

## Case Report

# Co-occurrence of syndrome of horizontal gaze palsy with progressive scoliosis with stroke in young: a case report

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## ABSTRACT

Horizontal Gaze Palsy with Progressive Scoliosis (HGPPS) is a rare autosomal recessive syndrome of hind brain maldevelopment associated with ocular and skeletal manifestations. To the best of our knowledge there have been no case reports from India although a family of Indian decent has been reported from Saudi Arabia. The clinical and MRI features along with its co-occurrence with stroke in young will be described in this paper.

**Keywords:** HGPPS, Stroke in young, ROBO 3 gene

## INTRODUCTION

An instance of co-occurrence of two relatively rare two rare neurological entities will be described (HGPPS and stroke in young). Horizontal Gaze Palsy with Progressive Scoliosis (HGPPS) is a rare autosomal recessive disorder probably secondary to mal development of dorsomedial brainstem structures. This disorder is characterized by congenital absence of conjugate horizontal eye movements, progressive scoliosis developing in childhood or adolescence along with intact vertical gaze and convergence. Horizontal pendular nystagmus is another described clinical feature. Mutations in the axon guidance molecule gene, ROBO 3 on chromosome 11q23-q25 have been identified.<sup>1,2</sup>

## CASE REPORT

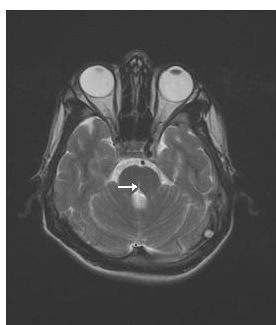
A 21 year old male presented to the casualty with acute onset of right hemiparesis with right facial weakness and slurring of speech of two days duration. He was born at term to consanguineously married parents. He was noted to have thoracolumbar scoliosis at birth which has been

progressing. He was also noted to have absence of conjugate horizontal eye movements by 3 years of age. His past medical history was otherwise uneventful. Patient's younger sibling (18 years old) was also noted to have similar skeletal and ocular manifestations.

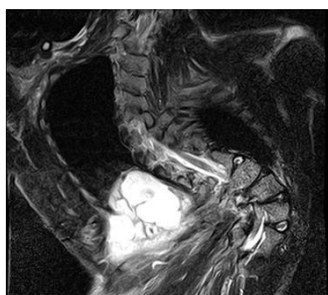
Examination revealed systemic hypertension, thoracolumbar scoliosis, right hemiparesis with facial weakness and right 9<sup>th</sup>, 10<sup>th</sup> and 12<sup>th</sup> cranial nerve involvement. Pendular nystagmus with absent horizontal eye movements was noted. Investigations revealed normal hemogram, lipid profile and renal function tests and transient hyperglycaemia. Echocardiogram revealed asymmetrical septal hypertrophy. Ultrasonogram of abdomen showed right pelviuretric junction obstruction with gross hydronephrosis and multiple calculi. His younger sibling had similar clinical features except those related to stroke and hypertension,

His MRI brain revealed hypoplasia of brainstem, midline cleft in posterior portion of pons (split pons sign) (Figure 1), tent shaped floor of 4<sup>th</sup> ventricle along with rectangular configuration of medulla. These features are said to be characteristic of syndrome of HGPPS.<sup>3</sup> Small

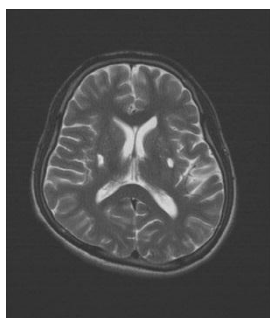
infarcts were noted within bilateral lentiform nuclei and right corona radiata (Figure 2). Spine imaging revealed gross scoliotic deformity of thoracolumbar spine (Figure 3). MRI brain and spine of patient's younger sibling revealed similar findings except for the absence of infarcts. Patient is currently under evaluation for hypertension.



**Figure 1: MRI image showing split pons sign.**



**Figure 2: MRI image showing bilateral basal ganglionic infarcts.**



**Figure 3: Spinal imaging showing gross scoliotic deformity.**

## DISCUSSION

HGPPS is a well-known clinical identity with earliest descriptions being in 1974.<sup>4</sup> This is a rare condition and to the best of our knowledge there are no reports of this syndrome occurring along with stroke in the young. The contributory causes for ischemic infarct could be systemic hypertension and hyperglycaemia. The causative factors for hypertension in this patient are under investigation. It is thought that a failure of decussation of axons of major motor and sensory tracts in hind brain is the main reason for clinical picture of HGPPS.<sup>2</sup> The failure of decussation of axons has been attributed to mutations in the human ROBO 3 gene, a gene homologous to roundabout genes in other species that are important in the regulation of axonal midline crossing during brain development. It is speculative whether the brainstem hypoplasia is one of the contributory factors for development of hypertension in this patient.

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