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Simpson Golabi Behmel Syndrome: A New Case and Review of the Literature

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Abstract **Case Report**

Simpson Golabi Behmel Syndrome (SGBS) is a rare syndrome characterized clinically by multiple congenital anomalies, pre and postnatal overgrowth, characteristic craniofacial anomalies, macrocephaly, and organomegaly associated with abnormalities of the skeletal system. On the molecular level, there are genomic rearrangements involving point mutations of the glypican-3 (GPC3) gene at Xq26. The spectrum of signs and symptoms associated with SGBS is wide, ranging from very mild to fatal forms, especially in affected men. We report a rare case of a child affected by SGBS type 1, emphasizing the clinical, paraclinical, therapeutic and monitoring modalities of this possibly serious syndrome.

Keywords: Simpson Golabi Behmel syndrome, genetic study, management, prognosis.

INTRODUCTION

Simpson-Golabi-Behmel syndrome (SGBS) is a rare X-linked genetic disorder characterized by accelerated pre- and postnatal growth, accompanied by variable congenital anomalies. We report a rare case of this syndrome with emphasis on its clinical, para-clinical, genetic, prognostic and therapeutic features.

OBSERVATION

A 12-year-old child from a non-consanguineous marriage consulted for tallness, mental retardation and school difficulties. The clinical examination finds a statural advance: his weight is 43 kg (+1 Standard Deviation (SD)) and his height is 168 cm (between +2 and +3 SD) with macrocephaly, facial dysmorphism made up of hypertelorism, an erased nasal root with a nasal saddle, a macrostomy with a thin upper lip, an everted lower lip with the presence of dental caries (Figure 1). Furthermore, the patient presents with supernumerary breasts (Figure 2), left post-axial polydactyly (Figure 3) and cryptorchidism. The skeletal assessment reveals rib synostosis. Ophthalmological, cardiovascular and abdominal examinations were normal.

The biological and hormonal dosages are unremarkable. The genetic study suggested the diagnosis of SGBS type 1. The molecular study of the GPC3 gene showed that he carries a de novo nonsense mutation (c.271C>T: p.Gln91X), because his mother does not carry this mutation.

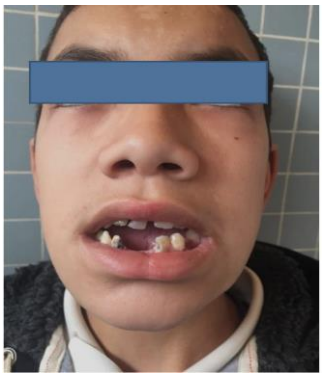

Figure 1: The characteristic face of the child with Simpson Golabi Behmel syndrome

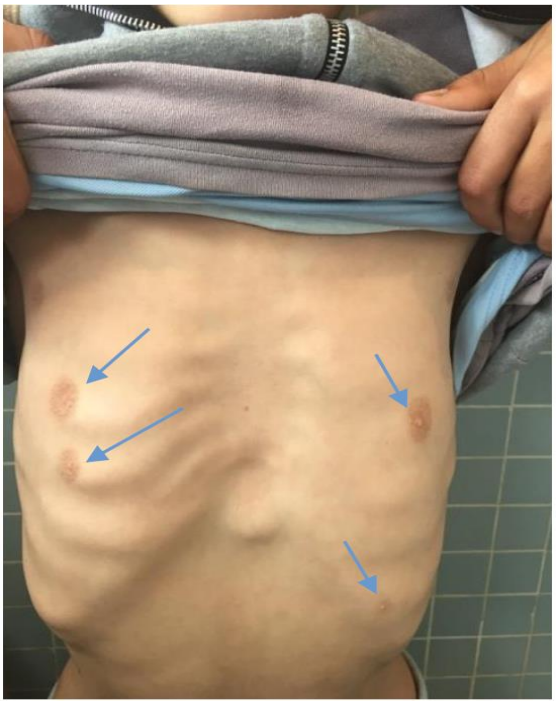

Figure 2: The supernumerary breasts of the patient with Simpson Golabi Behmel syndrome

Figure 3: The hand of the child with Simpson Golabi Behmel syndrome showing polydactyly

DISCUSSION

Simpson Golabi Behmel Syndrome was initially described by Joe Leigh Simpson in 1975. Its prevalence is unknown, it is less common than Beckwith-Wiedemann and Sotos syndromes.

It is a rare syndrome characterized clinically by multiple congenital anomalies, pre- and postnatal overgrowth, characteristic craniofacial anomalies, macrocephaly and organomegaly associated with abnormalities of the skeletal system. The heart, central nervous system, kidneys and gastrointestinal tract may also be affected [1].

The major diagnostic criteria are: overgrowth (macrosomia, macrocephaly and/or pre-and postnatal overgrowth), coarse, characteristic facial appearance, midline defects and tumor predisposition. Other findings

are organomegaly, anomalies of the skeletal system, and congenital malformations of the heart, central nervous system, kidney, and gastrointestinal tract. Intellectual disability of variable degree may be present.

On the molecular level, there are genomic rearrangements involving point mutations of the glypican-3 (GPC3) gene at Xq26. Sometimes these rearrangements intellectual, kidney function and tumor risk (liver tumors, gonadoblastomas and neuroblastoma) [2, 3].

The spectrum of signs and symptoms associated with SSGB is wide, ranging from very mild to fatal forms, especially in affected men. The prognosis and life expectancy depend on the phenotypic spectrum and the severity of the damage. also include the glypican-4 (GPC4) gene. These Glypicans are heparan sulfate proteoglycans, which play an essential role in controlling cell growth and division.

Screening for Wilms tumor and hepatoblastoma with abdominal ultrasound and serum AFP level every three months from time of diagnosis until age four years; renal ultrasound every three months until age seven years; no specific tumor screening protocol has been established for neuroblastoma, gonadoblastoma, or medulloblastoma, but follow up with a cancer predisposition specialist every six months is recommended. Annual ophthalmologic and audiologic evaluations in childhood; sleep study if there are concerns about sleep dysregulation including sleep apnea; routine monitoring of kidney function if renal anomalies are present; evaluation for scoliosis at least annually or during periods of rapid growth; monitoring of serum glucose level in the neonatal period; monitoring of developmental progress at each visit through adolescence [4].

The spectrum of signs and symptoms associated with SGBS is broad, varying from very mild forms in carrier females to infantile lethal forms in affected males. A percentage of affected males die in the newborn period, some of them probably due to heart defects. Carrier females and people with milder cases often live into adulthood. Because of the varying degrees of manifestations and severity associated with the condition, prediction of prognosis and life expectancy most likely varies on an individual basis. Intellectual disability must be carefully evaluated due to the majority of patients have normal intelligence, and do not have the coarse facial and difficulties in speech as we expected for classical SGBS [5].

CONCLUSION

Our observation reports a new case of SGBS type 1. Despite its rarity, this syndrome requires early and, above all, multidisciplinary management in order to take care of all potential complications in time.

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