CASE REPORT

Co-occurrence of Treacher-Collins syndrome and Down syndrome: case report and conduct

Coocorrência de síndrome de Treacher-Collins e de síndrome de Down: relato de caso e conduta

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ABSTRACT

Treacher-Collins syndrome is a rare, inherited malformation of the first and second brancheal arches and Down syndrome or trisomy 21 is the most common human chromosomal alteration. This study describes the co-occurrence of these two syndromes, and is the second report so far ever to do so in the literature. It also shows that early diagnosis and proper treatment of craniofacial malformations are essential to prevent amblyopia and to improve quality of life.

Key words: Mandibulofacial Dysostosis; Down Syndrome; Amblyopia; Vision, Low.

RESUMO

A síndrome de Treacher-Collins é uma malformação hereditária rara do primeiro e segundo arcos branquiais, enquanto a síndrome de Down ou trissomia do cromossomo 21 é a mais frequente alteração cromossômica humana. Este estudo descreve a coocorrência dessas síndromes, constituindo-se no segundo relato até agora descrito na literatura. Evidencia também que o seu diagnóstico precoce e o tratamento adequado das malformações craniofaciais são fundamentais para prevenir a ambliopia e melhorar a qualidade de vida.

Palavras-chave: *Disostose Mandibulofacial; Síndrome de Down; Ambliopia; Baixa Visão.*

INTRODUCTION

The Treacher-Collins syndrome is a hereditary, autosomal dominant malformation of high penetrance of the first and second branchial arches[1,2] with great phenotypic variation.[3] It results in malar and mandibular hypoplasia, ear malformations, deafness, eyelid coloboma, macrostomia, cleft palate, and skeletal alterations.[1] The ocular structure is usually preserved and there may be canthal dystopia, lipodermoid, lacrimal duct atresia, distichiasis, and strabismus.[2] The Down syndrome, characterized by the trisomy of chromosome 21, is the most frequent human chromosomal alteration.[4-7] In addition to mental retardation and heart alterations, it is often associated with slanting palpebral fissures, epicanthic, ametropia, stains in the iris, cataracts, keratoconus, strabismus, and nystagmus.[3-5] These syndromes, although they share some features such as cleft palate, low implantation of ears, cardiac malformations, and hypertelorism,[8] are of distinct clinical presentation and with very rare co-occurrence; this case is the second description in the literature so far.

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CASE DESCRIPTION

Male patient, born in 1/11/03, followed up at the Children's Low Vision Service at the São Geraldo Hospital (HSG) since 4/12/04, when he was transferred by the Sectors of Cornea and Ocular Plastic due to sequelae from the Down and Treacher-Collins syndromes. Since then, he has been followed up in the early intervention and visual stimulation program. His personal history are of natural childbirth, to a term pregnancy, without complications, with the genetic diagnosis of Down syndrome by robertsonian translocation between chromosomes 14 and 21 and Treacher-Collins syndrome, presenting hypothyroidism, hypotonia, difficulty feeding, growth and developmental retardation, seizures, cleft palate, malar hypoplasia, and mandibular malformation with microtia ears and conduct agenesis and bilateral ossicular chains. Clinical treatment of expositive corneal infected ulcer in the left eye (OE) in 3/18/03 and expositive keratopathy in the right eye (OD) were registered. He presented coloboma of lateral three-quarters in the lower eyelids with lower left eyelid reconstruction in 5/5/03 and lower right eyelid reconstruction in 6/6/03. He is the only son of young parents, non-blood related, karyotypes 46, XX and 46, XY, respectively. Mother G1P1A0, affected by the Treacher-Collins syndrome, as well as two of her brothers, his father, his three paternal uncles, and his paternal grandfather (Figure 1). The eye examination (2004) showed visual acuity of 0.64 cy/cm (20/960) in both eyes (AO), fixing and following light, with bright fringe and of high contrast objects; non-viable retinoscopy in AO; biomicroscopy: calm OD with inferior opacity: and calm OE with central leucoma; fundoscopy: OD with stained optic disc with crisp contour and physiological excavation, macula with preserved reflection and apparent rotation, light coroidose, normal retinal vessels, transparent vitreous, and non-viable OE. The ocular ultrasound suggested orbital congenital malformation without evident alteration in the eyepiece. Visual stimulation and monitoring with a multidisciplinary team were sustained. The evaluation conducted at the Subnormal Vision service in 11/16/10 showed AV of 4.8 cy/cm (20/130) in the OD (ment with depression) and 0.23 cy/cm (< 20/1900) in the OE and retinoscopy and fundoscopy unviable by corneal opacities (Figure 2). The patient is waiting for corneal transplant in AO. Despite the limitations imposed by the clinical condition, he presents good development in daily activities.

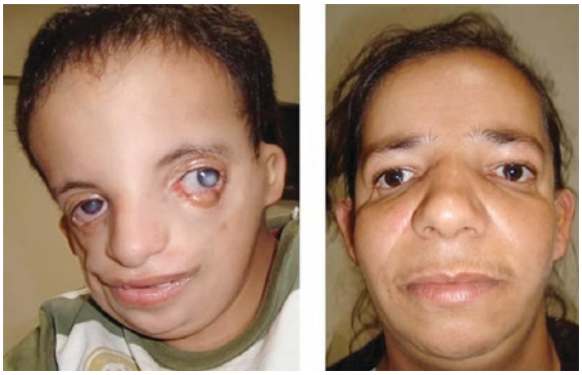

Figure 1 - Patient and his mother, both carriers of the Treacher-Collins syndrome (with the mother's permission).

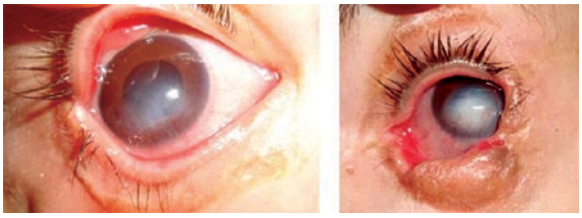

Figure 2 - Right and left eyes showing corneal opacities after expository keratitis by eyelid malformations already corrected.

DISCUSSION

Approximately 60% of patients with Down syndrome have some type of ophthalmologic abnormality, being the largest risk group for this type of abnormality in the pediatric population.[4] Different studies show high prevalence of refractive errors, cataract, strabismus, nystagmus, and infections among others.[4-7]

The Treacher-Collins syndrome brings ophthalmologic alterations in 100% of cases, and the most frequent are eyelid, strabismus, and ametropia alterations .[1,2] Because the eye structure is usually normal, refractive errors, anisometropia, strabismus, and ptosis in different degrees of association are the main responsible for amblyopia, which can therefore be prevented.[1,2]

Similarly to one report described to date of co-occurrence of these syndromes, this child presented craniofacial features more indicative of Treacher-Collins syndrome. In this study, unlike the only existing description, the Down Syndrome was characterized by a non-inherited translocation, which is responsible for their expression in only 2% of cases,[4] and the Treacher-Collins syndrome likely by high penetrance autosomal dominant inheritance and not by a new mutation. As in other patients with

Treacher-Collins syndrome, the low vision in this patient seems to be associated with eyelid alterations and their consequences (it is not possible to determine with certainty the role of ametropia and strabismus causing amblyopia).

Despite the complexity of genetic malformations, the diagnostic suspicion and referral for appropriate research and monitoring is on the doctor. Considering the importance of the first three years of life in visual development, ophthalmological monitoring should be established early, for better utilization of the residual vision and global development, seeking better quality of life and the adequate socio-economic and cultural inclusion of these patients.

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