

Successful Delivery in a High Risk Primigravida with Severe Bicuspid Aortic Valve Stenosis with Marfanoid Features: Case Report from a Tertiary Care Centre

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Introduction

- A high-risk case of a primigravida patient with Marfanoid features, contractual arachnodactyly, long arms and legs, breech presentation, bicuspid aortic valve with severe Aortic Stenosis, and abruptio placenta.
- Marfan Syndrome and Marfanoid features pose significant risks to patients. Valvular heart disease increases delivery complications in this patient subset.
- Severe left ventricular outflow obstruction and Marfanoid habitus present challenges during delivery, including heart failure and pulmonary oedema.

Methods

- A 31-year-old primigravida at 31 weeks gestation presented with abdominal pain and vaginal bleeding.
- Clinical examination revealed Marfanoid habitus, crowded maxilla, long arm span, high arched palate, and a systolic murmur in the aortic area.
- Obstetric ultrasound confirmed breech presentation and abruptio placenta with subchorionic bleed.
- Cardiac assessment using 2D Echocardiography indicated bicuspid aortic valve with significant aortic stenosis. No dilatation of ascending aorta or arch was observed. Normal left and right ventricular function with an ejection fraction of 60% was noted.

References: Braverman AC, Bicuspid AV & associated aortic disease; 4th Ed, Otto CM, Bonow RO, 2013, Pg 179

Discussion

- Contractual arachnodactyly is a rare condition affecting multiple body parts, characterized by tall stature, long limbs, and joint contractures.
- Cardiac manifestations may include mitral valve prolapse and the possibility of aortic root dilatation and regurgitant lesions.
- Baseline echocardiogram and trimester-wise monitoring during pregnancy are crucial to detect and manage complications such as aortic root dilatation and valvular lesions.
- Differential diagnosis should include Velo-Cardio-Facial Syndrome (VCFS), ruled out in this case due to the absence of cleft palate or cleft lip.
- This case highlights the successful pregnancy outcome in a patient with congenital heart disease, bicuspid aortic valve, severe aortic stenosis, Marfanoid features, and contractual arachnodactyly.

Conclusions

- Our report underscores the importance of vigilance in identifying rare entities like this high-risk case.
- A baseline echocardiogram and trimester-wise monitoring are crucial to identify and manage potential complications during pregnancy.
- Although these patients may present as Marfanoid, their unique features warrant differential diagnosis, ruling out conditions such as VCFS.
- This case demonstrates the successful management of a patient with congenital heart disease, bicuspid aortic valve, severe aortic stenosis, Marfanoid features, and contractual arachnodactyly during pregnancy.



Results

- Pregnancy-related hemodynamic alterations increased the risk of peri-partum complications in patients with Marfanoid features and bicuspid aortic valve with severe aortic stenosis.
- The patient's antenatal period was uneventful until she presented to the emergency department at 31 weeks gestation with bleeding and abdominal pain.
- A successful Caesarean Section was performed, delivering a male child weighing 1.4kg, who was transferred to the Neonatal Intensive Care Unit.
- The patient demonstrated complete recovery without any adverse events during her hospital stay. Genetic testing confirmed the diagnosis of FBN2 gene missense mutation associated with contractual arachnodactyly, which was received after abstract submission.