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Ophthalmological features in Goltz Syndrome

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Introduction

Goltz syndrome (GS) is a rare genetic focal dermal hypoplasia (FDH) due to abnormal mesoectodermal development. It was first identified by Goltz *et al*. in 1962.

It is inherited in an X-linked dominant manner and approximately 90% of females affected are heterozygous or mosaic mutations in PORCN gene.

GS is clinically characterized by skin abnormalities and defects affecting eyes, teeth, skeletal, urinary, gastrointestinal, cardiovascular, and central nervous system.

There are a variety of ocular manifestations in GS that may occur in up to 77% of the reported cases.

Ocular abnormalities described in patients with FDH include anophthalmos, microphthalmos, ectropion, hypertelorism, strabismus, nystagmus, coloboma, aniridia, retinal pigmentary changes, papillomas of the conjunctiva and eyelid, abnormality of lacrimal apparatus, and blocked lacrimal drainage.

Objective

In this present study, we report the ophthalmological features of two patients with typical clinical manifestations of GS.

Methods

The patients were conducted at CAVIVER, a nongovernmental organization (NGO) Institution and referral center dedicated to visually disabled children, at the city of Fortaleza, Brazil.

Ophthalmological evaluation included: assessment of visual acuity, anterior segment evaluation, ocular motility, binocular indirect ophthalmoscopy performed with mydriasis (tropicamide 1%), ultrasound biometry ~~ics~~ and pachymetry.

CASE 1

A six-year-old female with previously diagnosed GS by genetic and clinical study presented with triangular shaped asymmetric facies, atrophic linear skin lesions, following Blashko lines, focal alopecia, lobster claw deformity of the hands, low ear implantation, and dental and orofacial abnormalities. Family history was negative for GS.

Ophthalmologic evaluation disclosed visual acuity of 20/400 in the right eye (RE) and no light perception in the left eye (LE).

Microphthalmia and microcornea were observed in both eyes. In the RE was observed a transparent cornea, aniridia and posterior polar cataract; in LE: opaque cornea that makes it impossible to visualize iris and crystalline.

The horizontal diameter of the cornea in the right eye (RE) was 6 mm and in the left eye (LE), 3 mm. The LE presented a lower eyelid ectropion with symptomatology. The axial length measured in RE was 19 mm and due to extreme microphthalmia, axial length was impossible to obtain in the LE. Eye fundus examination of RE showed an extensive inferior coloboma of retina and optic nerve. Ocular ultrasonography disclosed a choroidal excavation cist in the RE and a total serous retinal detachment in the LE.

Pachymetry revealed corneal thickness of 680 microns in the RE and 580 microns in the LE. The patient was referred to the ocular plastic surgery department to evaluate the LE ectropion and to plan appropriate treatment.

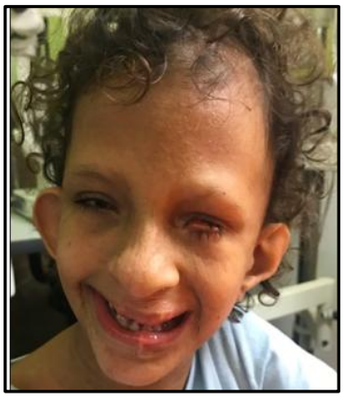

Figure 1: Triangular shaped asymmetric facies with ocular and dental deformities

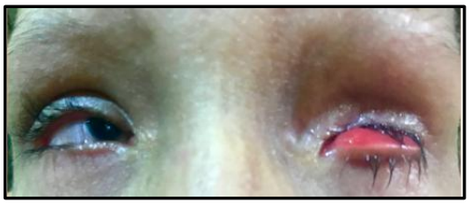

Figure 3: Eyelid ectropium

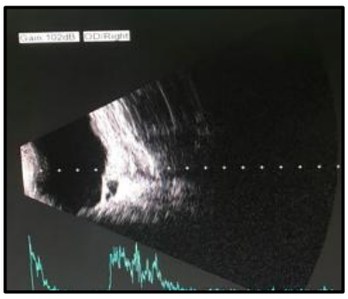

Figure 3:US of the right eye: choroidal excavation cist

CASE 2

A seventeen-year-old female with presumable GS and characteristic asymmetric hyperpigmented atrophic skin lesions at her right side of the face, dental abnormalities and left hand polidactylia was referred to CAVIVER NGO. Ophthalmologic evaluation disclosed visual acuity of no light perception in the RE and 20/60 in the LE , achieving 20/20 vision with the prescription of -1.00 spherical diopters.

Biomicroscopy evaluation in RE showed microcornea (8 mm), nasal iris coloboma, membranous lens with anomalous vasculature and posterior synechiae. Left eye was normal.

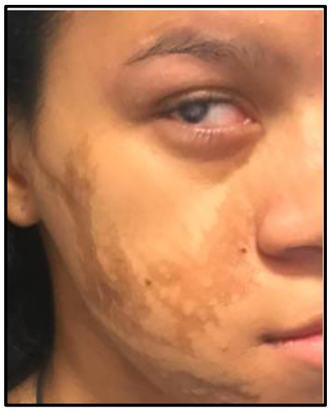

Figure 4: Atrophic skin lesions in the left side of the face

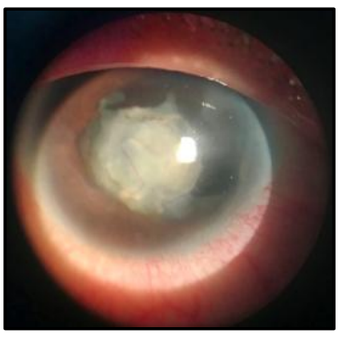

Figure 5: Vascularized and opacified lens with partial iris coloboma in RE

Figure 6: Polodactyly in left hand

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DISCUSSION

Focal dermal hypoplasia, or GS, is a multisystem disorder characterized by involvement of the skin (atrophy and linear pigmentation and herniation of fat through dermal defects occurring along the lines of Blaschko), skeletal system (limb hypoplasia, syndactyly, polydactyly, and oligodactyly), dental (hypoplastic teeth), eyes, and face (facial asymmetry, pointed chin, small underfolded pinnae and notched alae nasi). **[15,16]**.

Ophthalmic manifestations of FDH may occur up to 77% of cases. Ocular colobomas, strabismus and microphthalmia were described as the most frequent manifestations of the syndrome. Other ocular findings reported include anophthalmia, hypertelorism, ectropion, ptosis, nasolacrimal duct obstruction, lid margin or conjunctival papillomas, corneal clouding, blue sclera, aniridia, heterochromia, irregularity of the pupils, cloudiness of the vitreous, and optic nerve hypoplasia **[1,2, 10,11]**.

Multiple vascularized peripheral subepithelial corneal opacifications, retinal neovascularization with recurrent vitreous hemorrhage, and anterior persistent fetal vasculature were also associated with GS **[6,7,8]**.

The differential diagnosis includes incontinentia pigmenti and microphthalmia, dermal aplasia and sclerocornea (MIDAS) syndrome. The clinical history of incontinentia pigmenti includes cutaneous vesiculation and verrucous lesions with hyperpigmentation, which differ from the linear atrophic areas of FDH. A higher proportion of patients with incontinentia pigmenti have convulsions and neurological deficits than those with FDH. MIDAS syndrome presents with microphthalmia, dermal hypoplasia and aplasia, which are limited to the upper half of the body, mainly the face and neck **[8,10]**.

The cases reported here presented microphthalmia, microcornea, cataract, iris and retinal colobomas and lid ectropium.

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Both cases would not profit of a cataract surgery at the moment. No data about cataracts or cataract surgery were described in previous studies related to GS.

One patient could achieve good vision at least in one eye. The other patient has visual impairment and needed visual rehabilitation in order to achieve a better quality of life.

CONCLUSION

Treatment of Goltz syndrome is mainly supportive with a multidisciplinary team. It presents with a high prevalence of ocular abnormalities and ophthalmic evaluation of all children diagnosed with FDH is extremely important.

Ophthalmological assessment includes esthetical management of microphthalmia and surgical correction of lid ectropium if necessary.

Analysis of the refractive errors and glasses prescription may be helpful in improving vision in the affected patients to achieve a better quality of life.

It is important to give advice of preventive protection of the only seeing eye.

Therefore, an early diagnosis of ocular conditions is of great importance in order to achieve the maximum visual potential of these patients with both clinical and surgical interventions if necessary, and visual rehabilitation.

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