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A Rare Case of Congenital Cyanotic Heart Disease in Adult

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Abstract

Congenital heart diseases (CHD) in adults are uncommon. Especially C**yanotic** CHDs (CCHD) in the adult is very rare. We had an adult patient with central cyanosis, pan-digital clubbing and systolic murmur over the left parasternal area. Assuming it to be a case of Tetralogy of Fallot (TOF), being the commonest with these features in adults, we admitted him. On detailed elicitation of the history, perusing his old records, and with fresh investigations, we diagnosed it to be a case of **Double Outlet Right Ventricle (DORV)** with **pulmonary hypertension**. On reviewing the literature, we found that DORV in children has a very poor prognosis; they do not usually survive to an adult unless operated[1]. Moreover, this patient had developed the typical complications "Paradoxical embolism" and "secondary polycythemia". He had multiple cerebral abscesses with recurrent generalized convulsions.

Keywords: DORV, Adult cyanotic heart disease, Paradoxical embolism, secondary polycythemia

Introduction

While the incidence of Rheumatic Valvular Heart diseases is reducing down considerably[2], the Congenital Heart Diseases (CHD) are on the rise. This may be because of the better awareness & availability of advanced diagnostic methods.

CHD in adults is mostly **acyanotic** and the Tetralogy of Fallot's is the commonest cyanotic heart disease. The R to L reversal of shunt, the Eisenmenger syndrome could be the next possibility of Congenital Cyanotic Heart Disease (CCHD) in adults.

Case Summary

Mr. M, aged 21 years a resident of Othakalmandapam, Coimbatore came to our hospital on 08-10-2019 with the c/o left-sided chest pain, lasting for 45 minutes; not typical of classical angina. He gave h/o progressive effort intolerance for the past 10 years.

The parents informed h/o repeated Lower Respiratory Tract (LRTI0 in childhood and bluish discoloration of fingers & toes from the age of 3 years. But denied h/o unconscious spells or squatting during breathless periods. They sought medical attention at the age of 6 years and they were informed that the child had a birth heart disorder, requiring open-heart surgery; but because of the poor nutritional status, he could not be operated. The child was given some medicines and advised to improve general health. Then only at the age of 15, he revisited a hospital. He was again investigated with ECHO, (Report – 1 & Report - 5) and cardiac catheterization.(Report - 2). He was diagnosed as a case of Double Outlet Right Ventricle (DORV), Large-Sized Inlet Ventricular Septal Defect, (VSD) Severe Pulmonary Artery Hypertension (PAH)Because economical restraint and his health condition he was not operated. Within 3 months, he developed a headache and repeated generalized convulsions involving all the 4 limbs. Brain CT (Report -3) scan showed Bilateral Cerebral abscesses/granulomas. MRI. (Report- 4) showed bilateral multiple cystic lesions. Repeated seizure continued and Burr Holing was done. He was admitted in 2018 at our hospital and treated with parenteral antepileptics. There is no h/o any significant illness in the family.

Revising the history, antenatally the fetus showed no abnormality. All the trimesters had been uneventful and anomaly scans showed no abnormalities. There is no h/o radiation or drug intake by the mother. Delivery was Normal vaginal. No congenital abnormality was found

postnatally. No h/o cyanosis or distress at birth and infancy. Development was normal except a little short for age.

Clinical features on admission at our hospital: (08-10-2019)

His height was 154cm and the weight was 53kgs. On General Examination, there was no congenital deformity of the body except for the high arch palate (photo-1). Central Cyanosis (photo-2) was present with Pan-digital Clubbing (photos- 3,4). Pulse: rate 68/min. regular; No special character; accessible in all areas. Blood Pressure 110/70, Respiratory Rate: 23/ min. Cardiovascular System: S1 normal; S2: a single loud P2 over the pulmonary area, grade 2/6 msm was present over

Left Lower Sternal Border (LLSB). Respiratory system, Abdomen. & CNS were normal.

Investigations

Lab. reports done at our hospital (Report - 6) showed - 23.5 Gm/Dl; PCV: 77.3 and RBCcount was 10.9 mill/cu mm.

ECG. (Figure-1) Normal sinus rhythm HR. 64/m. First degree AV Block.(PR interval 0.24); RAD; Right ventricular Hypertrophy. (The tracing inV2,V3,V4 exceeded the paper width so we reduced the gain to 5mm/mV to fit in the ECG paper)

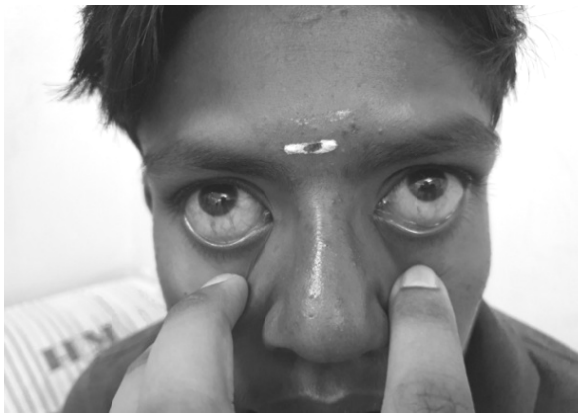

Photo - 1

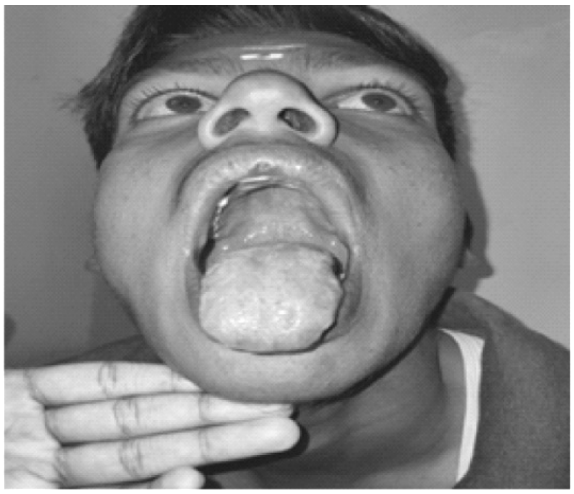

Photo - 2

Central cyanosis with a high arched palate

X-ray chest – (Figure- 2). Hilar congestion, enlarged main pulmonary artery.

ECHOs on 8-10, 9-10-2019 (Figures- 3,4) confirmed DORV with normally related Great Arteries, Large Inlet VSD with Subaortic type, severe PAH. BV functions were good.

To summarize the events

- Milestones:Appropriate for age.
- 3-5 years: H/o repeated Lower RTI infection
- H/o bluish discoloration of fingers &toes at 3-5 years :
- H/o of Grade II/III Dyspnoea
- Medical Attention was sought at 6 years.
- 6-15 years: H/o progressive dyspnoea, malnourishment
- 15 years: H/o Generalised Tonic Conic seizures, Brain Abscess
- Fits controlled with Burr Hole &anti epileptic drugs.
- The patient is on heart failure therapy
- 11-10-2019 - Discharged from our hospital.
- He able to do ordinary physical works;
- He is working in a petrol station

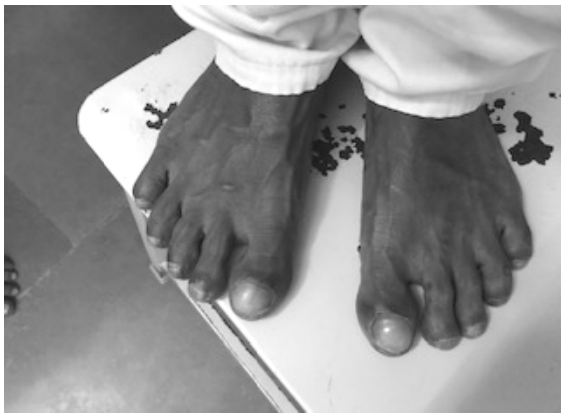

Photo - 3

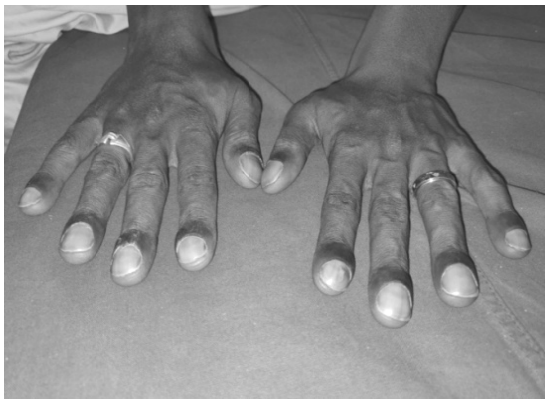

Photo -4

Bilateral Finger clubbing; bluishBilateral Finger clubbing; bluish

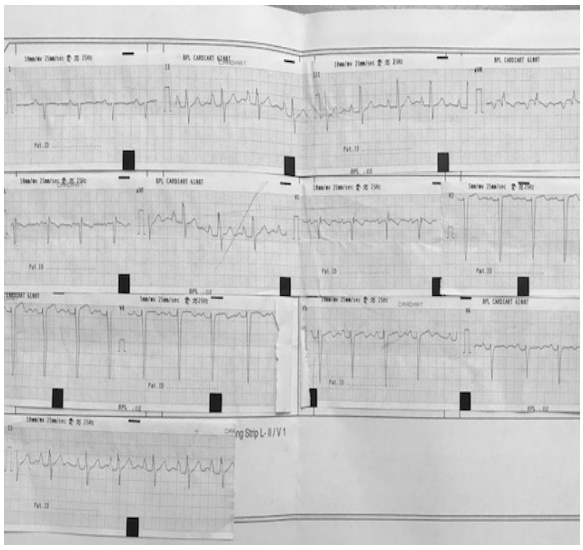

Figure- 1 ECG. 29.01.2020

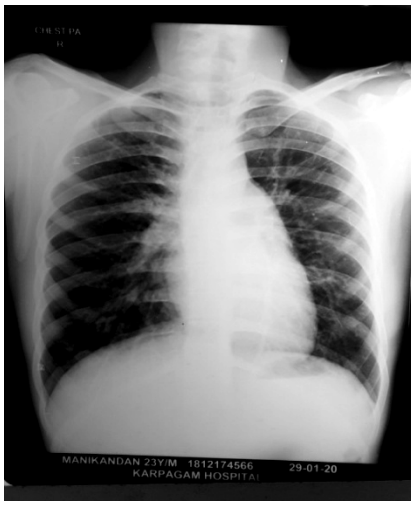

Figure- 2 X-Ray Chest- PA (29-01-2020)

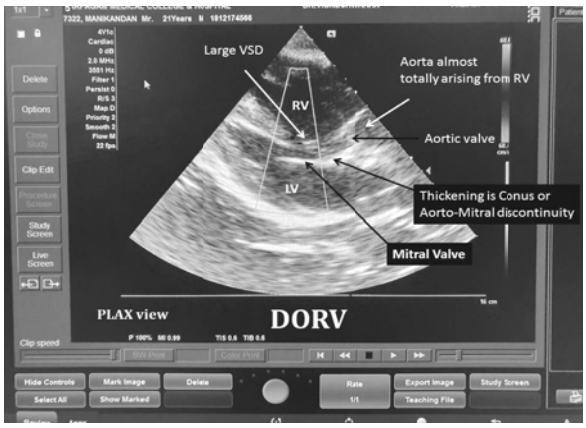

Figure – 3 ECHO Image (08-10-2019)

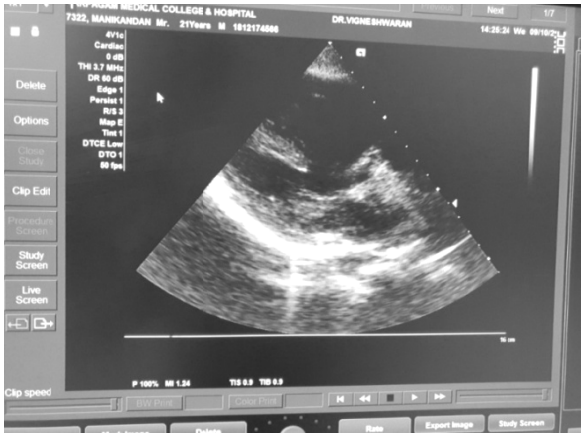

Figure – 4 ECHO Image (09-10-2019)

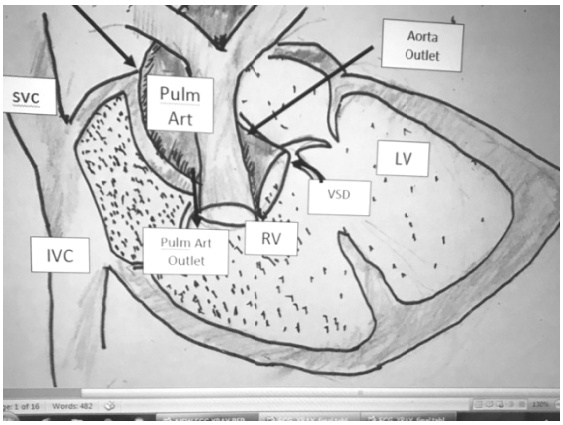

Figure – 5 Diagrammatic Representation of DORV

The double outlet RV, both Aorta &Pulm artery drain RV. The densely dotted area is deoxygenated blood (which also enters systemic circulation- Aorta) and loosely dotted oxygenated blood

Discussion

Double-Outlet Right Ventricle (DORV) — is a rare cyanotic congenital heart disease. The aorta and the pulmonary artery are partially or completely connected to the right ventricle (Figure-5). In one study in India the incidence of DORV is noted as 1.16% of all CHD[3] Only very few cases of DORV who survived over 30 years have been reported; our case is surviving to date (20-02-2020); he is over 22 years. The incidence of CHDs has a prevalence of 1.12/1000 births in India. A large study showed, about 88.5% were the acyanotics, and 11.5% were cyanotic heart patients3. Among the cyanotic heart diseases, Tetralogy of Fallot is the most frequent disease, seen in 48.0% of patients. (Table.1). Sometime DORV is associated with other lesions of the heart. (Table.2)

Table –1Prevalence of Congenital Cyanotic Heart Diseases

TOF is the commonest CCHD 48% of all the CCHD. In one study ref. DORV is 1%

CCHD	%
TOF	48
TGV	27
SINGLE VENTRICLE	07
TAPVC	06
TRICUSPID ATRESIA	02
TRUNCUS ARTERIOUS	09
DORV	01
TOTAL	100

Table-2Possible Associated Anomalies with DORV

Endocardial cushion defects,
Coarctation of the aorta
Mitral valve problems
Pulmonary atresia
Pulmonary valve stenosis
Right-sided aortic arch
Transposition of the great arteries

Pathogenesis

No associated Genetic defect is identified with DORV. Mostly it is sporadic; 22q11 deletion associated with some cases[4]. DORV appears to represent a primitive embryologic condition because of failure to achieve conotruncal rotation and left shift of the conus[5]

A spectrum of conotruncal anomalies which can fail to complete is:

VSD ⟶ TOF ⟶ DORV ⟶ TGA (Transposition of Great Arteries)

Congenitally corrected Transposition of Great Arteries is another rare malformation of this failure of rotation, where central cyanosis is not a feature.[6]

Classification of DORV5.

4 types are depending on the location of the mandatory VSD.

VSD - under the aorta, - with subaortic VSD

VSD - under the pulmonary artery - with subpulmonary VSD

VSD - under both of the great arteries, - with doubly committed VSD

VSD - not near the aorta or the pulmonary artery, - with non-committed, remote VSD

Report –1TRANSTHORACIC ECHO parameters 27-08-2014

Clinical Diagnosis

Situs & Looping	SDD
Systemic Veins	N
Pulmonary Veins	N
Atria	Dilated LA
Atrial Septum	Intact
AV valves	N
Ventricles	Dilated LV
Ventricular septum	Large sized inlet ventricular septal defect (22.0mm)
Outflow tracts	N
Semilunar valves	N
Branch PA	Good sized
Aorta and Aortic arch	Left aortic arch
PDA	No
Coronaries	N

Measurements

Parameter	Absolute (mm)		(mm)	(mm)
AO	27.6			
LA	36.8	Tricuspid Annulus		
RVid		Mitral Annulus		
IVSd	8.6	Aortic Annulus		
LVIDd	31.3	PA annulus		
LVIPWd	9.6	MPA		
IVSs	12.0	RPA		
LViDs	20.7	LPA		
LViPWs	15.4	Aortic Isthmus		
FS %	33	LV mass		
EF %	64	Others		

Of the Cyanotic CHDs in adults Eisenmenger Syndrome, Tetralogy of Fallot, and DORV are encountered. Other conditions like Ebstein Anomaly, Total Anomalous Pulmonary Venous Connection are extremely rare.

Our case is subaortic type VSD which is the Commonest form. The presence of pulmonary Stenosis and transposition of great arteries worsen the prognosis. The aorta is originating from the RV in our case. The difference between DORV and the Tetralogy of Fallot is in the degree of aortic overriding. It is generally accepted that the diagnosis of DORV when at least half of the aorta arises from the right ventricle. If the overriding of the aorta to the right ventricle is less than 50% it is in favor of a diagnosis of TOF and more than 50% is DORV.(50% RULE)[7]There is no pulmonary Stenosis or RV hypertrophy in our case. The prevalence of congenital **cyanotic** heart diseases is given in the **table1**.

Report-2

CARDIAC CATHETERISATION – 04-09-2014

- IMPRESSION:
- DOUBLE OUTLET RIGHT VENTRICLE.
- D-MALPOSED ARTERIES.

- LARGE SIZED INLET VENTRICULAR SEPTAL DEFECT.
- SEVERE PULMONARY HYPERTENSION

Report-3

CT BRAIN: 19.11.2014

IMPRESSION:

- BILATERAL CEREBRAL SOL-ABSCESS / GRANULOMAS
- DIFFUSE CEREBRAL EDEMA.

Report-4

MRI BRAIN: 22/11/2014

Features suggestive of multiple cerebral abscesses with extensive vasogenic edema, especially in the left cerebral hemisphere exerting mass effect on the left lateral ventricle and adjacent sulci.

Dd- tuberculomas since the patient does not have a fever but pyogenic abscesses.

Mild Midline shift to right and mild left uncal & descending transtentorial herniation exerting mass effect on midbrain

Report-5

The Echocardiogram: 19.11.2014

DORV

Normally related great arteries

Large inlet VSD with Sub-aortic extension- BD SHUNT

Confluent dilated branch PAS

Severe PAH

LVEF -65%

Report-6

INVESTIGATION: at our hospital 08-10-2019

SPO2: 84% Room Air & 94% On 2l O2 09-10-2019

LABORATORY: 09-10-2019

Hb - 23.5 G/Dl ; PCV : 77.3 ;

WBC : 6700 CELLS /c.mm. DLC : N-40.7%, L-48.4%, M-10.9%,

RBC- 10.9 mill/cu mm. PLT – 177000 cells/cu mm ;

Blood chemistry RBS – 83.0 mg/Dl ; Urea -25.8 mg/dl, Creat – 1.1 mg/dl, other blood chemistry were normal.

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