Gentle giant

Eswaradass Prasanna Venkatesan, Ramadoss Kalidoss, Balakrishnan Ramasamy, Gnanagurusamy Gnanashanmugham

Department of Neurology, PSG Institute Of Medical Sciences and Research, Coimbatore, Tamil Nadu, India

For correspondence:
Dr. Eswaradass Prasanna Venkatesan, Department of Neurology, PSG Institute Of Medical Sciences and Research, Coimbatore, 3, Vijayarahavachari road, Gandhi road, Salem - 636 007, Tamil Nadu, India.
E-mail: eprasanna2k1@gmail.com

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A 17-year-old male presented to us with two episodes of generalized seizures. He was first born child of nonconsanguineous parents. There was no history suggestive of hypoxic injury during child birth. He had delayed cognitive milestones and is diagnosed to have moderate intellectual disability. His siblings and parents were normal. On examination, he was a giant with height of 6'10". He had typical features of gigantism with large hands, large feet, dysmorphic and coarse facies, macrostomia, macroglossia, and congenital nevus over left face involving sclera [Figures 1 and 2]. He also had broad flat nasal bridge with short nose and accessory nipple on right side [Figure 3]. He demonstrated distorted articulation and stereotyped prosody. Further clinical examination revealed no murmurs, organomegaly, or spine abnormalities. There was family history similar illness in his maternal uncle. He was investigated for overgrowth syndromes. His magnetic resonance imaging of brain showed periventricular T2 white matter hyperintensity, but did not reveal any pituitary tumor [Figure 4]. Serum cortisol, thyroid function test, dexamethasone suppression test, and insulin-like growth factor (IGF) levels were normal. Ultrasound abdomen and echocardiogram were normal. Based on characteristic clinical features of coarse facies, supernumerary nipples, gigantism, and probable X-linked inheritance; a diagnosis of Simpson–Golabi–Behmel syndrome (SGBS) was diagnosed.

SGBS was first described by Simpson *et al*.,[1] in two male cousins with coarse face in 1975. It is also known as the "gigantism-dysplasia syndrome". It is an X-linked recessive disorder characterized by pre- and postnatal macrosomia and distinct craniofacial abnormalities.[2] The dysmorphic facies include macrocephaly, coarse facial features, macrostomia, macroglossia, and palatal abnormalities. Other classical features include mild-to-moderate intellectual disability, supernumerary nipples, cardiac defects, and genitourinary and skeletal abnormalities. SGBS is due to loss-of-function mutations of the glypican-3 (GPC3) gene at Xq26, which encodes a heparan sulfate proteoglycan. It plays essential

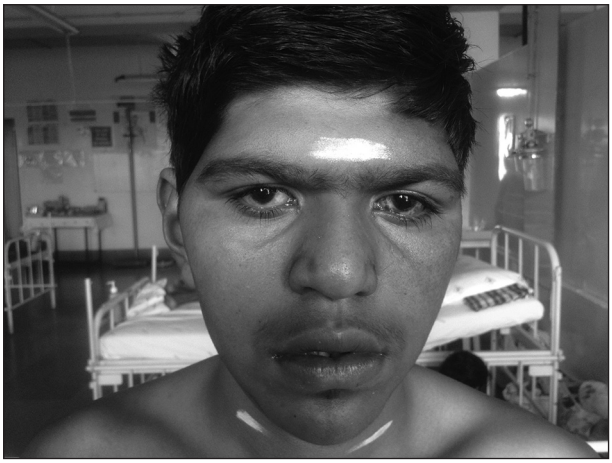

Figure 1: Dysmorphic facies and congenital naevus involving left face and sclera

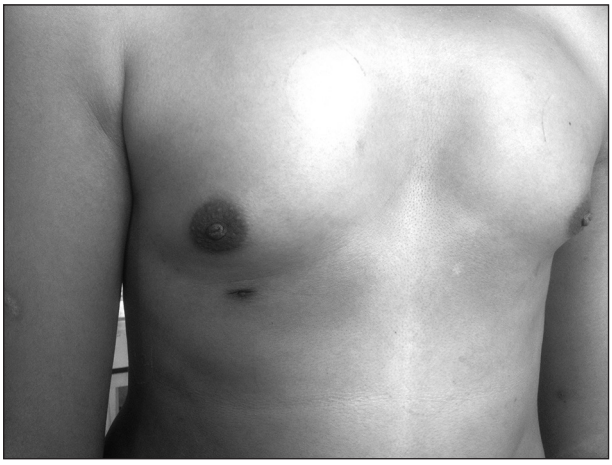

Figure 2: Right accessory nipple

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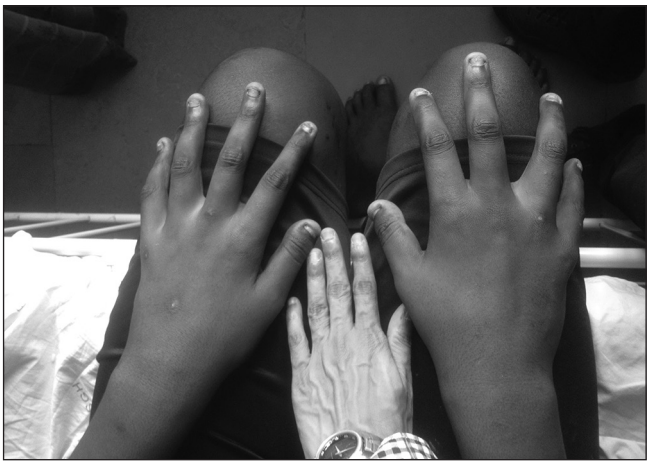

Figure 3: Large hands (acromegaly)

roles in development by modulating cellular responses to growth factors and morphogens.[3] Neonatal hypoglycemia and hypotonia are reported in most cases. There is high risk for embryonic tumors like Wilms tumor and neuroblastoma, and they have to be monitored.[4] SGBS closely mimicks other overgrowth syndromes like Beckwith–Wiedemann syndrome (BWS), Perlman syndrome, and Weaver syndrome. It can be differentiated from BWS by presence of coarse facies, skeletal abnormalities, and absence of omphalocele. Management of these patients involves multidisciplinary approach and close follow-up for malignancies.

The unique feature of our patient was he presented to us with epilepsy, which has not been associated with SGBS in previous reports. In conclusion, SGBS is a rare cause of gigantism which can be diagnosed easily by clinical examination.

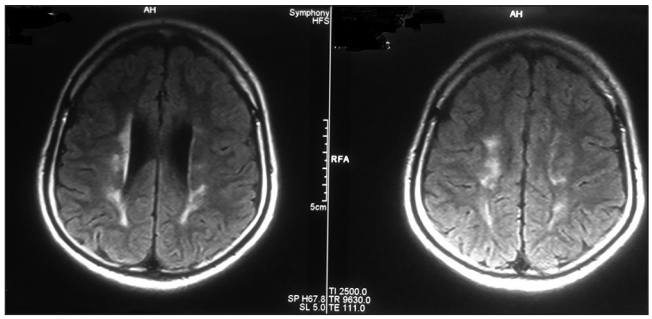

Figure 4: MRI Shows periventricular white matter hyperintensity

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