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Superior segmental optic nerve hypoplasia: differential diagnosis with glaucoma

Hipoplasia segmentar superior de nervo óptico: diagnóstico diferencial com glaucoma

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Abstract

Introduction: Congenital changes of the optic nerve are rare. Hypoplasia is the most common form of congenital alteration of the optic nerve. It is believed to be correlated with interruption of fetal development and low birth weight. It presents as a non-progressive anomaly with generally preserved visual acuity. We related a case of a patient with superior segmental hypoplasia with ocular hypertension after corticosteroid use, with a decrease in the nerve fiber layer. Patients with hypoplasia should be followed more closely if they have risk factors for glaucoma and should be considered as a differential diagnosis for normal pressure glaucoma.

Keywords: *Optic nerve hypoplasia; Glaucoma; Congenital optic nerve injury; Diagnosis, differential*

Resumo

As alterações congênitas do nervo óptico são raras. A hipoplasia é a forma mais comum de alteração congênita do nervo óptico. Acredita-se que seja correlacionada à interrupção do desenvolvimento fetal e ao baixo peso ao nascer. Apresenta-se como uma anomalia não progressiva com acuidade visual geralmente preservada. Relatamos um caso de uma paciente com hipoplasia segmentar superior com hipertensão ocular após uso de corticoide, cursando com diminuição da camada de fibras nervosas. Os pacientes portadores de hipoplasia devem ser acompanhados com mais rigor caso tenham fatores de risco para glaucoma e deve ser considerada como um diagnóstico diferencial para o glaucoma de pressão normal.

Descritores: Hipoplasia de nervo óptico; Glaucoma; Lesão congênita de nervo óptico; Diagnóstico diferencial

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Introduction

Congenital changes in the optic nerve are rare abnormalities mostly identified in pediatric examinations - they require proper diagnosis and follow-up. Optic disc coloboma or morning-glory syndrome are examples of congenital optic nerve abnormalities capable of affecting individuals' visual acuity and of leaving sequelae.[1]

Hypoplasia is the most common form of congenital optic nerve abnormality, which can affect one or both eyes. In addition, it can happen alone or in combination with functional and anatomical changes in the central nervous system.[2,3] It is featured by decreased number of axons, reduced optical disc diameter and by changes in individuals' visual field.[2,4]

Assumingly, hypoplasia is associated with dysplasia in the ganglion cell layer and with loss of retinal nerve fiber layer (RNFL) due to interrupted fetal development and low birth weight.[2,4,5] Studies have also suggested that the likely association between growth hormone (GH) deficiency and teratogenic effects of insulin on pregnancy can trigger vascular changes and impair the ganglion cell development process.[5–7] History of paternal ischemic heart disease is also a possible risk factor.[5]

Superior segmental optic nerve hypoplasia (SSONH) is an optic nerve hypoplasia subcategory whose structural dysfunction is limited to the upper region of the optic disc.[4] It is a non-progressive anomaly with overall preserved visual acuity[9] that presents four typical findings: superior entrance of the central retinal artery to the eye, superior optic disc pallor, superior peripapillary scleral halo and superior peripapillary RNFL thinning.[2,5,8] Patients' optical coherence tomography (OCT) and perimetry results may present RNFL thinning in the upper quadrant[8] and lower altitudinal field or sector defects[10], respectively, depending on clinical condition severity.

SSONH epidemiology is controversial. According to some studies, SSONH is often bilateral and mostly affects women,.[2,6] whereas other studies did not report statistical difference between sexes.[5]

Although SSONH is less prevalent than glaucomatous optic neuropathy, it should be taken into consideration in differential diagnosis of normal tension glaucoma (NTG).[10] Thus, the aim of the current study is to present a case report to draw physicians' attention to SSONH, with emphasis on the importance of identifying this rare congenital disorder, which is poorly described in the literature, so these professionals can avoid diagnostic errors and deepen the follow-up of patients presenting hypoplasia and risk factors for glaucoma.

Case report

White female patient, 27 years old, with positive family history of glaucoma, personal history of prematurity and partial growth hormone deficiency. Fundoscopy carried out during pediatric ophthalmology consultation had evidenced optic disc excavation and superior thinning on both eyes (Figure 1), as well as intraocular pressure (IOP) within the normal range during childhood. Perimetry has shown defect in the lower periphery of the right eye. Post-contrast magnetic resonance imaging of the head and orbit was performed; results did not show signs of optical pathway compression or occipital changes, neither ischemic nor demyelinating events.

In 2018, subepithelial infiltrates were diagnosed after adenoviral conjunctivitis and the patient was prescribed 1% topical prednisolone based on a 2-month weekly-weaning regime. Her

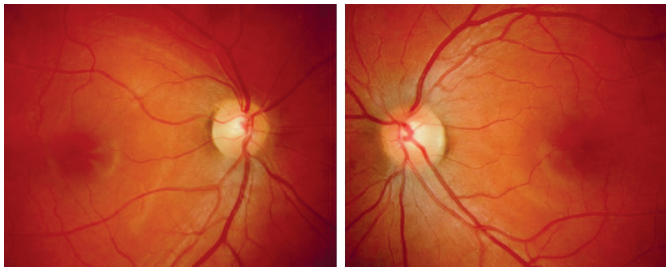

Figure 1: Retinography performed in patient's (A) right and (B) left eyes, in 2017; it shows superior fissure thinning in both eyes and changes in the superior peripapillary NRFL in the right eye.

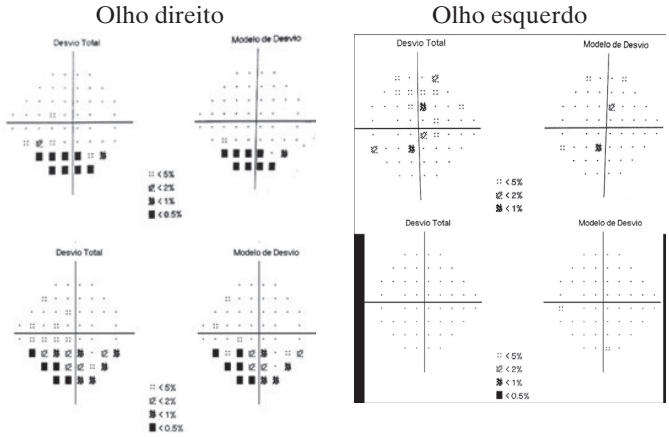

Figure 2: Humphrey perimetry (Carl Zeiss Meditec - HFA II 740) program 24-2 showing defect in lower right eye. The upper image performed in 2017 and the lower image performed in 2019 show increased defect, which suggests lesion progression in the right eye.

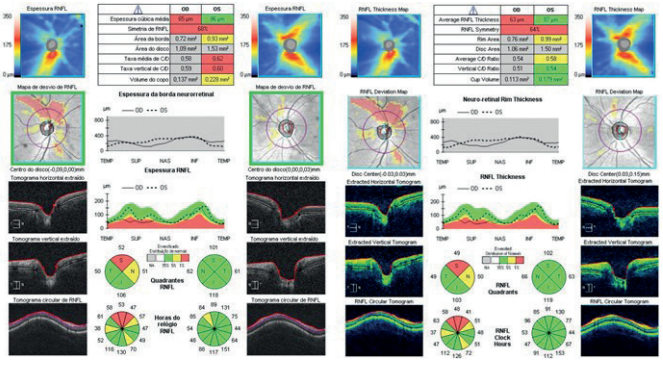

Figure 3: OCT image (Carl Zeiss Meditec - Cirrus) of both eyes. The first image was taken in 2017 and the one next to it was taken in 2019. They show decreased peripapillary nerve fiber layer in right eye quadrants associated with increased lesion on the RNFL deviation map. The ganglion cell complex evaluation also shows worsened density after IOP increase in the right eye.

IOP evolved to 32 mmHg in the right eye and to 31 mmHg in the left eye; it decreased to 14 mmHg in both eyes after she stopped taking the medication - some tests were carried out.

Pachymetry recorded 487 microns in the right eye and 475 microns in the left eye. Perimetry showed defect in the lower right eye (Figure 2) and unchanged left eye. OCT has evidenced localized loss of RNFL in the upper segment of the right eye (FIGURE 3). Patient was subjected to annual follow-up based on perimetry,

retinography and OCT in order to monitor lesion progression.

Discussion

The case of a patient with SSONH, who presented intraocular hypertension associated with the use of topical corticosteroids was reported and led to the hypothetical diagnostic of cortisone glaucoma. Prematurity and partial growth hormone deficiency during childhood were presented as risk factors for SSONH.[5]

Lack of lesion progression is a milestone in hypoplasia featuring processes.[10] Visual perimetry has shown peripheral inferior defect in the right eye; comparison between exams carried out in 2017 and 2019 suggested lesion progression due to intraocular hypertension (Figures 2 and 3).

The current study identified slight superior optic disc pallor and thinning, as well as decreased superior RNFL in the right eye, among the four features described for HSSNO (2,7). Some of these changes could only be diagnosed through OCT. [4]

The OCT carried out in 2017 has shown localized loss of peripapillary nerve fibers in the upper segment of the right eye (52 microns in the upper quadrant) and lack of changes in the left eye. The same examination was repeated in 2019 and it showed decreased peripapillary RNFL in all quadrants (49 microns in the upper quadrant) in the right eye. The contralateral eye did not show variation in peripapillary RNFL, although it underwent similar IOP increase (FIGURE 3). According to studies available in the literature, glaucomatous damages are more likely to progress in optic disc sectors presenting advanced or pre-existing lesions. [11]

Normal tension glaucoma (NTG) is the main differential diagnosis of SSONH. Both NTG and SSONH present IOP lower than 21 mmHg. The main differences between them are shown in Table 1. However, it is worth emphasizing that progression can only happen in NTG cases. [10,12]

So far, patients diagnosed with SSONH did not require additional follow-up tests, because they showed stable optic disc appearance and visual field sensitivity over the years. [9] However, assumingly, SSONH can be a risk factor for NTG or SSONH patients who can be more susceptible to loss of retinal nerve fibers due to high IOP. Therefore, it is necessary conducting RNFL follow-ups and IOP measurements, mainly in patients presenting risk factor for glaucoma.[7]

Table 1
Main differences between normal tension glaucoma and superior segmental optic nerve hypoplasia

	NTG	SSONH
Age	Adult individuals	Children/ young individuals
Sex	Female individuals	Prevalence among female individuals
Laterality	Bilateral	Uni or bilateral
Prevalence	Higher	Lower
Location	Upper temporal	Upper nasal
Disc bleeding	It may happen	It does not happen

NTG - normal tension glaucoma; SSONH – superior segmental optic nerve hypoplasia superior de nervo óptico

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