International Journal of Medical Science and Research
www.medicaljournals.in
Online ISSN: 2664-8768, Print ISSN: 2664-875X
Received: 19-06-2021; Accepted: 02-07-2021; Published: 16-07-2021
Volume 3, Issue 2, 2021, Page No. 01-02

A case report on maroteaux-lamy syndrome type IV with bilateral ventricular dysfunction

Susan Philip, Syama Priya Thampi, Gayatri Suresh, Raju Koneri
Banglore Baptist Hospital, Bellary Road, Hebbala, Bengaluru, Karnataka, India

Abstract
Mucopolysaccharidosis type IV (MPS IV), also known as Morquio syndrome, is a progressive condition which mainly affects the skeleton. Mucopolysaccharidosis type IV (MPS IV) or Morquio syndrome is an autosomal recessive lysosomal storage disease caused by deficiency of. The first signs and symptoms of MPS IV usually become apparent during early childhood. Affected individuals develop various skeletal abnormalities, including short stature, and abnormalities of the ribs, chest, spine, hips, and wrists. People with MPS IV often have joints that are loose and very flexible (hypermobile), but they may also have restricted movement in certain joints. A characteristic feature of this condition is underdevelopment (hypoplasia) of a peg-like bone in the neck called the odontoid process. The odontoid process helps stabilize the spinal bones in the neck. In people with MPS IV, the clear covering of the eye typically becomes cloudy, which can cause vision loss. Some affected individuals have recurrent ear infections and hearing loss. The airway may become narrow in some people with MPS IV, leading to frequent upper respiratory infections and short pauses in breathing during sleep (sleep aponea). Other common features of this condition include mildly thin tooth enamel, multiple cavities, heart valve abnormalities, a mildly enlarged liver (hepatomegaly), and a soft out-pouching around the belly-button (umbilical or lower abdomen (inguinal hernia). Unlike some other types of mucopolysaccharidosis, MPS IV does not affect intelligence.

Keywords: morquio syndrome, arylsulfatase B, peg-like, upper respiratory infections, inguinal hernia

Introduction

Maroteaux-lamy syndrome is a progressive condition that causes many tissues and organs to enlarge and become inflamed or scarred. The rate at which symptoms worsen varies among affected individuals. People with MPS VI generally do not display any features of the condition at birth. They often begin to show signs and symptoms like large head (macrocephaly), build up of fluids in the brain (hydrocephalus), distinctive looking facial features like macroglossia. Affected individuals develop hepatospleenomegaly and umbilical hernia. The airway may become narrow in some individuals leading to sleep apnea. The clear covering of the eye (cornea) typically becomes cloudy which can cause significant vision loss. Unlike other types of mucopolysaccharidosis, MPS VI does not affect intelligence. MPS VI causes skeletal muscle abnormalities, contractures that affect mobility. Individuals with this condition may also have dysostosis multiplex. Many children with MPS VI develops carpal tunnel syndrome and spinal stenosis which compress and damage the spinal cord. The life expectancy depends on the severity of symptoms. Heart diseases and airway obstruction are major causes of death.

Inheritance Pattern

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene.

Case Report

8 year old child who is a known case of MPS VI was admitted with pain in abdomen (mild to moderate in intensity) for 2 days. He had difficulty in breathing from 2 days which was progressively increasing with orthopnea since 1 day. The child had been advised tablet ramipril 2.5 mg OD,tablet lasix 1 tab BD and tablet bosentan. Drug compliance was poor with several doses missed over one week.

Fig 1

Past history

First admission: Admitted 2 years 3 months back with severe pneumonia and bilateral ventricular dysfunction. He required respiratory support with HFNC in PICU .He was worked up for MPS VI in view of corneal clouding on slit lamp examination, coarse faces, short and stubby fingers, dystosis multiplex and normal

Second admission: Admitted 1 year 3 months back for LRTI with cardiac failure. He required respiratory support with HFNC in PICU. Discharged on Lasix ,aldactone, ramipril and silymarin.

Third admission: Admitted with left ear discharge and breathing difficulty. ECHO suggestive of right ventricular dysfunction and increased pulmonary arterial pressures. Discharged on ramipril, bosentan and

Birth history

Term/AGA/LSCS. Birth weight was 3.5 KG. Received Phototherapy on day 5 of life. Delayed developmental history was present.

Family history

Second born to a third degree consanguineously married couple. Elder sibling died on day 20 of life in view of respiratory distress.

Physical examination

The child was in respiratory distress.
Pulse rate – 160/min.
Repiratory rate – 62/min.
Temperature – 96.8 F.
CFT < 3 seconds.
Clubbing, pallor present; short stubby fingers, short neck, widely spaced teeth, flexor contracture of lower limbs were present.

Systemic examination

Respiratory system: Bilateral air entry equal, bilateral NVBS present, bilateral basal crepitations present, SCR present, suprasternal retractions present.
CVS: S1, S2 present, no murmurs present, pericardial rub present.
CNS: Brisk reflexes present, sustained clonus present.

Investigations

2-D echo: mild to moderate left ventricular dysfunction with EF 40%.

Elevated liver enzymes: SGOT/SGPT -1060/1400.
Chest X-RAY: Cardiomegaly.
Hypokalemia

Management in hospital

Patient was started on oxygen, ramipril, furosemide and bosentan. Cardiologist opinion was to discontinue bosentan in view of no signs of pulmonary arterial hypertension.
Dobutimine infusion was started at 5 micro gram/kg/minute. HFNC was stared at 25 L/minute and FIO2 of 5%. Injection dobutimine and HFNC was tapered and stopped. It was advised by cardiologist to continue furosemide 20mg OD and tablet ramipril 2.5 mgOD.

References

1. Schieken RM, Kerber RE, Ionasescu VV, Zellweger H. Cardiac manifestations of the mucopolysaccharidoses. Circulation,1975:52:700-5.
2. McKusick VA, Kaplan D, Wise D, et al. The genetic mucopolysaccharidoses. Medicine (Baltimore),1965:44:445-83.
3. Lewis PW, Raine DN, Kennedy JF. Recognition of the mucopolysaccharidoses by 4 screening tests, including a refinement of the albumin turbidity test, and their differentiation by electrophoretic separation of urinary glycosaminoglycans. Ann Clin Biochem,1974:11:67-71.
4. Scott JE, Darling J. Differential staining of acid glycosaminoglycans (mucopolysaccharides) by Alcian blue in salt solutions. Histochemie, 1965, 221-33
5. Vanace PW, Friedman S, Wagner BM. Mitral stenosis in an atypical case of gargoylism. Circulation,1960:21:80-9.