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Case Report

Ophthalmological Findings in Goltz Syndrome: A Case Report

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Abstract: Focal dermal hypoplasia, or Goltz Syndrome, is a rare genetic x-linked autosomal dominant disorder. It affects multiple organs and systems, generally unilaterally, with extensive clinical variability. Despite the syndrome's manifestations being predominantly cutaneous, the visual system also suffers characteristic alterations that are worthy of better elucidation. It is a rare disease that is little described in the literature, including in the area of ophthalmology. The objective of this paper is to report on a case we attended to and to discuss existing evidence about this syndrome. EVRS, female sex, 11 months old, referred to the Ophthalmology Service for assessment. Born at 29 weeks, cesarean section, transverse fetal position at birth, weighing 1620g at birth, born with several malformations. Given her phenotypic characteristics, the patient had already been diagnosed as having Goltz syndrome. Ophthalmological examination shows microphthalmia, nystagmus and esotropia in both eyes. Presence of asymmetric corneal opacity, bilateral aniridia and extensive coloboma of the lower retina in the right eye. Opaqueness of cornea and lens in the left eye, making it impossible to see the fundus oculi. The patient remained hospitalized in a neonatal ITU for 4 months from birth, and was discharged in a good overall state, weighing 3370g, receiving diet for gastrostomy. Continues to have multidisciplinary follow-up. This case has reported non-ophthalmological and ophthalmological alterations characteristic of Goltz syndrome, as well as other rare and interesting alterations. Studying them is necessary in medical practice to ensure adequate diagnosis and follow-up of these patients.

Keywords: Focal Dermal Hypoplasia, Ectodermal Dysplasia, Microphthalmos

1. Introduction

Focal dermal hypoplasia, or Goltz Syndrome, is a rare genetic x-linked autosomal dominant disorder. It affects multiple organs and systems, generally unilaterally, with extensive clinical variability [1-6]. After being initially described by Lieberman in 1935, Goltz *et al*. in 1962 [5] and Gorlin *et al*. in 1963 outlined the main characteristics of the syndrome [1]. It mainly affects the female sex, in 90% of cases, and for the most part is lethal for male fetuses [1, 6-7]. Worldwide there are approximately 300 indexed reported cases, although its exact prevalence is unknown [2-3].

Despite the syndrome's manifestations being predominantly cutaneous, the visual system also suffers characteristic alterations that are worthy of better elucidation [1]. Ophthalmic involvement occurs in 40% of cases [4-5]. Ocular coloboma, strabismus and microphthalmia appear to be the most reported manifestations [4-7]. It is a rare disease that is little described in the literature, including in the area of ophthalmology.

The objective of this paper is to report on a case we attended to and to discuss existing evidence about this syndrome.

2. Case Report

EVRS, female sex, 11 months old, from the city of Viamão/RS, Brazil, referred to the Ophthalmology Service for assessment. Born at 29 weeks, cesarean section, transverse fetal position at birth, weighing 1620g at birth, born with

several malformations (Figure 1). Given her phenotypic characteristics, the patient had already been diagnosed as having Goltz syndrome. Mother aged 39, smokes 20 cigarettes a day, no prenatal care.

Non-ophthalmological alterations present since birth include scalp atrophy, cleft lip and palate, ear malformation and low ear position (figure 1A-B), supernumerary left nipple, mammary hypertelorism. Presence of atrophic lesions in the thorax region and lower limbs. Bilateral lobster claw hands and bilateral webbed toes (figure 1C-E). Cardiac auscultation murmur 2+/6+, respiratory system without abnormalities and no visceromegaly. Genitalia with hemiatrophy of the left labium majus.

Ophthalmological examination shows microphthalmia, nystagmus and esotropia in both eyes. Presence of asymmetric corneal opacity, bilateral aniridia and extensive coloboma of the lower retina in the right eye. Opaqueness of cornea and lens in the left eye, making it impossible to see the fundus oculi (figure 1F-H).

Radiological examination demonstrates scoliosis and reduction of cardiac area. Cranial ultrasound provides evidence of dysgenesis/ agenesis of the corpus callosum. Echocardiogram shows interatrial and interventricular communication. Heel prick and female chromosome set are normal.

At one month old, the patient had ocular secretion with *S. aureus*, and received topical ciprofloxacin for 10 days. The patient remained hospitalized in a neonatal ITU for 4 months from birth, and was discharged in a good overall state, weighing 3370g, receiving diet for gastrostomy. Continues to have multidisciplinary follow-up.

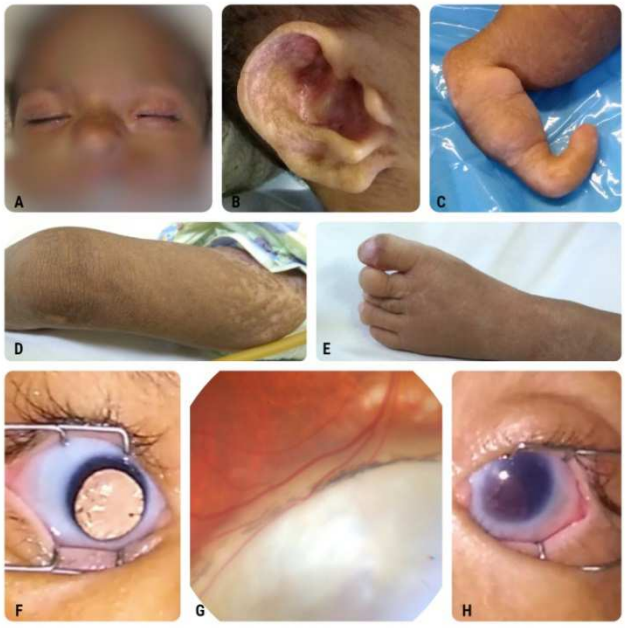

Figure 1. *Goltz Syndrome systemic phenotypic alterations. (A) Ear malformation. (B) Low ear position and ocular hypertelorism. (C) Lobster claw hands. (D) Atrophic lesions to lower limb. (E) Toe malformation. (F) External right eye examination. (G) Coloboma in right eye lower retina. (H) External left eye examination.*

3. Discussion

Focal dermal hypoplasia, or Goltz Syndrome, is a rare genetic x-linked autosomal dominant disorder, with involvement of the mesoderm and ectoderm [1-4, 7]. It affects multiple organs and systems, including the visual system. The syndrome's extensive clinical variability can be justified by earlier or later inactivation of the gene responsible causing it (PORCN gene on the X-chromosome short arm) [1-3, 5-8]. Despite unilateral involvement being more frequent [1], this case reports a patient with bilateral manifestations, including bilateral ophthalmological manifestations.

The main cutaneous alterations reported include telangiectasias and hyper/ hypochromic and/ or achromic atrophic lesions with a reticular pattern. The lesions follow the Blaschko lines [1-3, 5-6] and mainly affect the torso and the limbs [3]. There is a relationship with a significant increase in type III collagen in the dermis and absence of type IV collagen in the basement membrane area [1-2]. Among the adnexa, thin and fragile hair, apocrine gland abnormalities and nail deformities are the main alterations found [1].

The skeletal system is frequently compromised, whereby 60- 80% of cases are affected [3, 8]. There are reports of syndactyly, hypoplasia or shortness of long bones and costovertebral abnormalities [3, 5-6]. Scoliosis is found in approximately 20% of patients [6].

Other systemic manifestations reported include omphalocele, agenesis of the corpus callosum, hydrocephaly and myelomeningocele. Cardiovascular, pulmonary and gastrointestinal tract alterations may also occur [1]. Renal agenesis, renal hypoplasia and horseshoe kidney are genitourinary alterations that can be found [1, 3, 5-6].

Ocular involvement occurs in 40% of cases [4-5]. Depending on clinical variability, acuity can vary widely [2]. Ocular coloboma, strabismus and microphthalmia appear to be the most reported manifestations [4, 7] and were found in the case reported here. Other findings include anophthalmia, hypertelorism, ectropion, ptosis, nasolacrimal duct obstruction, corneal opacity, aniridia, heterochromia, vitreous turbidity, optic nerve hypoplasia, eyelid coloboma and eyelash malformations [4, 8-13]. Aniridia, present in our patient, is an extremely rare ophthalmological finding in this syndrome [5]. With regard to cataracts, cortical and subcapsular cataracts are the most reported [2, 8].

Clinical criteria for diagnosis require the presence of three or more characteristic cutaneous findings and at least one characteristic malformation of the limbs [5, 8]. In view of the diverse phenotypic alterations found in our patient, her clinical diagnosis becomes possible.

Depending on clinical variability, intellectual disability can affect 15- 18% of patients who have the syndrome [5, 14]. Behavioral and emotional alterations are also reported [5].

Reconstructive surgery also plays an important role for these patients, providing better quality of life and social inclusion [1, 4- 6, 15].

4. Conclusion

Detailed systemic and multidisciplinary examination are extremely important in patients with Goltz syndrome. As such, it is possible to diagnose, monitor and treat the different compromised systems. This case has reported non-ophthalmological and ophthalmological alterations characteristic of Goltz syndrome, as well as other rare and interesting alterations. Studying them is necessary in medical practice to ensure adequate diagnosis and follow-up of these patients.

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