

Prenatal diagnosis of Tetralogy of Fallot and its variants- Case series analysis

Brinda Sabu, Dhanya Shenoy, Vidyalekshmy R

Department of Maternal Fetal Medicine, KIMSHEALTH, Trivandrum, Kerala, INDIA

INTRODUCTION

Tetralogy of Fallot (TOF),the most common cyanotic heart defect(CHD)is characterised by : Ventricular septal defect(VSD),overriding aorta,and pulmonary stenosis in utero. Addition of the outflow tract views in prenatal screening ultrasound (USG) has improved the detection rate to a greater extent. Postnatal outcome depends on the type of surgery and the time at which it is performed. There are 3 types of TOF -TOF with pulmonary stenosis, pulmonary atresia (PA) and absent pulmonary valve (APVS). Though classic forms of TOF tend to be well tolerated, TOF with PA and APVS still have higher perinatal morbidity and mortality.In this setting, the survival, medical care, and developmental disabilities depend on the time of the diagnosis,delay of the treatment,and severity of the CHD. Hence early fetal diagnosis of this treatable CHD reduces the risk of perinatal morbidity and mortality. Here we discuss the three variants of TOF and their associations in a case series analysis.

CASE REPORTS

Case 1: 26-year-old Primigravida,First-trimester aneuploidy screen(FTS)-low risk, at 18+ 6 weeks, on a detailed fetal echocardiogram had the following findings: Situs solitus, left Axis deviation, venoatrial (VA)/atrioventricular(AV) concordance, Perimembranous VSD with overriding Aorta,right Aortic arch,small branched pulmonary arteries-TOF/pulmonary stenosis.

Case 2: 24-year-old Primigravida, 18 + 6 weeks, FTS – LR, had the following echo findings: Situs solitus,VA/AV concordance, main and branched pulmonary arteries not imaged, subaortic VSD with overriding Aorta, presence of Major Aortopulmonary collaterals (MAPCAs),retrograde flow in Ductus arteriosus-TOF/Pulmonary atresia

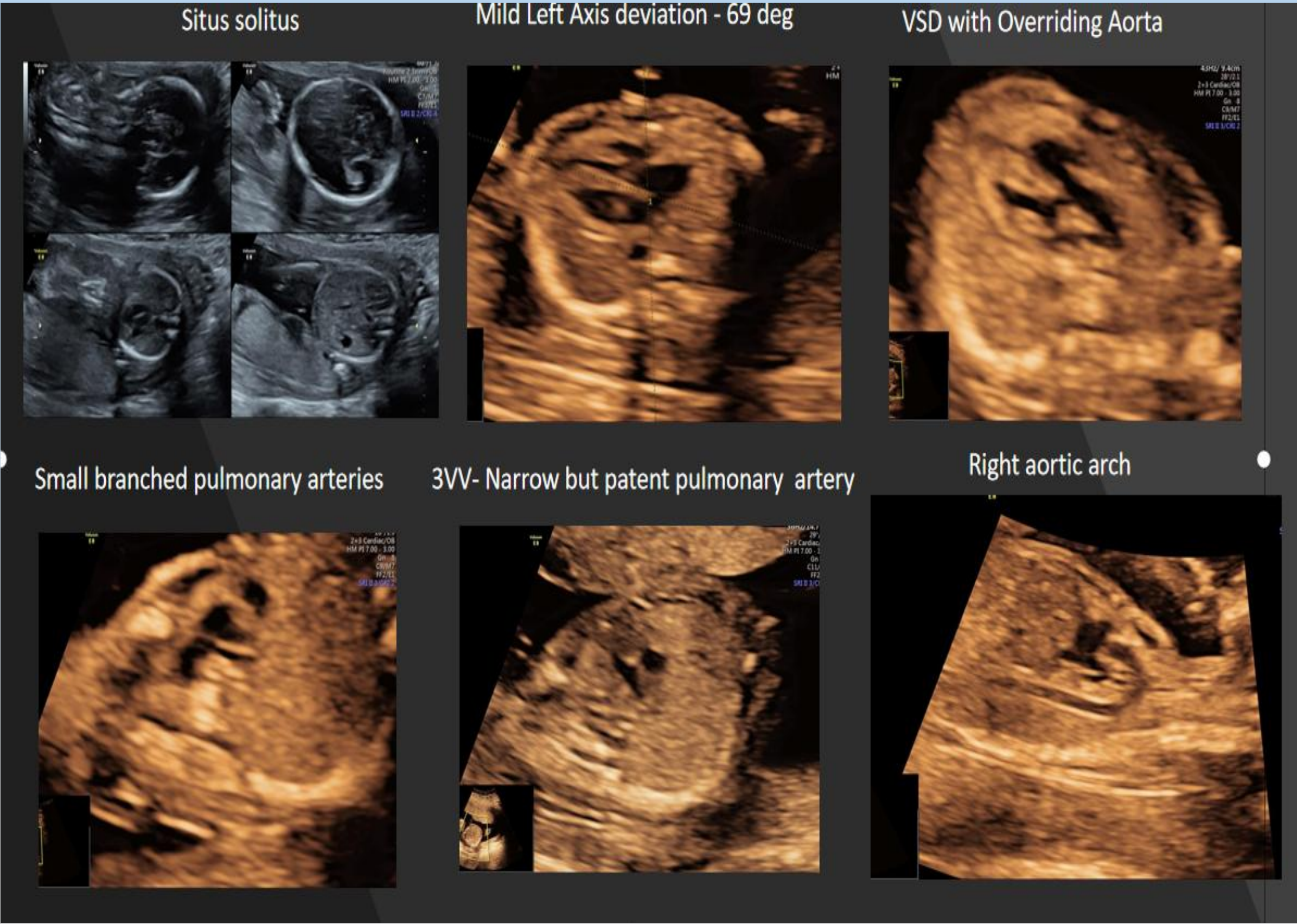
Inspite of genetic and pediatric cardiologist counseling, both parents opted for termination of pregnancies(TOP).

Case 3: 32 years old, G3P1L0, previous baby with Pulmonary atresia and Infant death, Present pregnancy at 34 weeks-Echo findings were: Chamber asymmetry,dilated right ventricle,VSD with overriding aorta,dysplastic Pulmonary valve,massively dilated MPA and branched PA’s,absent ductus arteriosus,high velocity across pulmonary valve annulus (250cm/sec)-TOF/Absent Pulmonary valve syndrome. A male baby weighing 2.2kg was delivered by LSCS.Postnatal ECHO confirmed antenatal findings.He was suspected to have Velocardiofacial syndrome and CMA confirmed the same.Though the baby underwent Pulmonary arterioplasty on D24,he succumbed 2 weeks post-surgery due to multiorgan dysfunction.

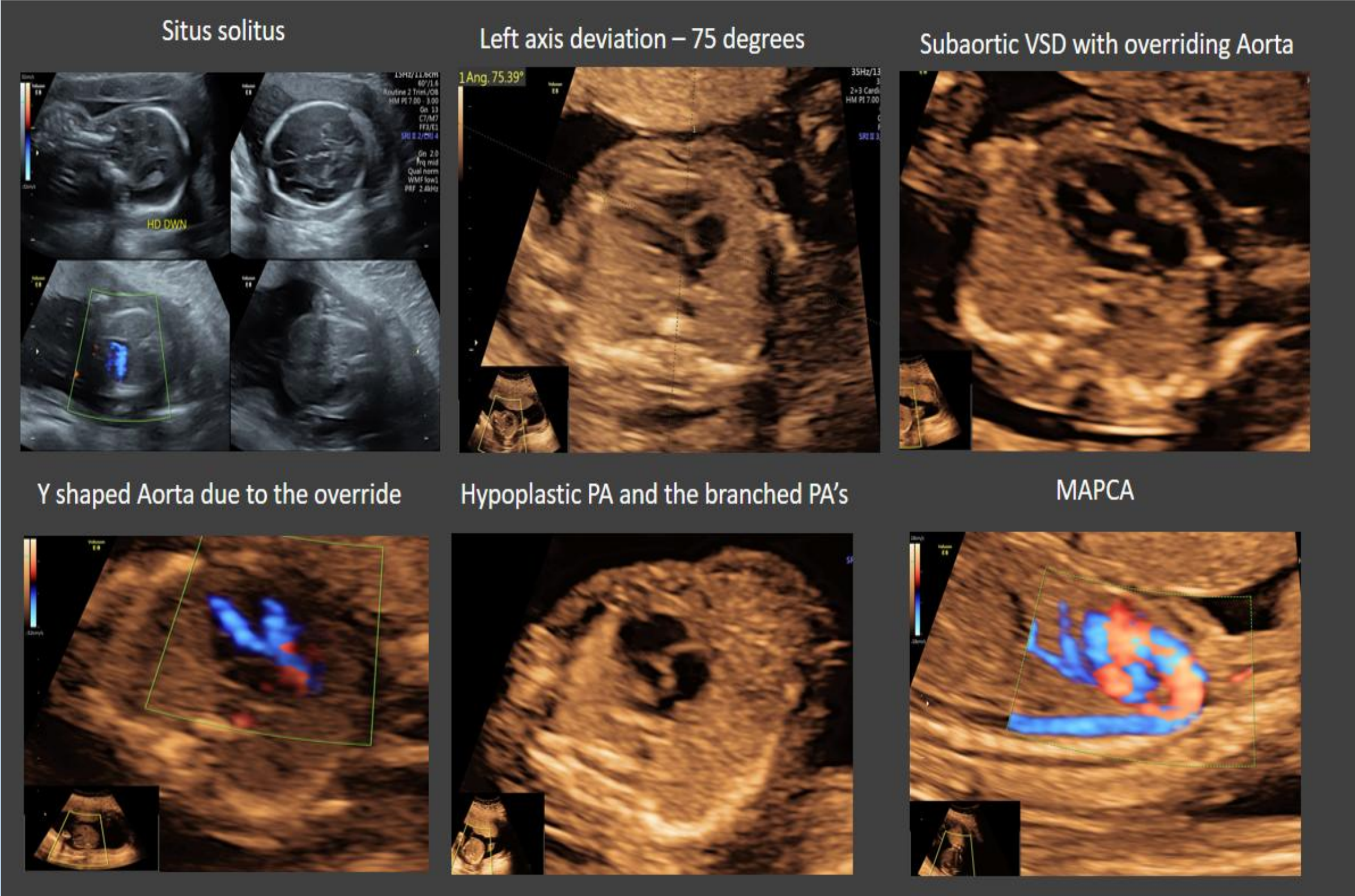
DISCUSSION

TOF is the most common cyanotic heart defect at a frequency of 7% to 10% in children with CHD.Fetal diagnosis of TOF certainly leads to a high rate of TOP because of associated anomalies. Survival rates of TOF with Pulmonary Atresia and APVS is worse than Pulmonary stenosis. Chromosomal anomalies are more commonly associated with Pulmonary stenosis while 22q11 microdeletion is more common in APVS and Pulmonary atresia. Right aortic arch is a/w TOF with Pulmonary Atresia while absent ductus arteriosus is a/w APVS. Detailed cardiac and extracardiac evaluation followed by Invasive testing (Amniocentesis) to rule out chromosomal anomalies and microdeletions should be advised to distinguish Isolated from Complex TOF. Pediatric cardiologist input needs to be obtained for

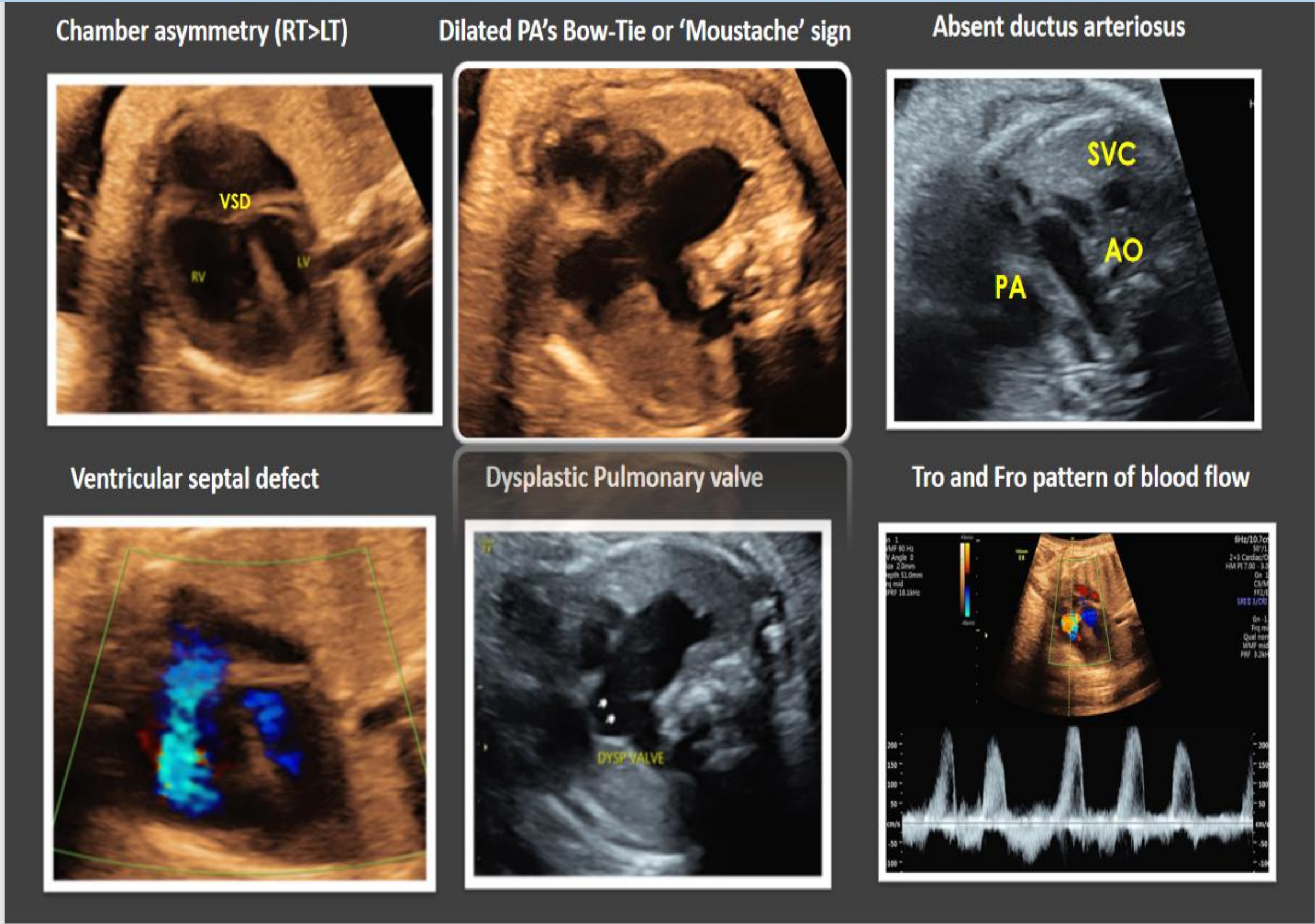
Case 1- TOF with Pulmonary stenosis



Case 2- TOF with Pulmonary atresia



Case 3- TOF with APVS



planning the immediate neonatal magement and long term surgical options.Classic forms of TOF/PS tend to being well tolerated during the neonatal period and the patient may be discharged for follow-up, usually after the first week. In general, the surgical correction is performed in the first year of life, or may require a prior palliative surgical Blalock-Taussig operation. In cases of TOF/PA with pulmonary flow ductal-dependence, prostaglandin infusion should be initiated with a surgical approach soon after birth. The forms of TOF/APVS may evolve with congestive heart failure and higher perinatal mortality.

CONCLUSION

Though prenatal diagnosis of TOF leads to a high rate of TOP because of associated anomalies, isolated TOF with normal chromosomal makeup has good postnatal outcomes. Sonographic findings, genetic testing results and postnatal outcomes are different among subtypes of TOF. Documenting those details at diagnosis can help specialists better counsel their patients. A multidisciplinary team approach involving fetal medicine expert,perinatologist,and pediatric cardiologist/surgeon will help the parents in optimal decision making process and postnatal preparation.

REFERENCES

- 1) Rabih Chaoui,Alfred Abuhamad - A practical guide to fetal echocardiography- 3rd edition
- 2) Deborah S. Fruitman et al-Prenatal assessment of Tetralogy of Fallot: A multicenter prospective cohort study, Progress in Pediatric Cardiology, Volume 60, 2021,

Cardiac views	TOF with Pulmonary stenosis	TOF with Pulmonary atresia	TOF with Absent pulmonary valve
Four chamber view	Normal	Normal	RV dilated
Five chamber view	VSD +Overriding aorta	VSD + Overriding aorta	VSD + Overriding aorta
Pulmonary Artery	Narrow, antegrade flow	Very narrow, or even not visible; retrograde flow	Markedly dilated with to-and-fro flow
Aortic root	Dilated +	Dilated ++	Normal Size
Ductus Arteriosus	Antegrade or retrograde flow	Tortuous with retrograde flow	Typically Absent
MAPCAs	Not Typical	Present in more than 30- 40% of cases	Absent
Prognosis	Good	Guarded	Guarded
Deletion 22q11	10%-15%	20%-25%	30%-40%