and no history of previous health problems, was referred to our service by a general practitioner due to problems with reduced visual acuity (CA) in both eyes and strabismus. His mother, who accompanied him, reported that the symptoms of strabismus began approximately a year and a half ago. She also mentioned that she was properly vaccinated during pregnancy against infections that could cause birth defects and denied any complications during pregnancy or childbirth. There is no relevant family history of ophthalmologic diseases.

At the initial ophthalmologic evaluation, the patient had a corrected visual acuity of 20/80 in the right eye and 20/120 in the left eye. The external examination showed divergent strabismus in the primary position of the gaze. Biomicroscopy showed bilateral corneal opacities, with a decrease in the red reflex. In addition, the iris appeared flat and glued to the cornea, exhibiting a narrow iridocorneal angle. The lens had congenital cataracts with posterior displacement.

Further evaluation, including measurement of intraocular pressure, was normal, and an ocular ultrasound confirmed congenital cataract and posterior displacement of the lens. Based on these clinical findings, the diagnosis of Peters Syndrome Type I was strongly considered, given the absence of systemic manifestations and the results of biomicroscopy. The family was properly informed about the condition, its implications, and the treatment options available.

3 DISCUSSION

Characterized by corneal opacity, Peters syndrome is a rare congenital disorder due to a defect in the migration of neural crest cells. Thus, the patient with Peters Syndrome will have a malformation of the anterior segment of the eye due to the interference of the etiological factor during the 6th and 16th weeks of gestation. The etiology of Peters Syndrome is still uncertain, encompassing genetic and environmental factors, such as infections, trauma, and toxic substances that interfere with the period of embryonic differentiation of the anterior chamber. (MEYER, 2010).

In 1906, Albert Peters first described Peters Syndrome by noting the association between central corneal leukoma, iridocorneal synechiae, the presence of a narrow anterior chamber, and a defect in Descemet's membrane. Alterations in the embryonic development of the different layers of the cornea differentiate Peters Syndrome into two phenotypes. Type 1, which is more restricted, without systemic symptoms, is characterized by central corneal clouding and iridocorneal synechiae. Type 2, on the other hand, is clinically more severe, with denser corneal opacifications, deterioration of the lens, and the presence of systemic symptoms. There is also Peters Plus Syndrome, described by Schooneveld, et al., in 1984, in which the patient, in addition to the typical clinical of Peters Syndrome, presents symptoms such as cleft lip and palate, dysmorphia of the face and hands, short stature,

anomalies of the pinna, and mental retardation (FERREIRA, 2020).

Patients with Peters Syndrome may present a greater association with other ocular pathologies, such as glaucoma (occurs in 50% of cases), cataracts, microcorneas, microphthalmos, flat cornea, sclerocornea, irian coloboma and ptosis (MEYER, 2010).

According to studies conducted by Bhandari et al, in 2011, Peters' Anomaly prevails among males, affecting 56% of cases, and in 67.2% the symptoms manifested bilaterally, with a notable association with systemic malformations (71%).

Patients with mild corneal opacity, not associated with cataracts, have adequate vision evolution and a good prognosis, with treatment often reserved for peripheral iridectomy. On the other hand, those with Peters Syndrome, with extensive densification, associated with systemic and ocular alterations, have a poorer prognosis, encompassing broader therapeutic measures, such as penetrating keratoplasty (MEYER, 2010).

The management of Peters Syndrome aims to provide an ideal visual stimulus to these patients, avoiding the triggering of complications (CALIXTO, 2023).

Among the complications of Peters Syndrome, we can mention amblyopia, unilateral or bilateral, due to sensory deprivation (consequent to central corneal opacification, cataract and glaucoma), retinal detachment, and spontaneous corneal perforation (FERREIRA, 2020).

There is no cure for the syndrome, or for anterior desgenesis of the eye. The only way to avoid aggravation and visual recovery is total ceatoplasty (corneal transplantation). This should be considered for corneal involvement before 3-6 months of age; and also surgical intervention in cases of glaucoma. Periodic follow-up with a pediatric ophthalmologist is essential, and the use of corticosteroids should be avoided in these children due to the high risk of developing glaucoma.

Peters Syndrome is a rare congenital disorder with uncertain etiology that requires early diagnosis and treatment, enabling better prospects for the patient and reducing the risks of visual impairment. Thus, adequate ophthalmologic investigation and efficient pediatric follow-up are essential, with the purpose of early detection of any alteration that may aid in the diagnosis.

4 CONCLUSION

Peters Plus Syndrome is an autosomal recessive genetic disorder with significant consanguineous involvement. It consists of Peters' Anomaly associated with short stature, brachydactyly and cognitive alteration of varying degrees, as well as a cause of fetal death in severe cases. It may also present cardiovascular and genitourinary involvement. When non-lethal, it is responsible for important comorbidity and impaired quality of life. (ALMEIDA et al, 1991; CANDA et al, 2018)

Diagnosis is uncertain and is based primarily on ectoscopy and physical examination of the

patient. Family history can also be important. Complementary tests may be relevant for the investigation of adjacent involvements, such as echocardiography for constitutional defects of the heart and basal vessels and ultrasonography of the kidneys and urinary tract for deformities, for example. Alterations in the sequential analysis of the B3GLCT gene may be present, showing homozygous mutation in the offspring and heterozygous mutation in the parents. In this way, the autosomal recessive inheritance of the pathology is elucidated. (DEMIR et al, 2020; HESS et al, 2008)

Detection by imaging is possible even in the intrauterine period and enables early intervention of the pathology, attenuating the repercussions resulting from the syndrome and helping to maintain a better quality of life. Morphological ultrasound, performed in the prenatal period, is useful for the verification of some findings that indicate the presence of the syndrome, such as agenesis of the corpus callosum, microcephaly, short limbs and polycystic kidneys. Case If the doubt persists, the MRI can bring even more details: hypotelorism and bilateral cataracts. (DEMIR et al, 2020; CANDA et al, 2018).

The possibilities of intervention for the condition are limited and consist only of treating the clinical manifestations of the condition. Bilateral corneal transplants may be considered, and motor physiotherapy may also be indicated, with better results in milder cases. (DEMIR et al, 2020).

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