Original Research Paper

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Anaesthesiology

A RARE CASE OF MARFAN'S SYNDROME POSTED FOR EMERGENCY APPENDICECTOMY

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ABSTRACT Marfan syndrome (MFS) is an autosomal dominant condition with a reported incidence of 1 in 3000 to 5000 individuals. The syndrome is associated with classic ocular, cardiovascular, and musculoskeletal abnormalities, although involvement of the lung, skin, and central nervous system may also occur. Decreased life expectancy occurs primarily due to aortic complications.(1) We are presenting a case of Marfan's syndrome posted for appendicectomy and the risks dealt by us with this multisystem dysfunction.

KEYWORDS : Risks of Anaesthesia, Marfan syndrome, acute appendicitis. appendicectomy

INTRODUCTION

It is an autosomal dominant inheritance mutation in Fibrillin 1 gene located on chromosome 15 with dysregulation of Transforming Growth Factor beta (TGF beta)

Among the triad of skeletal deformity, ocular manifestations, and cardiovascular involvement are usually present in the affected individuals, cardiovascular involvement being the most serious.[2] Aortic dilation and aortic dissection can cause up to 50-70% of deaths in Marfan's syndrome.

There is no specific laboratory test except for molecular genetic testing for the diagnosis of MFS.[3] There is no specific treatment that cures MFS

CASE REPORT

A 20-year-old male was admitted with complaints of acute abdominal pain for 2 days.

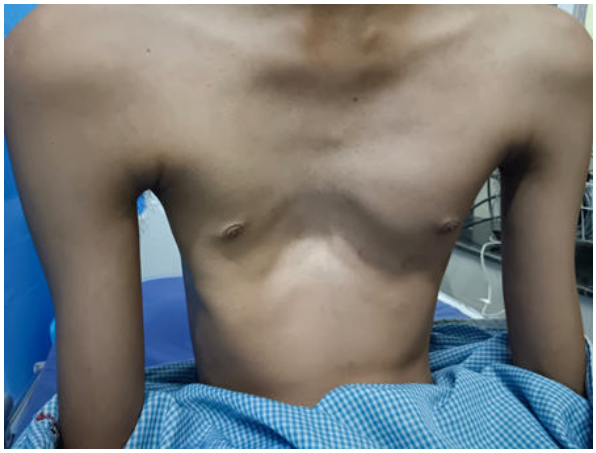
He was diagnosed as Acute Appendicitis

Arachnodactyly
Positive Thumb(Steinberg Sign)
Positive Wrist (Walker Sign)

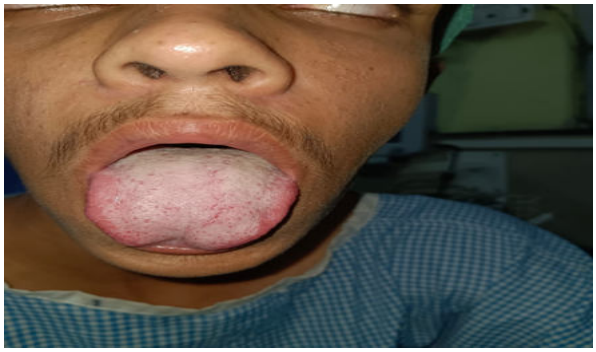
Significant clinical features are

- Height:181cms, Thin (Weight:35kg) BMI:10.74
- Disproportionately long extremities in comparison to the length of the trunk, or dolichostenomelia.
- Arachnodactyly with a positive thumb sign where the entire distal phalanx protrudes beyond the ulnar border of a closed fist.
- A positive wrist sign where the top of the thumb covers the entire fingernail of the fifth finger when wrapped around the contralateral wrist.
- Pectus excavatum.
- Pes planus, or flatfoot deformity (4)
- Micrognothia
- Scoliosis deformity of the thoracic spine
- He is not on any medication.

PECTUS EXCAVATUM

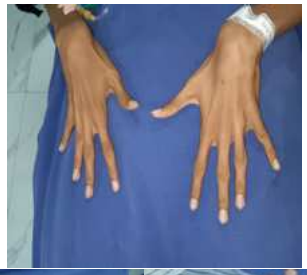


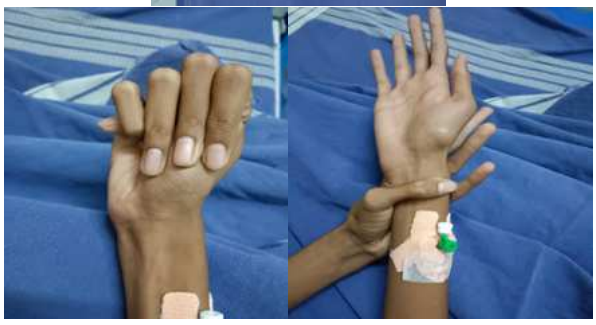
MACROGLOSSIA



MPG CLASS IV

MICROGNATHIA





Positive Thumb(Steinberg Sign) Positive Wrist (Walker Sign)

THORACIC SCOLOIOSIS

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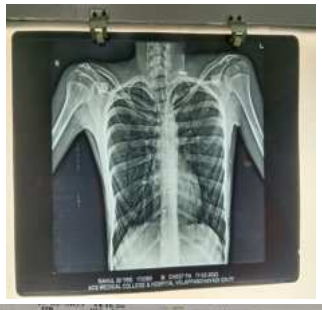

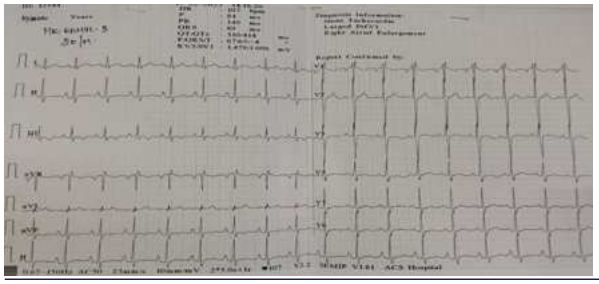

INVESTIGATIONS DONE

- Routine blood count - normal
- Blood sugar – fasting and postprandial
- Renal profile – normal

ECG – suggestive of Normal Sinus Rhythm with T wave inversion in Lead III,avF and V1

2D Echo-Moderate MR,Mild to Moderate AR with Prolapsed Anterior and Posterior Mitral leaflets with LVEF-59%

- Echo – moderate Mitral Regurgitation, Mild to Moderate Aortic regurgitation, Prolapsed Anterior and Posterior Mitral leaflets, Ejection fraction 59%
- Ultrasonography to confirm the clinical diagnosis of Appendicitis

The patient and the parents have explained the existing problems and the risk during anaesthesia and surgery. A high-risk informed consent was obtained

Anaesthetic Technique

It was decided to take up the surgery under spinal anaesthesia.

The patient was preloaded slowly with 15ml/kg of Ringer lactate solution

Anaesthesia work station checklist was done, emergency crash cart was kept ready

Premedicated with Inj. Glycopyrrolate 0.2 mgm and Midazolam 0.1 mgm.

The patient was connected to Monitors – Pulse oximetry, Non Invasive Blood Pressure, ECG, Temperature

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Inj. Bupivacaine 3.5 ml with 25 micrograms of Fentanyl at L2- L3 space. Level of analgesia was T6.

100%Oxygen was supplemented via face mask at a rate of 5L /minute Appendicectomy was performed. There was one episode of hypotension which was corrected with Inj.Phenylephrine 25 microgram.

After the end of surgery, the patient was observed in Post Anaesthesia Care Unit, then shifted to Medical ICU. His postoperative period was smooth and uneventful.

DISCUSSION

'Elastic fibre degeneration', lacking of smooth muscle cells in aorta and mucopolysaccharide deposition in between the cells of the media are the main features of aorta in Marfan syndrome.(5)

Reduced distensibility of aorta, with haemodynamics, occurs. The goals of anaesthetic management of Marfan's syndrome are thus to decrease aortic pressure and to monitor the signs with preparations for possible aortic stenosis (6)

We opted for spinal analgesia, because the patient 's EF was normal, and the vital signs of the patient were normal, in spite of cardiac deformity

Inj.Midazolam a benzodiazepine is used as a premedicant drug. It causes anxiolysis, mild sedation, and antegrade amnesia.

The advantage of adding fentanyl in subarachnoid space increases the duration of analgesia, reduces the requirement of postoperative analgesia, and the MAP was under optimal control.

The goals of the anesthetic management of Marfan syndrome patients are thus first to decrease aortic pressure

The expected life span of Marfan syndrome is only 32 years.

The use of beta-blockers, noninvasive monitoring, restriction of vigorous physical exercise, and elective repair of the aorta has greatly improved the life span.

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