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Case Report

Congenital heart defects and skeletal malformation syndrome with congenital hemiplegia

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ABSTRACT

Congenital heart defects and skeletal malformation syndrome is very rare syndrome. Most of the patients had germline mutations in ABL1 gene. A 30-year-old gentleman presented with history of congenital heart disease (ventricular septal defect) and skeletal malformations which are typical of CHDSKM. Patient also had congenital hemiplegia which is rare in CHDSKMS. Patient also had lactose intolerance since childhood. Patients were evaluated thoroughly to rule out other causes. Current report is one of the rare case reports of CHDSKMS, only few case reports have been published till now.

Keywords: Congenital heart defects, Skeletal malformations, Germline mutations, Congenital hemiplegia

INTRODUCTION

Congenital heart defects and skeletal malformations syndrome (CHDSKM) is characterized by heart defects like ventricular septal defects and aortic root dilation in adulthood, other heart defects can also be present in CHDSKMS are atrial septal defect and patent dutus arteriosis. Skeletal defects are variable in incidence and include pectus excavatum, scoliosis, and finger contractures and some patients may also exhibit joint laxity. Failure to thrive is observed in infancy and childhood.[1]

Clinical features of congenital heart disease and skeletal malformation syndrome includes: scoliosis, failure to thrive, abnormal facial shape, flexioncontractures, ventricular septal defect, abnormality of the skeletal system, short nose, abnormality of cardiovascular system morphology, pectus excavatum, abnormality of the genital system. Other rare features are also reported in CHDSKMS include hearing impairment, lipodystrophy like features, renal hypoplasia, and ocular abnormalities.[1]

This is rare syndrome mainly has autosomal dominant inheritance caused by germline mutation in ABL1 gene.[2,3]

CASE REPORT

A 30 years old male patient presented with, on and off loose stools, increased after taking milk and milk productsand breathlessness of NYHA class 1 since 2 months. Patient does not have history of fever, vomiting and pain abdomen. Patient does not give history of cyanotic spells and recurrent respiratory tract infections.

Past history of congenital heart disease was present, no history of surgery for the same. He had weaknesses of right upper and lower limb noticed by his parents at the age of 2 months. On examination it was observed that; vitals were stable, facial asymmetry in the form of right facial atrophy, elongated face, reduced length of clavicle on right side, pectus excavatum, kyphoscoliosis convexity towards right side, right upper and lower limb shortening and finger contracture (Figure 1).

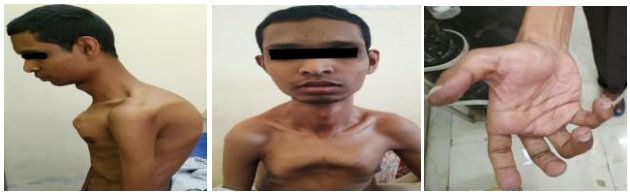

Figure 1: Skeletal malformations.

Patient was evaluated for the cause of chronic diarrhea and also for the malabsorption including stool tests and Colonoscopy found to have lactose intolerance. Precordial bulge, apical impulse was present in sixth ICS in anterior axillary line, hyperdynamic in character. parasternal heave of grade 3, systolic thrill in left parasternal area, pan systolic murmur of grade 4/6, high pitched best heard with diaphragm of stethescope. Functional tricuspid regurgitation present. Loud P2 heard. Wasting of right upper and lower limbs, hypertonia and power 3/5 in right upper and lower limbs. DTR's brisk on right side, plantar bilateral flexor. No abnormality seen in other system examination. Family history was not able to elicit because patient stays in orphanage and lost his parents in early childhood.

Investigations

2D ECHO showed congenital heart disease, situs solitus, large sub aorticventricular septal defect, R>L shunt, moderate aortic regurgitation. CBC RFT and LFT were normal. CT brain showed old infarct with cortical and subcortical gliosis in left fronto parietal and high parietal regions. Genetic analysis was not done. Colonoscopy was normal.

DISCUSSION

Congenital heart defects and skeletal malformations syndrome is characterized by atrial and ventricular septal defectswith aortic root dilation in adulthood. Skeletal defects are variable and include pectus excavatum, scoliosis, and finger contractures, and few patients exhibit joint laxity.[1]

Wang et al studied affected individuals from 4 unrelated families who presented with dysmorphic facial features, congenital heart disease, skeletal abnormalities, joint problems, failure to thrive, gastrointestinal problems, and male genital anomalies.[1] Presentation was different in younger children like dysmorphic features included broad forehead, small nose, deep-set eyes and small chin, whereas in older individuals, the face appeared elongated, with a narrow maxilla, long and narrow nose, and pointed chin. Common skeletal abnormalities which were present in above study were pectus excavatum, scoliosis, finger contractures and hind foot deformity. Congenital heart defects, which were present includes atrial and ventricular septal defect. Joint laxity was present in 3 patients. Our patient had congenital hemiplegia, which is not described in CHDSKMS literature. Till date three variants are reported which are de novo missense variants, c.407C>T (p.Thr136Met), c.746C>T (p.Pro249Leu), and c.1573G>A (p.Val525Met).One more recurrent variant, c.1066G>A (p.Ala356Thr) observed in six patients.[4,5] This is a very rare autosomal dominanant disorder, very few cases reported till now. Genetic analysis couldnot be done due to financial constraints.

CONCLUSION

This article is a rare association of skeletal malformation and congenital heart disease with congenital hemiplegia. This is one of its kind and only fewer reports have been published till date and requires further higher studies in this regard.

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