Shprintzen-Goldberg Syndrome: Case Report

Shprintzen-Goldberg Sendromu: Olgu Sunumu

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Abstract

Shprintzen-Goldberg syndrome is a rare syndrome with craniosynostosis of coronal, sagittal or lambdoidal sutures, dolichocephaly, typical craniofacial features, skeletal abnormalities, scoliosis, joint hyperextensibility or contractures, neurological findings and brain malformations. Fifteen months old male patient with hydrocephalus and dysmorphic facial appearance was referred to our clinic for genetical evaluation. In his dysmorphic examination, the findings were dolichocephaly, prominent forehead and glabella, hypertelorism, prominent eyes, proptosis, depressed nasal root, anteverted nostrils, small nose, low-set and posteriorly rotated, dysplastic ears, microretrognathia, short philtrum, fish mouth of mouth, plump cheeks, high and narrow palate, secondary alveolar arch, pectus carinatum. Hands were small and he had arachnodactyly. Toes were thin. He had also hypotonia and umbilical hernia. With these findings, he was clinically diagnosed as Shprintzen-Goldberg syndrome. This is the first reported case of Shprintzen-Goldberg syndrome from Turkey according to literature review.

Keywords
Shprintzen-Goldberg syndrome, dolichocephaly, craniosynostosis

Öz

Introduction

Shprintzen-Goldberg syndrome (SGS), is a syndrome characterized by typical dysmorphic features, craniosynostosis and marfanoid
features, skeletal and neurological abnormalities. Craniosynostosis, dolichostenomelia, proptosis, low-set ears, arachnodactyly, camptodactyly, pes planus, pectus excavatum or carinatum, scoliosis, joint hyperextensibility or contractures, hydrocephalus, dilatation of lateral ventricles, Chiari 1 malformation may be seen in this syndrome. Cardiovascular anomalies like prolapse of mitral valve, mitral regurgitation, aortic regurgitation and aortic root dilatation may accompany. Minimal increasing of adipose tissue, abdominal wall defects, myopia and cryptorchidism are also rarely seen in SGS. The weight of patients generally tends to increase with age, while their weights are generally below the third percentile (1).

Craniosynostosis is seen in 40% of SGS patients. The anterior fontanel is large and often has a wider secondary alveolar arch. Hydrocephalus is seen in 40% of cases. Umbilical hernia is among the commonly identified findings in patients (1).

Diagnosis is based on clinical and radiological findings (C1-C2 anomalies, large anterior fontanel, thin ribs, square shaped vertebrae and osteopenia) and the SKI gene is known to be only gene associated with this syndrome (1).

Nutritional problems, cyanosis due to respiratory problems, obstructive apnea are frequently seen during the infantile period. The linear growth rate is increasing with age.

Case Report

A 15-month-old male patient was referred to our genetics diagnosis center for genetic screening by the department of pediatric neurology where he was followed up for hydrocephalus.

A healthy 24-year-old father and a 24-year-old healthy mother, who were not consanguineous; told that antenatal follow-ups of the mother regularly performed and developmental retardation was detected in the prenatal follow up of baby. The case was the first pregnancy of the mother and was delivered with spontaneous vaginal birth at the hospital, weighing 2.500 g. The family did not remember birth height and head circumference.

Postnatal physical examination was found to be normal. The patient was taken to the pediatrics department by the family with the complaint of failure to thrive at the age of 2.5 months. After the examination, the child was followed up for further examination and treatment by the department of pediatric neurology and brain surgery due to hydrocephalus.

On physical examination, body weight was 9200 g (10-25 p), height was 72 cm (3-10 p), head circumference was 45 cm (25-50 p). On the dysmorphic examination, dolichocephaly, large anterior fontanel, prominent forehead and glabella, hypertelorism, macrophthalmic-proptotic appearance, flattened nasal root, antevert nostrils, small nose, low, posterior and displaced ears, microretrognatia, short philtrum, fish mouth, high and narrow palate, secondary alveolar arch, pectus carinatum (Figure 1). We found also, his hands were small and associated with arachnodactyly and his toes were thin. Hypotonia and umbilical hernia were present in the patient.

Developmental milestones were retarded. He did not say a meaningful word at the age of 15 months.

In brain magnetic resonance imaging (MRI), cerebellar tonsillar ectopia and supratentorial hydrocephalus (Arnold Chiari syndrome); in cervical-thoracal-lumbar MRI cerebellar tonsillar ectopia and pectus carinatum were detected.

The case was diagnosed as SGS because of dolichocephaly, large anterior fontanel, hypertelorism, ocular proptosis, posteriorly located, low-set ears, microretrognatia, pectus carinatum, umbilical hernia and hydrocephalus.

The patient’s family and ethics committee have been granted permission for this study. There was no statistical analysis in this study.

Figure 1. Dysmorphic appearance of Shprintzen-Goldberg syndrome patient
Discussion

SGS is a syndrome characterized by marfanoid habitus, dolichocephaly, craniosynostosis, mental retardation, ocular proptosis, hypertelorism, down slanting palpebral fissures, strabismus, low-set ears, micrognathia, arachnodactyly, and camptodactyly. Mental retardation is a frequent finding, patients are hypotonic and developmental milestones are retarded. Findings such as thin and sparse hair, Chiari malformation, bifid uvula, choanal atresia/stenosis, vocal cord paralysis, dental malocclusion, aortic root dilatation, mitral valve prolapse, inguinal hernia, hyperelastic skin, hypospadias have also been described less frequently in the patients.

SGS was first presented by Sugarman and Vogel in a 17-year-old boy with plagiocephaly, multiple craniofacial, vertebral and skeletal anomalies, umbilical and inguinal hernia, hypotonia and mental retardation. Shprintzen and Goldberg, 1982, described this syndrome as a separate clinical form (1). In our case, hydrocephalus, dolichocephaly, large anterior fontanel, hypertelorism, ocular proptosis, low-set, posteriorly located ears, microretrognatia, pectus carinatum, umbilical hernia were present.

Phenotypic characters can be variable and differential diagnosis must be done carefully. In a few cases localized fibrillin-1 (FBN1) mutation has been identified in the 15q21.1 region. However, SKI is the only gene which is reported as associated with SGS (1). The E3692A mutation of the FBN1 gene (C1221Y) was described by Kosaki et al. (2) in a Japanese case whom clinical findings were consistent with SGS. Pauliks et al. (3) described complex congenital heart disease in a new SGS patient. Elmistekawy et al. (4) reported the first double-valve surgery in a patient with severe mitral and tricuspid regurgitation with SGS. Pavone et al. (5) in a 16-year-old case, after 12 years of follow-up, was found to have dental malformations among the various clinical features of SGS. Because of the compatibility of clinical findings, we did not have the opportunity to perform molecular analysis of the patient diagnosed with SGS.

Although SGS has typical findings, it has similar clinical features with Loeys-Dietz syndrome (LDS) and Marfan syndrome (MFS). Hypotonia and mental retardation are more common in SGS, but they are rare in LDS and MFS. Some radiologic findings (C1/C2 anomalies, 13 pairs of ribs, square shaped vertebra, Chiari 1 malformation) are more common in SGS than LDS and MFS. Aortic root dilatation is common in LDS and MFS, although it is rare in SGS, it can be a very serious clinical entity (6). Aortic aneurysm is also more common in LDS. Congenital contractual arachnodactyly, fronto-metaphyseal dysplasia, Melnick-Needle syndrome, Idaho syndrome and Antley-Bixler syndrome should also be considered in the differential diagnosis. When all the differential diagnoses are considered, our patient complies with the SGS clinic with dysmorphic features and other clinical findings.

This is the firstly reported case of SGS from Turkey. It is an example in terms of clinically encountered patients because it carries typical clinical findings.

Ethics

Informed Consent: Informed consent form was taken.

Peer-review: Internally peer-reviewed.

Authorship Contributions


Conflict of Interest: No conflict of interest was declared by the authors.

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References