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CASE REPORT

Clinical diagnosis of prenatal Goltz syndrome: a case report

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Abstract

This case report describes the follow-up of a child with Goltz Syndrome, a rare mesoectodermal dysplasia, from prenatal diagnosis to current medical care. The diagnosis of the patient and her mother were established during pregnancy, based on skin lesions presented by the mother and ultrasounds that revealed skeletal deformities, omphalocele and other typical characteristics of the syndrome. Post-birth medical follow-up included a series of complementary examinations, such as x-rays and MRIs, which provided a detailed assessment of the neurological anomalies associated with the syndrome. Furthermore, the report highlights the importance of genetic monitoring and a multidisciplinary approach to ensure the best management and quality of life for patients with rare conditions.

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INTRODUCTION

Goltz Syndrome, also known as focal dermal hypoplasia (FDH), is a rare meso-ectodermal dysplasia first described by Liebermann in 1935 as "macular linear atrophoderma and congenital papillomatosis" and later by Goltz in 1962[1]. It has an extremely low incidence, and approximately 200 to 300 cases have been documented. The etiology involves mutations in the PORCN gene on the X chromosome, which is part of the WNT signaling pathway[2].

This condition exhibits an X-linked dominant inheritance pattern, which is lethal in hemizygous males[3]. Approximately 90% of women affected by the syndrome are either heterozygous or have mosaicism concerning a pathogenic variant in the PORCN gene[4].

OBJECTIVES

This study presents a case of GOLTZ-GORLIN Syndrome diagnosed prenatally in 2019 during a pregnancy monitored at a secondary hospital in the Federal District. It also reviews the literature on the syndrome, analyzes the clinical case of the patient in question, and establishes correlations between the case studied and information available in the medical literature.

CASE REPORT

A 4-year-old female preschooler, the first daughter of a non-consanguineous couple, was born to a mother clinically diagnosed with Goltz Syndrome, with no other cases of malformation in the family. The diagnosis was established based on gestational ultrasounds and skin lesions presented by the mother, which were observed during consultations, as depicted in Figure 1. The gestational ultrasounds revealed a fetus with multiple malformations and signs consistent with Goltz Syndrome, including skeletal deformities and omphalocele, among others. Significant diagnostic findings included skeletal dysplasia, omphalocele, and agenesis of the leg and foot [2]. However, these alone were insufficient in this case. The skin lesions exhibited by the mother were crucial in diagnosing the condition. This antenatal medical approach underscores the importance of thorough investigation during pregnancy.

Complementary tests

After birth, during follow-up with the Genetics Service, the patient underwent further confirmatory diagnostic tests



Figure 1. The case report describes the clinical manifestation of Gorlin-Goltz syndrome in the patient's mother. The distinct phenotypic characteristics of the syndrome were observed during a prenatal consultation, enabling the patient's early diagnosis.

(see Figure 2). X-rays revealed congenital skeletal deformities, including partial absence of the tibia and complete absence of the fibula in the right lower limb, as well as congenital amputation at the knee level in the left lower limb. Magnetic resonance imaging of the brain and lumbosacral spine provided a detailed assessment of the neurological anomalies associated with the syndrome, including plagiocephaly, non-closure of parts of the posterior arch of sacral vertebrae, asymmetry of the roots of the cauda equina within the dural sac, and absence of the left piriformis muscle, left obturator internus muscle, and gluteal muscles on the left side, highlighting the complexity of the condition.

The present study was duly submitted to and approved by the Research Ethics Committee. It was registered in *Plataforma Brasil* and assigned Certificate of Submission for Ethical Assessment (CAAE) number 75225023.9.00005553.



Figure 2. Clinical manifestations in the infant with Gorlin-Goltz syndrome described in this case report.

DISCUSSION

Goltz Syndrome, or Focal Dermal Hypoplasia, is a rare meso-ectodermal dysplasia.

A thorough physical examination of the patient and their mother, focusing on specific skin characteristics like cribriform atrophic lesions and bilateral ectrodactyly, was essential for defining the phenotype and starting genetic counseling during pregnancy. A comprehensive interview, which included gathering family history, identified additional diagnostic factors. This strategy could aid in the early detection and management of similar disease cases.

Follow-up with Medical Genetics:

It is imperative to involve the Medical Genetics team to adjust the therapeutic approach and monitor the evolution of the syndrome. Early recognition facilitates effective interventions and may be valuable for future cases with similar presentations. Regular follow-up allows a more comprehensive understanding of the clinical and genetic challenges associated with FDH, contributing to developing more effective management strategies. Specialized follow-up plays a central role in optimizing patient care.

CONCLUSION

The case emphasizes the complex nature of Goltz Syndrome, also known as Focal Dermal Hypoplasia, and the crucial importance of early diagnosis in this rare meso-ectodermal dysplasia. It is important to note that a thorough clinical evaluation of the pregnant mother was vital for the prenatal diagnosis of the patient. The careful physical examination of the mother uncovered subclinical signs of the syndrome, which, along with the clinical context, enhanced the diagnostic suspicion with greater accuracy and speed than laboratory-based methods.

The condition's prenatal identification triggered genetic counseling and the definition of an appropriate care strategy for the mother and newborn. The Medical Genetics team's intervention within a multidisciplinary framework was vital for monitoring, adjusting, and optimizing patient management and improving quality of life.

This case underscores the importance of careful medical surveillance, highlighting the primacy of clinical evaluation in detecting rare genetic syndromes. In the ever-evolving field of medical genetics, it is crucial for health professionals, especially obstetricians and pediatricians, to be well-informed about these conditions to facilitate early and effective diagnosis. Such knowledge is vital to ensure competent care from the beginning of follow-up and to enhance the quality of life for

patients with complex genetic conditions. Ongoing investment in research is essential to broaden our understanding and diagnostic capabilities, enabling clinical practice to progress in anticipating and addressing the needs of these patients more efficiently and humanely.

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