

Pierre Robin Syndrome associated with type III familial Duane Retraction Syndrome

Síndrome de Pierre Robin associada à Síndrome de Duane familiar do tipo III

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ABSTRACT

The Pierre Robin Syndrome (PRS) consists of a triad of anomalies characterized by micrognathia, glossoptosis and fissure of the palate, usually associated with other syndromes e occasionally associated with ocular variations. In Duane Retraction Syndrome (DRS), there is a failure in the lateral rectus innervation by the VI cranial nerve, with anomalous innervation of the lateral rectus by fibers of the III nerve. Even though PRS has already been associated with more than 50 other syndromes, there is not any report in literature of association with familial DRS. Thus, this work aims to report a case of this association in a 29 years old patient with recurrence of the syndromes in the family.

Keywords: Ocular retraction syndrome; Pierre Robin syndrome/genetics; Abducens nerve; Eye diseases, hereditary

RESUMO

A síndrome de Pierre Robin (PRS) consiste em uma tríade de anomalias caracterizada por micrognatia, glossoptose e fissura de palato, comumente associada com outras síndromes e ocasionalmente com alterações oculares. Na Síndrome de Duane (DRS), há uma falha na inervação do reto lateral pelo VI nervo, com inervação anômala do reto lateral por fibras do III nervo. Ainda que a PRS já tenha sido associada com mais de 50 outras síndromes, não existe na literatura relato de casos de associação com a DRS familiar. Dessa forma, esse trabalho tem por objetivo relatar um caso dessa associação em um paciente de 29 anos com recorrência das síndromes na família.

Descritores: Síndrome da retração ocular; Síndrome de Pierre Robin/genética; Nervo abducente; Oftalmopatias hereditárias

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Study carried out at Hospital das Clínicas of UFTM.

The authors declare no conflicts of interests.

Received for publication 15/01/2017 - Accepted for publication 28/11/2018.

INTRODUCTION

Duane Retraction Syndrome (DRS) is characterized by a congenital defect in the innervation of the lateral rectus muscle by the cranial nerve VI with reduction of the palpebral fissure, retraction of the ocular globe, and frequent anomalous innervation of the lateral rectus by fibers of nerve III, which may lead to an upshoot or downshoot movement of the eye in the adduction attempt.⁽¹⁾ DRS is almost always bilateral, although the involvement of one eye is often discreet. About 30% of those affected by the syndrome have other congenital defects associated associated such as deafness of perception, speech disorders, alterations in the kidneys, heart and skeleton.⁽²⁾ Usually there is single binocular vision in the primary position, often with face rotation. The eye affected usually has the following motility defects: restricted abduction, restricted adduction, globe retraction in adduction, upshoot or downshoot in adduction, and convergence deficiency. According to Huber⁽³⁾, DRS can be classified as: Type I (most common): absent or limited abduction, normal or discretely limited adduction, ocular parallelism in primary position of the gaze, or small angle esotropia. Type I (less common): limited abduction, normal or discretely limited abduction, ocular parallelism in primary position of the gaze, or small angle esotropia. Type III: limited abduction and adduction, ocular parallelism or small angle esotropia in primary position of the gaze, upshoot or downshoot.

Pierre Robin Syndrome is described in the literature as a triad of abnormalities characterized by micrognathia, glossoptosis, and cleft palate.⁽⁴⁾ Some researchers propose the etiological theory of the intrauterine anomalous position of the fetus during its formation, whereas others believe in genetic factors that would lead to mandibular hypoplasia and oropharyngeal and muscular impairments⁽⁵⁾ due to reports of positive family history, but the genetic factor was not confirmed. There are reports of association between PRS and Moebius Syndrome and Goldenhar Syndrome with no reported cases associated with Duane Retraction Syndrome.⁽⁶⁻⁸⁾

CASE REPORT

Patient F.F., male, 29 years old, with history of right eye (R/L) hypertropia since birth, a constant deviation according to the mother, with first appointment on 07/21/2004. Patient with Pierre Robin Syndrome (micrognathia, cleft palate and glossoptosis), with investigation for genetic syndromes being negative, and with suspected keratoconus. Presence of congenital talipes equinovarus and radial longitudinal deficiency, and delayed neuropsychomotor development. F. F. was born at 8 months and a half of gestation, vaginal birth, neonatal hypoxia, did not cry at birth due to posterior cleft palate and glossoptosis. He underwent surgery when 11 days old to correct the cleft palate. There is also a report of viral meningitis at age 2.5, but without neurological

sequelae. The family history shows 1 uncle with Pierre Robin Syndrome associated with Duane Retraction Syndrome type III (Figures 1 and 2), and 1 cousin with Pierre Robin Syndrome in association with Duane Retraction Syndrome (unspecified type). The mother is highly myopic and has keratoconus.



Figure 1: Downshoot of the LE of the patient's maternal uncle in the clinical case



Figure 2: Patient's maternal uncle presenting Duane Syndrome III in the left eye with increased palpebral slit of the LE at abduction attempt.

Ophthalmological examination (07/21/2004):

Visual acuity: S/C RE fixes poorly up to 10 cm;
LE fixes up to 6 m.

Tonometry: 14/14 mmHg. Eye Fundus Myopic choroidis, normal digging papillae. Presence of vicious head position (VHP) - raising the chin and tilting to the left (Figure 3). Reduction of fissure in the right eye (RE) to adduction, and increased fissure at abduction (Figures 4 and 5). Presence of significant upshoot in the RE at supraduction and adduction attempt of the RE (Figure 6). Abduction restriction and RE adduction. Presence of small deviation L/R in the primary position of the gaze (PPG). Duplications in the left eye (LE) without alterations. In subsequent examinations, downshoot was also observed in the adduction attempt of the RE, and small exotropia (XT) of the RE. Thus, Duane Retraction Syndrome type III (abduction and adduction restriction of the RE) was diagnosed with

Table 1
Exam history of the patient

		RE spherical	RE cylinder	axis	LE spherical	LE cylinder	axis
Static Refraction	21/07/04	-1.25	-0.25	90°	+1.75	-1.25	180°
Dynamic Refraction	17/07/13	-4.00	-1.50	100°	plano	-1.25*	90°
Static Refraction	17/07/13	-4.25	-3.00	100°	+0.75	*	*
Dynamic Refraction	03/08/18	*	*	*	+5.00	-7.50*	100°
Static Refraction	03/08/18	+0.75*	-4.75 *	100°	+2.00	-3.00	115°

* Poor range impairing cylinder check



Figure 3: Patient in infra, PVC with raised limb, with no apparent deviation.



Figure 5: Increased palpebral slit at abduction attempt of the RE.



Figure 4: Decreased RE slit at adduction attempt



Figure 6: Presence of Up-Shoot and slit reduction at adduction attempt of the RE.

Upshoot and Downshoot. The case was dealt expectantly, and no ophthalmologic surgery was carried out. Refraction exams (Table 1) show a poor outcome and difficulty in gauging due to poor cooperation and probable keratoconus.

Ophthalmological examination (02/03/2018):

Visual acuity: S/C RE fixes poorly up to 20 cm;
LE fixes up to 5 m.

DISCUSSION

Pierre Robin syndrome has been associated with more than 50 other cranial syndromes and abnormalities, several of which involving facial and ocular anomalies such as the Stickler Syndrome (COL2A1 or COL11A2 gene on chromosomes 12 and 6), Marshall and gene microdeletion 2q33, causing retinal detachment, congenital cataract, and prominent nose/dental anomalies, respectively. On the other hand, Duane Retraction Syndrome is also associated with anomalies due to alterations on chromosomes 8, 2, 4, 6, 7, 10, 12, 19, 20 and 22 causing alterations in the ear, kidney, heart, upper limbs and skeleton, and the main anomalous locus of DRS is 2q31.1.(2, 5) Thus, it is possible to observe the concomitance between associated syndromes, such as Goldenhar syndrome which was associated with PRS and DRS.^(5,9)

Thus, it is possible to observe some overlap between the chromosomes affected in both syndromes, even with the nearby locus, for example, between the cause of DRS (2q31.1) and the microdeletion of gene 2q33 due to PRS, both in the long arm of chromosome 2. Such factors suggest that an association between DRS and PRS is provável, and an investigation of similar cases is necessary since there are no similar records in the literature, especially in familial cases pointing to genetic correlation.

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