MURCS Association: A Rare Association with Patent Ductus Arteriosus and Bicuspid Aortic Valve

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Abstract
An 18 yr old female referred to us as hypertrophic obstructive cardiomyopathy with bicuspid aortic valve. On initial history and examination patient also had primary amenorrhea, differential cyanosis in lower limbs, differential clubbing, polydactyly, syndactyly, high arched foot, pectus carinatum and scoliosis. Oxygen saturation was 94% at room air and complete blood count was suggestive of polycythaemia (Hb 20 g/dl, Hct 60%, Tc-16500, RBC count- 6.29 million /cumm, Platelet count – 1, 88000).

Imaging studies were done, ultrasonography showed absent uterus, absent left kidney, right ectopic kidney. X-ray foot showed 6th metatarsal with phalanx. 2D ECHO was suggestive of Patent Ductus Arteriosus with reversal of shunt with severe aortic stenosis and bicuspid aortic valve.

All these anomalies form part of a syndrome complex called MURCS ASSOCIATION (Mullerian agenesis/aplasia, renal anomalies, and Cervicothoracic Somite deformities).

Introduction
MURCS Association is a type of MRKH (Mayer Rokitansky Kuster Hauser) syndrome.

MRKH syndrome is divided into two types
Type 1 – isolated form
Type 2 – associated with renal, vertebral, auditory and rarely cardiac anomalies known as MURCS ASSOCIATION.

The incidence of MURCS Association is 1:4500 female births being more common than isolated MRKH syndrome. Majority of the cases appear to be sporadic, although familial cases have been reported, the mode of inheritance being autosomal dominant with incomplete penetrance and variable expressivity. MRKH syndrome is characterised by congenital aplasia of uterus and upper 2/3rd of vagina in women showing normal secondary sexual characteristics and normal 46 xx karyotype.

Fig. 1 : Extra metatarsal with phalanx
MURCS Association is characterised by congenital aplasia of uterus and upper 2/3rd of vagina with

- Renal anomalies – unilateral agenesis, ectopia of kidneys or horse shoe shaped kidney.
- Skeletal abnormalities mainly vertebral anomalies – Klippel Feil anomaly, fused vertebra, hemivertebra and scoliosis and digital anomalies like polydactyly, syndactyly.
- Hearing defects and rarely
- Cardiac anomalies – tetralogy of fallot, atrial septal defect, pulmonary valvular stenosis and aortopulmonary window defects.

**Case Report**

An 18 year old female presented to the department of medicine with a history of fever and breathlessness since 4 days. She was diagnosed as having bicuspid aortic valve with hypertrophic cardiomyopathy in 2010. She was born out of a nonconsanguineous marriage, family history being unremarkable. She had primary amenorrhoea with near normal secondary sexual characteristics and normal phenotype. Physical examination revealed multiple malformations including pectus carinatum, scoliosis, polydactyly (Figure 2), syndactyly and high arched foot. Clinical examination revealed differential cyanosis and differential clubbing in lower limbs. Following were oxygen saturation in limbs.

- SpO2 in right upper limb 93 % left upper limb 94 %
- Right lower limb 88 % left lower limb 86 %

**Complete blood count showed**

- HB- 20 g/dl, WBC count 17500, platelet count 1,88 000, RBC Count 6.9 million/cumm, PCV 60%, MCV 86.9 fl, MCH 28.8 pg, MCHC 33.1 G/L which was suggestive of secondary polycythaemia due to chronic hypoxia. Peripheral smear examination showed normocytic normochromic blood picture.

2D ECHO findings were suggestive of large PDA with reversal of shunt (Eisenmenger’s physiology) with bicuspid aortic valve and severe aortic stenosis (Figure 3).

- LV : RvDd 0.74 cm, IVSd 2.32 cm, LVDd 2.66cm, IVSs 2.38cm, LVDs 1.36cm, EF 82%, Biventricular
hypertrophy.

Ultrasonography of the abdomen: absent uterus, absent left kidney, right ectopic kidney.

MRI of abdomen and pelvis showed: absent uterus, absent left kidney, right ectopic kidney and L4 hemic vertebra (Figure 4).

Liver and renal function test were normal. Audiogram was normal. ECG was suggestive of biventricular hypertrophy. Karyotype was normal 46 xx.

X ray of chest s/o cardiomegaly. X ray of dorsal spine s/o scoliosis to right and lumbar scoliosis to left (Figure 5).

X-ray of foot showing 6th metatarsal with phalanx (Figure 1).

Discussion

MURCS Association is a rare syndrome complex with mullerian aplasia, renal anomalies, and cervicothoracic deformities of unknown aetiology with an incidence of 1:4500 female births. Most cases are sporadic; few familial cases with polygenic and multifactorial cause have been noted. MURCS usually presents with primary amenorrhoea however as in this case congenital heart disease or other components of syndrome complex may be the presenting feature. Nevertheless all patients with primary amenorrhoea and absent uterus should be evaluated for MURCS. Once the patient has been told that she cannot conceive due to absence of uterus it is a huge psychological trauma to both patient and family members, counselling of patient and family members is utmost important. Other treatment options include surgical or nonsurgical creation of neovagina, non surgical method being first line of approach should be preferred whenever possible. For infertility, invitro fertilization of her own eggs and use of surrogate mother is an option. In our patient we had to refer the patient to cardiology institute for management of cardiac disease.

Cardiac anomalies are rare in MURCS association and we report this very rare association of PDA and Bicuspid Aortic valve in a patient with MURCS Association. Till date there is no case report of such association.

References