A CASE REPORT OF TWIN TO TWIN TRANSFUSION WITH TETRALOGY OF FALLOT IN BOTH TWINS

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Abstract:
We report an instance of twins born to a 22 year old G2A1 mother with no adverse antenatal factors having twin to twin transfusion. It was noted that both babies had Tetralogy of fallot(TOF)with pulmonary atresia. The rarity of the association of twins having a similar heart disease and twin to twin transfusion is highlighted.

Keyword: Twin to twin transfusion, Tetrology of Fallot

INTRODUCTION:
Twin to Twin transfusion is a commonly noted phenomenon among monochorionic twins. A Hemoglobin (Hb) difference of 5 g/dL or more is taken as indicative of twin to twin transfusion. Case reports of twin pregnancies with Tetrology of Fallot have been rarely reported in the world. There is only one such published case report from India though in older children. Also, there are no published case reports of twin to twin transfusion coexisting with TOF in existing literature.

We are bringing to your notice such a rare case.

CASE REPORT:
A 22 year old G2A1 mother delivered prematurely at 32 weeks of gestation, twin babies with birth weights of 1000g and 1560g respectively. There were no adverse antenatal risk factors other than the twin gestation. The mother had pretermprelabour rupture of membranes (PPROM) and underwent a caesarean section 13 hours after PPROM. Placentation was monochorionicdiamniotic. She delivered twin girl babies with severe twin to twin transfusion with a hemoglobin(Hb) of 5.7 g/dL and 25.5g/dL respectively. The recipient twin ,the larger of the discordant twins, was severely polycythemic (Hb 25.5 gm%). The hemoglobin levels came down to 18.2 gm% post exchange.

Baby had baseline saturation in the mid 80s from 24 hours onwards with no respiratory distress and no increase in saturation
noted on adding supplemental oxygen. There was no murmur. Echocardiography done showed large non-restrictive VSD with 8 mm bidirectional shunt and 50% aortic override. Pulmonary artery confluence was seen with adequate pulmonary artery anatomy - MPA 2.8 mm, RPA 2.4 mm. Pulmonary valve was atretic. Multiple aortopulmonary collaterals were seen with left sided aortic arch. Impression was Tetralogy of Fallot with pulmonary atresia and aorto-pulmonary collaterals.

The other baby had severe pallor and was mildly hypotonic since birth. On auscultation, the baby had muffled heart sounds. Routine blood investigations other than anemia were within normal limits. She had a very high percentage of nucleated red cells (620/100 wbc) suggestive of intrauterine stress. The chest X-ray showed massive cardiomegaly and normal to increased pulmonary vascularity. Baby had severe respiratory distress since birth and so was on CPAP with a maximum PEEP of 5 cm H2O and this was continued for 37 hours. In view of increasing oxygen requirements from 36 hours of age, an echocardiogram was done which showed similar features of Tetralogy of Fallot with Pulmonary atresia and aortopulmonary collaterals like the other twin. Baby received packed cell transfusion after birth. She was also started on low dose Dopamine for poor cardiac contractility. The baby’s clinical condition was worsening since birth, developed metabolic acidosis and she died at 44 hours of age. Parents were not willing for any cardiac intervention in view of baby being extremely low birth weight and guarded prognosis.

A FISH analysis for 22q 11 deletion was sent in both babies and was negative for Di George, Velocardiofacial syndrome, TUPLE1-HIRA locus on chromosome 22q11.2.

DISCUSSION
Congenital heart diseases are more common in monozygotic twins compared to dizygotic twins. The incidence of concordance, that is both twins being affected, is about 5% in dizygotic twins compared to 25% in monozygotic twins (1). One of the commonest genetic association seen in babies with congenital heart disease is 22q11 deletion. This deletion can easily be identified by FISH (Fluorescent in situ hybridisation test). These deletions are commonly associated with conotruncal anomalies. However, incidence of Fallot's tetralogy and other cardiac lesions have been reported with this deletion. It has been reported that there is about 25% incidence of concordance for CHD among monozygotic twins. It is a matter of interest to know whether the concordance refers to any involvement of the heart or denotes the similarity in the case of heart lesion. It is quite common for the heart lesion to be phenotypically different (1). There are few reports of Tetralogy of Fallot in twins. However, only one of the case reports showed 22q11 deletion. Lu J et al (2) give an account of twin pregnancy (monozygotic) with both children having TOF and associated pulmonary atresia. This was a proven case of 22q11 deletion contributing to the cardiac problem. However, their extracardiac great vessel patterning was different thus showing that phenotypic variability of great vessel development cannot be always explained on basis of genotypic variations. There was no twin to twin transfusion in this case. There are multiple case reports of twins with TOF but negative for 22q11 deletion. Jensen H et al (3) reported a case of monozygotic twins with TOF, with no major chromosomal abnormalities but submicroscopical chromosomal...
aberrations. Laugel et al. (4) discussed a case report showing monozygotic twins, both affected by TOF but there was no 22q11 microdeletion when assessed prenatally. The author postulated that other genetic factors may play a role. In another report from 1970, a similar association between monozygotic twin and TOF was found (5). Cassidy et al. identified triplet siblings with TOF. The cases were identified in infancy when they presented with a cardiac murmur (6).

Nora et al. (7) also identified one set of twins with TOF when she conducted an analysis of monozygotic twins. She also gave an association of 25% concordance for heart involvement in monozygous twins after combining her results with nine previous studies. Alva et al. (1) reported 8-year-old monozygotic twins with TOF, but were negative for microdeletion in 22q11. The author analysed the presence of concordant cardiac lesion and noted the rarity (total 4 cases). He also analysed another monozygotic twin pregnancy with pulmonary stenosis but negative for 22q11 microdeletion. There are no published reports so far from India on twins with Tetrology of Fallot in the newborn period. There is a case report in older children in India. Patel et al. (8) first reported in India 7-year-old monozygotic twin sisters with TOF. This was the case report of concordancy in heart disease among twins though detected in older children. Ours may be the first case report of concordant heart lesion-TOF, reported from India in the newborn period. Alan Fryer et al. (9) brought forward a case report showing a family tree with 22q11 deletion and the family having various cardiac anomalies and phenotypic variations. It was postulated that in addition to the genetic makeup, in vivo factors also played a role for expression of heart lesion. Ross (10) defines the influence of environmental factors in the discordancy of the lesions in monozygous twins. There are instances of 22q11 deletion in one of the twins with that baby alone having TOF. Kadaretal (11) reported a twin pregnancy with 22q11 deletion with one of them having Truncus arteriosus and the other having TOF. He emphasised the importance of antenatally detecting 22q11 deletion and their importance in the further management of the patient. Goodship et al. (12) reports a twin pregnancy born at 36 weeks of gestation with the smaller twin diagnosed with TOF at 8 weeks of life. This was a case of monozygotic twins with 22q11.2 deletion but there was cardiac involvement in only one baby (smaller) even though they looked phenotypically the same. It was postulated that 22q11 deletion predisposed the baby to a heart problem and the twinning process made one of the baby to manifest. Yamagaishi et al. (13) also has a case report of monozygotic twins with twin to twin transfusion with 22q11.2 deletion and one of the twins (smaller one) having TOF. Husain et al. (14) discussed the case report of a parasitic twin with imperfectly formed organs attached to the midline of the host twin, with the host twin having TOF and omphalocele containing the parasitic kidney and bowel. He postulated a single causative factor may have caused the sequence of events. The presence of microdeletion was not reported in this case. Singh et al. (15) has commented that the discordancy of monozygous twin in case of heart lesions may be explained by differences in the epigenetic mechanism involving DNA methylation which are sensitive to in vivo effects during development and differentiation.
The fact that the majority of the case reports had absence of 22q 11 deletion brings us to the fact that other factors may also have a play in the causation of the heart defect. The fact that the heart lesions may be discordant even in monozygous twins brings into the picture that environmental factors may play a role. In our study, as mentioned above, we had discordant twins having identical congenital heart disease along with severe twin-to-twin transfusion. The anemic twin also had cardiac failure probably secondary to the anemia and the multiple aorto-pulmonary collaterals. Cardiac failure is rarely seen in Fallot physiology. FISH for 22q deletion was negative.

REFERENCE:


