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Dysmorphology

Simpson-Golabi-Behmel Syndrome: A Case Report

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Abstract **Case Report**

Simpson-Golabi-Behmel syndrome (GBS) is a rare, X-linked recessive genetic disorder caused by loss-of-function mutations in the GPC3 (Xq26) gene, with polymorphic clinical presentation and evolutionary profile, characterized by growth accelerated pre- and postnatal, facial dysmorphism and visceral and skeletal malformations, varied and inconstant, as well as an increased tumor risk. We report the case of a 4-month-old infant, the result of a poorly followed pregnancy carried to term, birth weight was 4200g with notion of delayed cry. The parents are non-consanguineous, no similar case in the family. Our patient is followed for hemicorporal tonic-clonic epileptic seizures evolving since birth, controlled by phenobarbital. Clinical examination found psychomotor retardation, height and weight advance, macrocrania, enlarged anterior fontanelle, facial dysmorphism with hypertelorism and obliterated nasal root, supernumerary nipple, umbilical hernia and polydactyly. Examination of the external genitalia reveals cryptorchidism bilateral. A paraclinical assessment was requested to look for associated malformations came back normal. In the light of the literature and through this observation, the authors support the phenotypic, genetic and evolutionary particularities of the syndrome by Simpson-Golabi-Behmel.

Keywords: Simpson Golabi Behmel syndrome-dysmorphism-mutation-GPC3 gene.

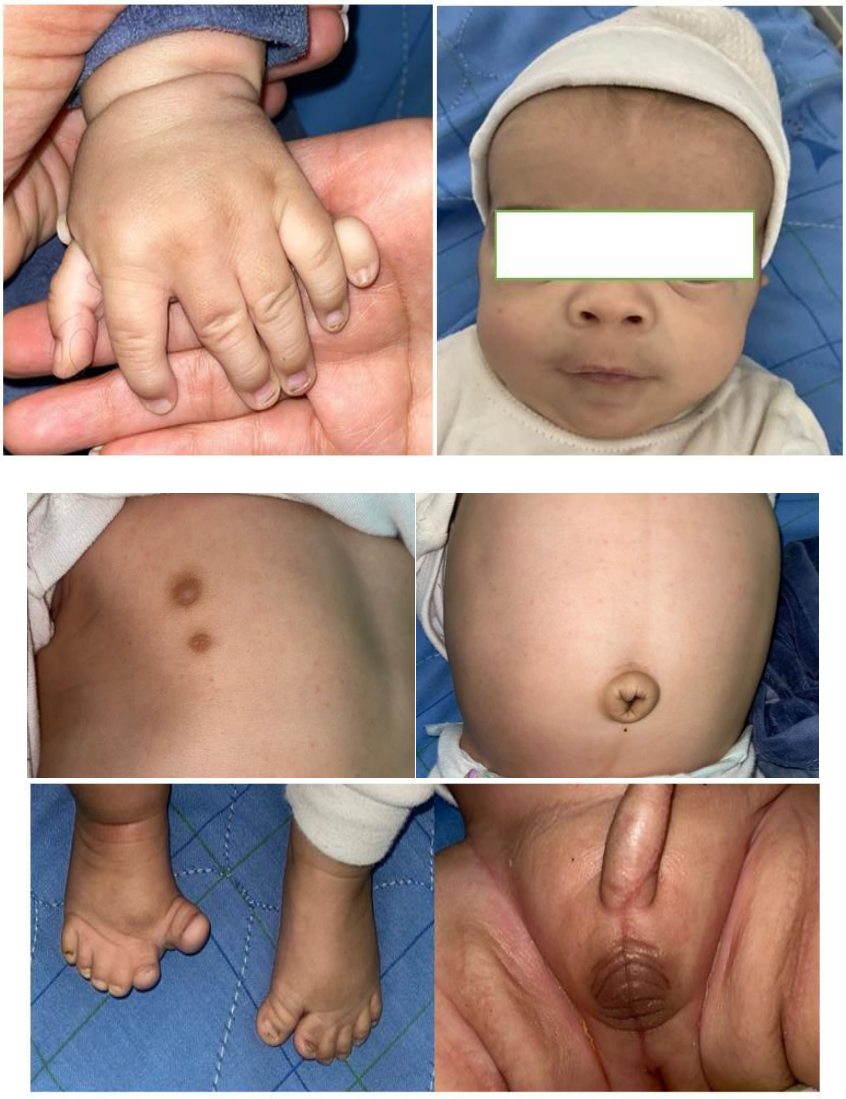
INTRODUCTION

Simpson-Golabi-Behmel syndrome (SGB) is a rare, X-linked recessive genetic disorder due to loss-of-function mutations in the GPC3 gene (Xq26), with a polymorphic clinical presentation and evolutionary profile, characterized by accelerated pre- and postnatal growth, facial dysmorphia, and visceral and skeletal malformations, which are varied and inconsistent, as well as an increased tumor risk.

OBSERVATION

It is about a 4 months old infant, resulting from a badly followed pregnancy carried to term, j1 of a twin pregnancy, male, birth weight was 4200g with notion of delayed cry, J2 (her twin) female with a birth weight of 2400g, notion of immediate cry. The parents are not consanguineous. No similar case in the family. Our patient is followed for tonic-clonic hemicorporeal epileptic seizures evolving since birth, controlled by phenobarbital. Clinical examination found psychomotor retardation, advanced staturo-ponderal, macrocrania, enlarged anterior fontanel, facial dysmorphia with hypertelorism and effaced nasal root, supernumerary nipple, umbilical hernia and polydactyly. Examination of the external genitalia revealed bilateral cryptorchidism. A paraclinical assessment was requested to look for associated malformations came back normal.

A genetic study to look for a mutation of the GPC3 gene was requested.



DISCUSSION

Simpson-Golabi-Behmel syndrome (SGBS) has a broad clinical picture and varying degrees of severity. It is secondary to mutations in the GPC3 gene that result in hyperactivation of Hh signaling, which leads to overgrowth and cancer. It is characterized by pre- and postnatal growth retardation with macrosomia, characteristic dysmorphia (macrocephaly and coarse facial features, macroglossia, hypertelorism, dental malocclusion, palate malformations), supernumerary nipples, congenital heart defects and arrhythmias, segmental vertebral malformations, abdominal visceromegaly, diaphragmatic hernia, umbilical hernia, limb anomalies (polydactyly/brachydactyly, skin syndactyly, nail hypoplasia) and genital involvement (cryptorchidism, hypospadias). Central nervous system involvement results in varying degrees of intellectual disability, motor retardation and language delay. Sleep disturbances have also been reported. Patients with GBS have an increased risk of embryonal tumors (Wilms' tumor), hepatoblastoma, adrenal neuroblastoma, gonadoblastoma and hepatocellular carcinoma.

Diagnosis is based on clinical signs, family history, genetic testing for GPC3 mutations, and array comparative genomic hybridization (aCGH) analysis of genomic imbalance at Xq26.

This variability in clinical presentation is the reason for a multidisciplinary management. It is important to make the diagnosis early in order to search for associated malformations, to ensure regular surveillance given the high predisposition to cancer, and to provide the patient with appropriate genetic counseling to evaluate the risk of transmission to the offspring.

CONCLUSION

The prognosis of this syndrome depends on the associated malformations, the degree of neurological involvement as well as the quality of surveillance given the increased tumor risk.

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