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Case Report

Cataract in Treacher Collins Syndrome

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Abstract

A 13-years old patient with Treacher Collins Syndrome presented with lens opacification. Initial visual acuity was 20/200 at the right eye and 20/100 at the left eye. A lamellar opacification of the lens, as banana-tree-leaves-shaped cataracts, was observed in both eyes. Phacoemulsification through corneal incision, with an anterior circular capsulorhexis was performed in both eyes. Intraocular lens implantation was performed. Post cataract surgery visual acuity was 20/30 in both eyes. Cataract is a rare feature in Treacher Collins Syndrome and the lens opacity morphology detected in this patient was very unique. The authors outline the importance of a multidisciplinary team approach, including pediatrician, geneticist, psychologist, neurologist, speech therapist, pediatric ophthalmologist and plastic surgeon to take care for patients with TCS.

Keywords: *Cataract; Pediatric Cataract; Treacher Collins Syndrome*

Abbreviations: TCS: Treacher Collins Syndrome

Introduction

Treacher Collins Syndrome (TCS) or mandibulofacial dysostosis is a genetic disorder characterized by typical *facies* with malar and mandibular hypoplasia, antimongoloid eyelid position, auricular pavilion malformations, conductive deafness, cleft palate, and a bird's beak-shaped nose [1,2].

The syndrome's target gene, TCOF1, was mapped on the distal portion of the long arm of chromosome 5 (5q31.3-q33.3) with autosomal dominant transmission of variable expressivity. This gene responsible for TCS has recently been cloned, and a protein product has been identified with homology to a family of nucleolar-cytoplasmic transport proteins. Almost all mutations identified in TCS result in premature termination of the protein product, suggesting that the pathogenetic effects result from haplo insufficiency of the gene product during embryogenesis. The precise function of this protein product and its role in TCS pathogenesis remain unknown [2].

The clinical manifestations are probably due to a congenital malformation involving the first and second pairs of pharyngeal arches, with an incidence ranging from 1:40,000 to 1:70,000 cases per birth [1].

Lower eyelid coloboma is frequently seen in TCS and is the main reason ophthalmologists are consulted as part of the multidisciplinary team to treat patients with this syndrome. Other ocular findings, more rarely, may be part of TCS, such as microphthalmia, tear duct atresia and cataract [2,3].

A large multicenter study was conducted by Rooijers et al. to analyze ocular involvement in children diagnosed with TCS. Inferior lid coloboma was a major ocular manifestation, this defect was observed in 98.5% of the patients and no cataract was detected in this group with TCS [1]. There is a prior report of cataract surgery in TCS, in a 13 months old child, that presented a late onset cataract [3].

Cataract in patients with TCS is not a common oc-

currence and has been rarely described in previous literature [3]. We report a case of a patient with bilateral cataract associated with TCS.

Case report

A 13-year-old male diagnosed with TCS that presented with mild hearing loss, low ear implantation, and bird´s beak-shaped nose (Figure 1), was referred to CAVIVER Eye Clinic, in the city of Fortaleza, CE, Brazil, for ophthalmological evaluation. The patient´s parents were first cousins, showing no symptoms or signs of TCS. The patient was referred from the Genetic Clinic.

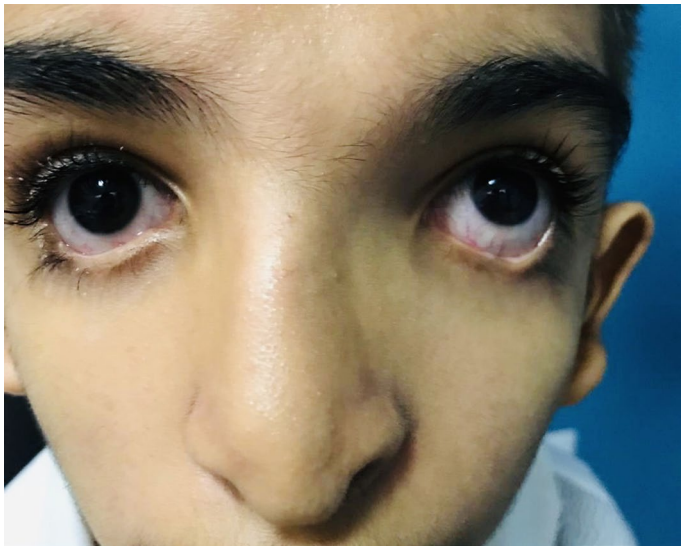

Figure 1: Facial features: partial bilateral inferior eyelid coloboma, nose and ear malformations

The patient complained of progressive bilateral vision loss. A comprehensive ophthalmic evaluation disclosed visual acuity of 20/200 and 20/100 in the right eye (RE) and left eye (LE), respectively. Indirect binocular ophthalmoscopy revealed normal eye fundus. Refraction was not feasible in RE due to dense lens opacity. The refraction of the LE was - 2,25 sph = - 4,50cyl (135°). Neither nystagmus nor strabismus was observed. A partial bilateral inferior eyelid coloboma was present.

A bilateral lamellar lens opacification resembling banana-tree-leaves was observed through slit-lamp examination. The peripheral lens cortex was clear in both eyes (Figure 2).

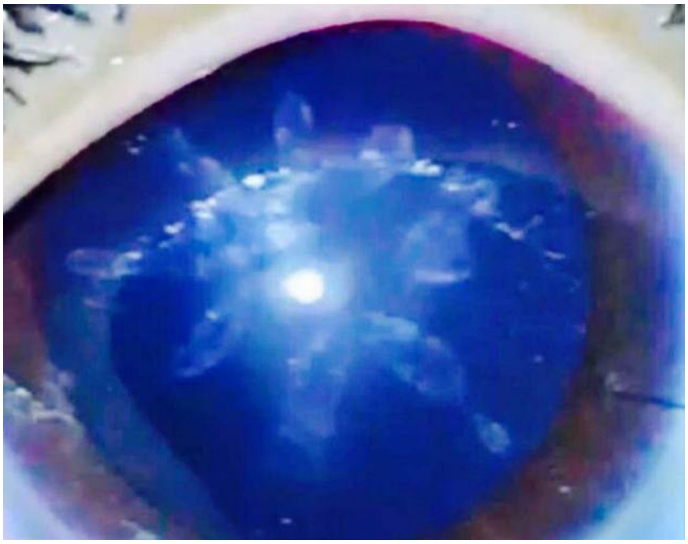

Figure 2: Morphology: banana-tree-leaves-shaped cataract

Axial length was 26.7mm in RE and 25.5mm in LE. Keratometric readings were 42.25 x 45.75 diopters (120°) in RE and 42.00 x 46.50 diopters (145°) in LE. Intraocular lens power calculation was +11.0 and +13.0 diopters in the RE and LE, respectively.

A phacoemulsification through corneal incision, with an anterior continuous circular capsulorhexis, was performed in both eyes, in different dates and settings. Viscoelastic substance was injected to maintain stability of the anterior chamber during surgery. Aspiration of the soft nucleus needed low ultrasound power. All nucleus and lens cortex were carefully aspirated. A hydrophobic mono-focal foldable acrylic intraocular lens was implanted in the bag. The posterior capsule was clear and was left intact bilaterally. The procedures were uneventful. Post-operative refraction was + 0.75 sph = - 3.00 cyl (40º) and - 1.00 sph = - 4.00 cyl (145°), respectively in RE and LE. The final best corrected visual acuity achieved 20/30 in both eyes.

Discussion

Treacher-Collins syndrome is a genetic condition with a marked craniofacial abnormality. The gene responsible for TCS at chromosome 5 (5q31.3-q33.3) was studied and genetic testing for prenatal diagnosis is available for affected families [2].

Rooijers et al. studied the prevalence of ocular and adnexal anomalies in 194 patients with TCS. Primary ocular anomaly, as inferior lid coloboma,

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was reported in 98.5% of cases, and secondary anomalies in 34.5%, strabismus in 27.3%, refractive errors in 49.5%, and visual impairment in 4.6%. No cataract was reported in this multicenter study with a large number of affected patients [1].

Only one published paper reported lens opacification in TCS. Biebesheimer and Frederick reported bilateral extracapsular cataract extraction with posterior chamber lens implantation in a 13-month-old patient with delayed-onset nuclear cataract [3].

The report in our case highlights a rare association between TCS and cataract, contributing to our understanding of the ocular manifestations in this genetic disorder. In this patient, visual complaints in an early stage of the cataract formation made possible the observation of the initial morphological aspect of the lens opacification. Younger children usually don't complain about loss of vision, thus leading to late diagnosis of cataracts in preschool children.

Pediatric cataract morphology may be associated with specific systemic disorders suggesting the possible aetiology of the lens opacification and suggesting heritability [5]. Potential mechanisms behind the development of cataracts in TCS, other than genetics, were not totally elucidated, and environmental factors seem not to be involved.

Cataract morphology can be a visual prognosis indicator. Different morphological types may have a better visual prognosis than others, with lamellar cataracts doing well and total cataracts relatively poorly. Lamellar cataracts frequently develop at a later stage and can be progressive. A published study of children with congenital cataracts showed that visual prognosis might depend on the morphological type, with less favorable outcomes in cases of total cataracts [6,7].

Pediatric cataracts can exhibit phenotypical heterogeneity. In a previous study, 207 patients with pediatric cataract were studied. The age of the patients ranged from 19 days to 12 years. Zonular cataracts occurred most frequently in 72 (33.8%) patients. Among the zonular cases, the lamellar subtype was the most common (66.7%). Total cataract occurred in 31.9% of the patients. Among the patients with genetic disorders or syndromes, 87,5% had bilateral cataract. Morphology or laterality of pediatric cataract may be indicative of its aetiology [8].

The banana-tree-leaves-shaped cataract morphology was a very unique finding in the case of TCS enorted here. This cataract morphology was not previously described in the literature and could be associated with early stages of the lamellar lens opacification in TCS patients. The morphological heterogeneity of pediatric cataracts makes the morphological classification challenging, but morphology is of great importance in guiding aetiology diagnosis, and treatment options.

The banana-tree-leaves-shaped cataract morphology is a novel finding that adds to the phenotypic heterogeneity observed in pediatric cataracts. The specific effect of this cataract on this child's demonstrated a progressive diminishing of visual acuity and altered visual function, with complaints of sensitivity to light and disturbance in his ability of contrast vision. In this patient blurring of vision occurred and a rapid progression toward severely decreased vision was detected.

Within a couple of months, progressive lens opacification resulted in visual acuity of light perception due to the development of a total cataract in both eyes. Phacoemulsification and intraocular lens implanted in the bag was performed in both eyes with no complications [6]. Due to the late onset of the lens opacification, which occurred after to the amblyogenic period, and due to early detection and treatment, a good visual acuity was restored in this case reported here after cataract surgery.

The lack of investigation of cataract genetic panels may advocate for overlapping genetic diseases in the same patient, thus knowing that consanguinity was reported in this present case. Further genetic analysis must be prescribed for this patient to clarify possible occurrence of other altered genes.

Genetic analysis could offer vital insights into the etiology of cataracts in this patient and possibly reveal novel genetic markers linked to this condition and might substantiate the link of this cataract phenotype to TSC. It was not possible to obtain genetic analysis at the moment of surgery. Thus, there is no strong evidence that the banana-leaves-shaped cataract is a specific phenotype of

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TCS.

The authors outline the importance of a multidisciplinary team approach, including pediatrician, geneticist, psychologist, neurologist, speech therapist, pediatric ophthalmologist and plastic surgeon to take care for patients with TCS. Ophthalmologic evaluations are important to analyze the visual acuity, ocular manifestations as cataract formation, integrity of cornea, and adnexal structures anomalies, as lower eyelid defects. Early ophthalmological evaluation is recommended in this genetic disorder.

The uniqueness of the cataract morphology in this case might have potential implications for diagnosis and management of TCS patients, as well as the need for further research to understand if the physiopathology of this specific cataract type in TCS is linked to the protein product identified at the genetic researches in TCS.

Accurate diagnosis of the aetiology of pediatric cataracts is important for epidemiological studies and to promote ocular health preventive programs with aim to prevent childhood blindness.

Conclusion

The banana-tree-leaves-shaped cataract morphology in TCS is a rare and a unique finding in the case described here. Healthcare providers should be vigilant about ocular involvement, decreased vision and complications in TCS patients, even if they are uncommon. Patients with TCS need a multidisciplinary approach to achieve a better quality of life. Routine ophthalmological evaluation is recommended in order to avoid visual impairment.

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Authors declare

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