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Case Report

VACTERL rare association with rare survival pattern

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ABSTRACT

Truncus Arteriosus (TA) as a cardiac anomaly is a rare occurrence in VACTERL (**V**ertebral defects, **A**norectal malformations, **C**ardiac defects, **T**racheo-**E**sophageal fistula, **R**enal & **L**imb anomalies) association and portends an extremely poor prognosis. The authors report here the case of a 37-year-old male who presented with complaints of recurrent respiratory tract infections and occasional syncopal attacks. The patient was subsequently found to possess an extremely rare combination of VACTERL association with Complex Congenital Cyanotic Heart Disease (Truncus Arteriosus with Right Sided Aortic Arch, Single Coronary Artery, Situs Inversus with Levocardia). The relatively symptom-free survival of the patient into adulthood with this combination of birth defects constitutes a medical rarity and is therefore being presented here.

1. Introduction

Truncus Arteriosus (TA) is an uncommon anomaly, constituting 0.7–1.4% of all congenital heart diseases[1,2] in which a single arterial trunk arises from the ventricle supplying the pulmonary arteries, the coronaries and the systemic circulation. Classified originally by Collett and Edwards to include four subtypes depending upon the origin of the pulmonary artery,[3] an alternative classification system was later developed by Van Praagh and Van Praagh.[4] Patients with TA present with cyanosis and congestive cardiac failure in the neonatal period and survival up to adulthood is rare.[5,6]

VACTERL represents the non-random association of multiple congenital malformations represented by at least three of the following: V = Vertebral, A = Anorectal, C = Cardiac defects, TE = Tracheo-Esophageal fistula, R = Renal & L = Limb anomalies,[7] with a prevalence of 1:10,000 to 1:40,000 births.[8] Ventricular Septal Defect (VSD), Atrial Septal Defect (ASD) and Tetralogy of Fallot (TOF) are the most frequent cardiac defects associated with VACTERL. TA constitutes a rare chance occurrence in VACTERL association and carries a bleak prognosis. Although the maximum survival ever reported for this association is 38 years,[5] untreated, it is extremely rare for such patients to survive into adulthood. The further co-existence of a right sided aortic arch, single coronary artery, situs inversus with levocardia draws the present case into the rarest of rare occurrences in a single surviving adult patient.

We report here such a case of a patient who, having survived to adulthood (37 years) with minimal symptoms, was

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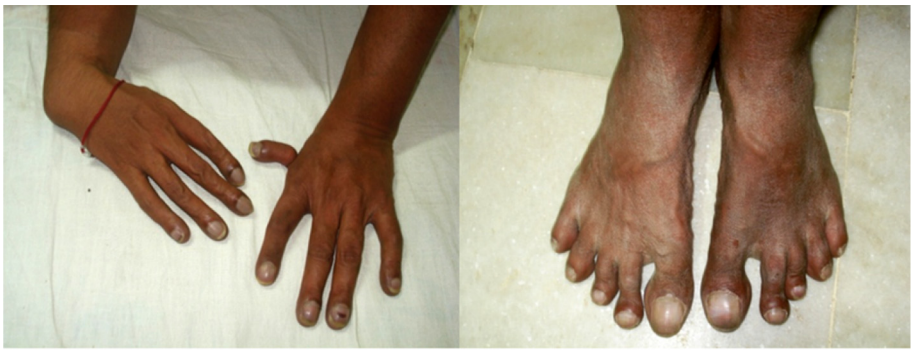

Fig. 1 – Chronic hypoxia induced grade III clubbing of all fingers and toes.

discovered by chance during routine course of investigations, to possess an extremely rare and hitherto fatal combination of congenital anomalies. The case is unique for the extreme rarity of associated anomalies, the unusual presentation and the diagnostic challenge it constituted and is thus being considered worthy of presentation.

2. Case report

A 37-year-old male patient presented to the medicine OPD with complaints of recurrent attacks of respiratory tract infections, intermittent episodes of swelling mostly appearing in the feet and bothersome lower limb pain. The patient also reported having experienced 3–4 episodes of fainting lasting for seconds to minutes with spontaneous recovery of consciousness. The patient's brother had noticed him turning blue during the fainting episodes. The patient, however, was not aware of the same. The patient also seemed unconcerned about the odd shape of his right hand, simply dismissing it as having been there "since birth". Examination was remarkable for presence of grade III clubbing in hands and feet (Fig. 1) and marked central and peripheral cyanosis. The right forearm was dysplastic with an absent thumb. A rudimentary thumb was present in the left hand (Fig. 2). Mild degree of scoliosis was evident in the standing posture. Cardiovascular system examination revealed precordial bulge with visible suprasternal pulsations. A grade IV/VI pansystolic murmur, heard best in the 3rd left intercostal space and radiating to the entire precordium. Blood pressure was normal in both upper and lower limbs. Other system examinations revealed no abnormality.

Blood investigations showed Hb to be 24.4 g/dl with a hematocrit of 76%. X-ray of the hands showed the right radius to be hypoplastic with bowing of ulna and absent 1st metacarpals bilaterally (with absence of corresponding phalanges of 1st metacarpal on the right side) (Fig. 3). A rudimentary left thumb was present. A mild degree of scoliosis was evident in X-ray of the spine. However, no obvious vertebral anomalies could be appreciated. Colour Doppler study of the arterio-–venous system of the hands showed a non-bifurcating right brachial artery. The study of the left arm and lower limbs was unremarkable. USG showed the right kidney to be absent and was remarkable for the presence of a left sided liver with a right sided spleen (Situs inversus). CT scan of the chest and abdomen further corroborated the above findings and demonstrated the presence of a common arterial trunk with right sided aortic arch and the possibility of a separate origin of a single hypoplastic pulmonary artery (PA) (Fig. 4). 2D ECHO with colour Doppler performed subsequently lent further proof to the above showing a large subaortic VSD (20 mm) with more than 50% over-ride of a single arterial trunk taking origin from the left ventricle (Figs. 9 and 10). An atretic PA was seen to take separate origin, being fed by systemic-pulmonary collaterals and a single large MAPCA (Figs. 11 and 12). (Truncus Arteriosus type IV – Collett & Edwards/Pseudo truncus). The truncus was quadri-valvular and mild truncal regurgitation was demonstrated. SVC and IVC were both left sided feeding into the right atrium (Figs. 9–12). Cardiac catheterization was then performed which, apart from supporting the ECHO findings of a single arterial trunk with right sided aortic arch (Figs. 5 and 6), large VSD (Fig. 8) and an atretic PA being fed through aorto–pulmonary collaterals, also demonstrated the presence of a single coronary artery taking origin from the arterial trunk (Fig. 7).

Based on the clinical examination findings and reports of X-ray, USG, CT, ECHO and cardiac catheterization, a diagnosis

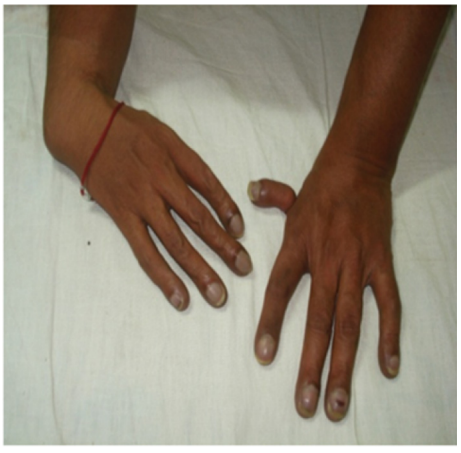

Fig. 2 – Dysplastic (R) forearm with absent thumb and rudimentary (L) thumb.