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Case Report

Shprintzen-Goldberg syndrome presenting as generalised epilepsy in a child: A rare presentation

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Abstract

The Shprintzen–Goldberg syndrome (SGS) is a rare connective tissue disorder characterised by craniosynostosis, distinctive craniofacial features, skeletal abnormalities, marfanoid body habitus, neurological, cardiovascular and intellectual disability. We hereby present a case of a thirteen-year-old Indian child who presented to our clinic with generalised epilepsy from 4 years of age. Child also had intellectual disability, delayed developmental milestones, characteristics facial features, umbilical hernia. As there is paucity of literature showing generalised epilepsy as a part of this syndrome, this case promote to create a high degree of clinical suspicion to diagnose a case of Shprintzen-Goldgerg syndrome in patient of generalised epilepsy present with distinctive craniofacial features along with features of craniosynostosis and marfanoid habitus.

Keywords: Shprintzen-Goldberg syndrome; generalised epilepsy; craniosynostosis, marfanoid habitus.

1. Background

Shprintzen-Goldberg syndrome is a rare connective tissue disorder characterised by craniosynostosis, a marfanoid habitus, distinctive craniofacial features, and skeletal, neurologic, cardiovascular, and connective tissue anomalies. Characteristic craniofacial features include hypertelorism, downslanting palpebral fissures, high-arched palate, micrognathia, and low-set posteriorly rotated ears.[1] In central nervous system commonly reported manifestations include hypotonia, developmental delay, hydrocephalus, ventricular dilatation, and Chiari I malformation and in cardiovascular system, mitral valve prolapse and aortic root dilatation are frequently reported. The most common skeletal manifestations are arachnodactyly, pectus deformity, camptodactyly, scoliosis, and joint hypermobility and inguinal or umbilical hernia.[2-4]

2. Case report

Thirteen years old girl, second in birth order, born to non-consanguineous couple, out of normal vaginal delivery presented to us with complaints of seizures since four years of age and poor school performance. There was no family history of craniosynostosis, musculoskeletal problems, or intellectual disability. She had history of umbilical hernia repair at 3 year of age. On clinical examination her anthropometry parameters are as follows: Weight 38 kg (b/w 25 and 50th centile), Height 143cm (10th centile), HC 55.5 cm, arm span 153 cm. Her facial dysmorphism features (Figure 1) included long face, bifrontal prominence, down slant eyes, squint, bulbous nose, malar hypoplasia and high arched palate. She had a marfanoid body habitus with scoliosis and pectus carinatum along with long and slender fingers and toes and camptodactyly of third, fourth and fifth left fingers (Figure 2a). There was hallus valgus of both great toes and syndactyly of 2nd and 3rd toes. (Figure 2b)

Routine blood investigations, including haemogram, coagulation profile, and biochemical profile were within normal limits. Cerebrospinal fluid analysis was within normal limit. Ultrasound of the abdomen and pelvis and chest roentgenogram was unremarkable while X-ray spine depicted scoliosis (Figure 3a) and X-ray hand suggested camptodactyly and arachnodactyly (Figure 3b). Two dimensional echocardiography showed mild mitral regurgitation and other cardiovascular functions were normal. Magnetic resonance imaging brain suggested Arnold chiarri malformation type1 with mild hydrocephalus (Figure 4). Karyotyping and genetic analysis could not be performed owing to the poor affordability of the parents.

Antiepileptic drugs were started to control seizure and behaviour therapy for intellectual disability. She received regular physiotherapy and occupational therapy. A cardiology opinion for mitral regurgitation was obtained and the patient advised regular follow up. Genetic counselling was advised for the parents.

Figure 1: Facial dysmorphism features - long face, bifrontal prominence, down slant eyes, squint, bulbous nose, malar hypoplasia along with marfanoid body habitus with scoliosis

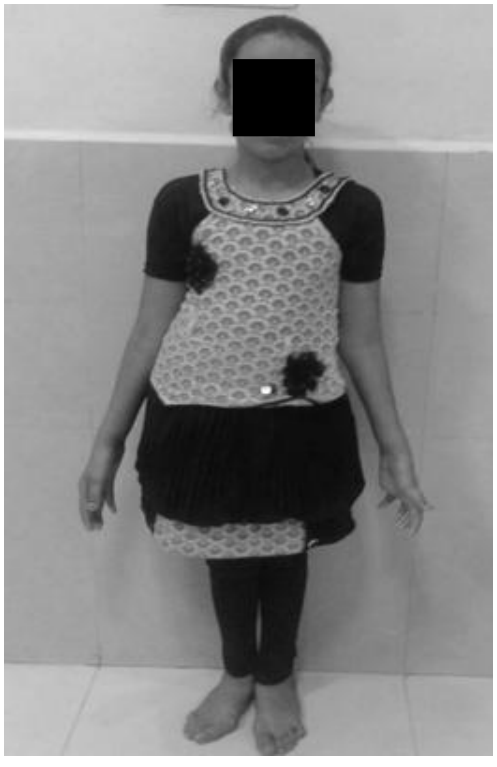


Figure 2a: Camptodactyly of 3rd,4th,5th left fingers

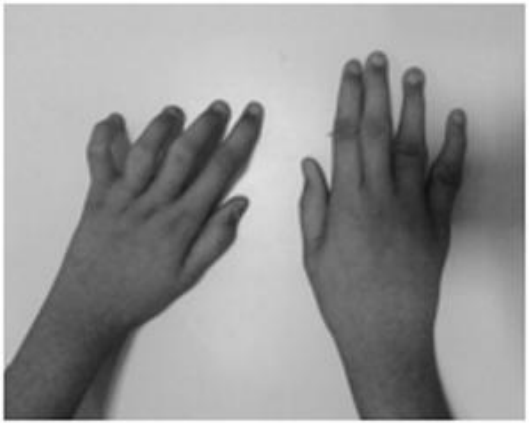


Figure 2b: Hallus valgus of both great toes and syndactyly of 2nd and 3rd toes

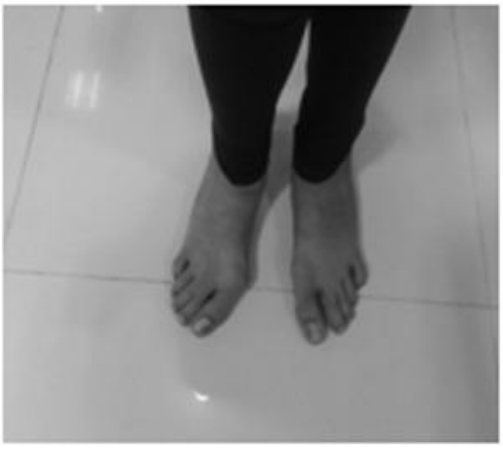


Figure 3a X-Ray Spine showing scoliosis

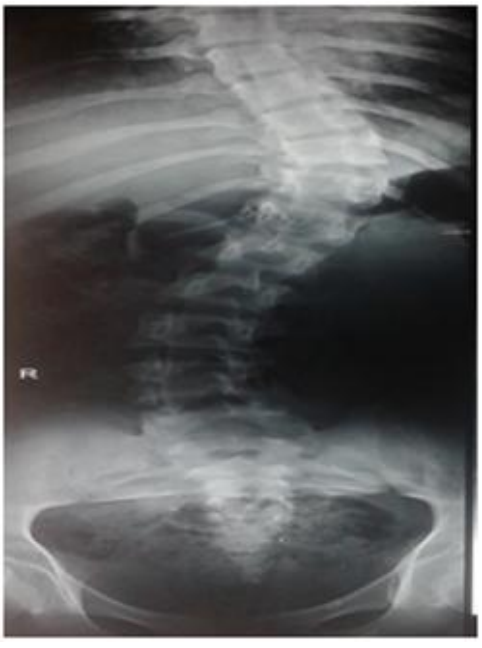


Figure 3b: X-ray hand showing camptodactyly and arachnodactyly

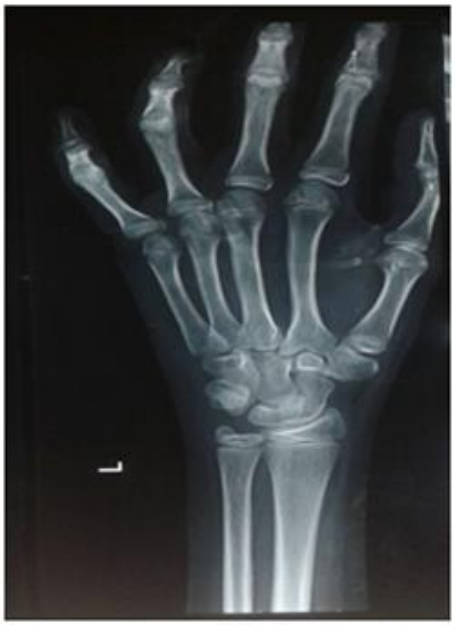
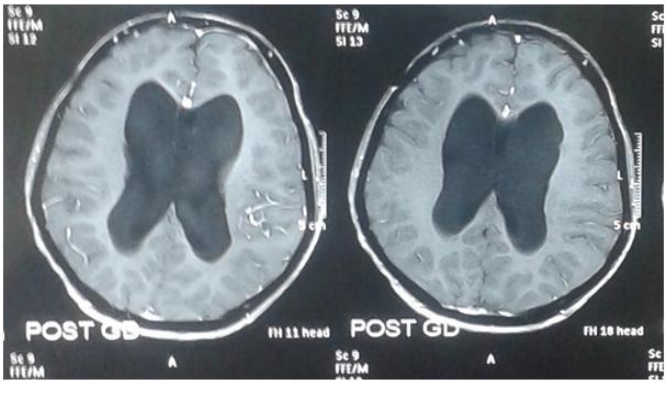


Figure 4: MRI brain suggesting Arnold chiari malformation type1 with mild hydrocephalus



3. Discussion

Shprintzen-Goldberg syndrome also known as craniosynostosis with arachnodactyly and abdominal hernias. [5] This syndrome first described by Sugarman and Vogel [6] and established as a separate clinical entity by Shprintzen and Goldberg in 1982.[4] Till now, only 60 cases of Shprintzen-Goldberg syndrome has been described in medical literature. [5] Our case describes rarity of generalised epilepsy as a presenting symptom of this syndrome. Shprintzen-Goldberg syndrome (SGS), occur due to heterozygous mutation in SKI gene. It is inherited in an autosomal dominant manner. Most individuals with SGS are sporadic cases, having unaffected family members or parents suggesting de novo mutation in affected individual or as a result of germline mosaicism in one of the parents. Our case was also a sporadic case. Mutation in the fibrillin-1 gene (FBN1) with locus in the long arm of chromosome 15 also attribute for few cases.[7] Consistent with symptoms described by Shprintzen and Goldberg [4] and Greally *et al* [5], this child also shared similar craniofacial, skeletal and CNS features.This child is also operated for umblical hernia which is described by Bhushan *et al*. in an Indian child.[8] Aortic root dilatation was described by Carmignac *et al* in SGS but not found in our patient.[9] Differential diagnosis of this syndrome included Marfan syndrome, Loeys-Dietz syndrome, Idaho syndrome-II, Antley-Bixler syndrome, congenital contractural arachnodactyly and several other craniosynostotic syndromes.[10] Management of SGS is best conducted through the coordinated input of a multidisciplinary team of specialists including surgeon, physician, pediatrician, ophthalmologist, cardiologist, clinical geneticist, otorhinolaryngologist, speech and language pathologist, radiologist, physiotherapist. Early detection is essential for prevention of morbidity, mortality and disability.

4. Conclusion

As generalised seizures is very common symptom in children but Shprintzen-Goldberg syndrome is rare entity, this case demonstrates need of high degree of clinical suspicion in a child presented with generalised seizure along with craniosynostosis, distinctive craniofacial and skeletal features supported by radiological findings. This syndrome has variable phenotypes and various systemic abnormalities, a detailed and thorough clinical examination is essential in every child suspicious of SGS syndrome. Genetic testing should be offered to every case, as confirm diagnosis need genetic study and also important for prenatal testing for at risk pregnancy.

Contribution: PM wrote the first draft of manuscript. SS was the consultant in charge of the case.MG helped in detailed diagnosis of case and all authors contributed in critical review of manuscript writing.

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