EBSTEIN'S ANOMALY IN CONGENITAL RUBELLA SYNDROME - A RARE ASSOCIATION - A CASE REPORT.
KANNAN D DURAI SAMY
Department of Paediatrics, MADRAS MEDICAL COLLEGE AND GOVERNMENT GENERAL HOSPITAL

ISSN 2455-2852
2019, Vol. 5(1)

Abstract: Congenital Rubella Syndrome (CRS), is caused by maternal rubella infection which presents with classical triad of cataract, deafness and congenital heart disease (CHD). We report a case of female child, 9 years old, a case of congenital rubella syndrome who had Ebstein's anomaly, is reported for its rarity.

Keyword: EBSTEIN'S ANOMALY, CONGENITAL RUBELLA SYNDROME, DEAFNESS, CATARACT.

INTRODUCTION:
Congenital rubella syndrome (CRS) was first described by Australian ophthalmologist Norman McAllister Gregg (1941). He observed the association of cataract and congenital heart disease in children born to mother who had rubella infections during early pregnancy. The evidence about the true burden of CRS in India, is not available. But approximately 10-30% adolescent female are susceptible to rubella infection in India.

A female child of 9 years age, first born of non consanguineous parents with perinatal period being uneventful admitted with H/O hard of hearing, inability to speak and left microcornea was referred for cardiac evaluation. On detailed antenatal history, mother had exanthematous fever at 8th week of gestation for which no treatment was given and she recovered spontaneously. She was immunized as per national immunization schedule and was not given any optional vaccine. She was not on any medication (like lithium which could cause ebstein's anomaly,etc..) during antenatal period. Full term vaginal delivery, low birth weight of 2.2 kg with no significant post natal events. The child had normal motor, social and adaptive development. Examination showed the child was comfortable at rest, cyanosed with pandigital clubbing and left microcornea. Her height, weight and head circumference were within normal limits. In Cardiovascular examination, apical impulse displaced laterally. On auscultation first and second heart sounds were normal, multiple clicks, grade 3/6 pan systolic murmur in tricuspid area, grade 3/6 ejection systolic murmur in pulmonary area. Neurological system was normal except for bilateral (B/L) hard of hearing. Fundus examination of both eyes showed salt and pepper retinopathy suggestive of congenital The child was investigated and biochemical investigations, thyroid profile were within normal limits. Audimetry: B/L severe sensori-neural hearing loss. Rubella IgG antibody level 4.5( Negative <0.8, positive >1.1, equivocal 0.8-1.1). Chest X-ray showed cardiomegaly with right atrial enlargement and pulmonary oligemia. Electrocardiogram showed normal sinus rhythm with normal atrio-ventricular conduction, right atrial enlargement, right ventricular hypertrophy, Himalayan P waves. Echocardiography was done which showed situs solitus, levocardia, atrial septal defect (ASD) of ostium secundum (OS) type -9 mm in size with bidirectional shunt, Small patent ductus arterious (PDA), left to right shunt, downward displacement of tricuspid valve of 2.6 cm, tricuspid regurgitation, mild right pulmonary artery (RPA) stenosis with impression of Ebstein's anomaly with small PDA, ASD OS ,mild RPA stenosis. With this clinical picture of B/L sensori-neural hearing loss, CHD, salt and pepper retinopathy child was clinically confirmed as a case of CRS. WHO surveillance reference: Clinically confirmed CRS case: “An infant in whom a qualified physician detects at least two of the complications listed in (a) below or one in (a) and one in (b):
(a) Cataract(s), congenital glaucoma, congenital heart disease, loss of hearing, pigmentary retinopathy
(b) Purpura, splenomegaly, microcephaly, mental retardation, meningocoealitis, radiolucent bone disease, jaundice that begins within 24 hours after birth” 5. Parents were counseled regarding the condition, child was issued hearing aids and taught speech therapy. Nutritional, immunization and medical advice. Follow up was planned in general medical, ophthalmology, ENT, cardiology and cardio-thoracic OPD. The child is being in the regular follow up.

This case of CRS is reported for its rare association with Ebstein's anomaly with CRS.

DISCUSSION:
The incidence of CRS is decreasing with the advent of rubella/MMR/MMRV vaccine. Maternal viremia associated with rubella infection during pregnancy may result in infection of the placenta and fetus. Only a limited number of fetal cells become infected.
The growth rate of infected cells is reduced, resulting in fewer numbers of cells in affected organs at birth. The infection may lead to deranged and hypoplastic organ development, resulting in structural anomalies in the newborn. Timing of the fetal infection determines the extent of teratogenic effect. In general, the earlier in pregnancy infection occurs, the greater the damage to the fetus. Infection during the first trimester of pregnancy results in abnormalities in the infant in about 85% of cases, whereas detectable defects are found in about 16% of infants who acquired infection during the second trimester. Birth defects are uncommon if maternal infection occurs after the 20th week of gestation. Importantly, maternal infections can produce these anomalies as well.

The classic triad of CRS is cataract, sensory neural deafness, congenital heart disease, patent ductus arteriosus being the commonest 3. Clinical manifestations – Heart defects (patent ductus arteriosus-78%, right pulmonary artery stenosis-70%, left pulmonary artery stenosis-40%), valvular pulmonary stenosis-67%, low birth weight-60%, psychomotor retardation-45%, neonatal purpura-23%, diabetes mellitus-20%, thyroid dysfunction (5%), microcephaly, prematurity, hepatitis, linear streaking of bone, developmental delay, autism spectrum disorders, schizophrenia, growth retardation, learning problems, progressive rubella panencephalitis (PRP), death (35%)

Treatment -
- No specific treatment, only supportive to the individual problems
- Prevention - Vaccination of all women by any means (>99% prevention)
- 2 doses - one @ 15 months of age, next @ 4-6 years of age, ideal @ 5th year when DPT, OPV booster 3.
- After vaccination pregnancy should be postponed for at least 3 months (infection 11 weeks prior to pregnancy can cause CRS 3). The association between CRS and CHD has been well studied. In the first report on this syndrome, Gregg et al found congenital heart disease in 44 of 78 patients recognized through the presence of congenital cataracts. Twenty years later (1961) Campbell et al indicated that patent ductus arteriosus continued as the most frequent cardiovascular defect following maternal rubella during pregnancy. His list of defects included ventricular septal defect, atrial septal defect, pulmonary stenosis, tetralogy of Fallot, coarctation of the aorta, aortic valve stenosis, transposition of the great arteries, and tricuspid atresia.

The pathophysiology of this relationship was explained by Stuckey et al. He argued that damage produced in the heart by this infectious agent was related to gestational age at the time the infection occurred. He reasoned that "the cardiac septa, the main vessels, and their valves are only susceptible to damage by environmental factors for a short period of some 4-5 weeks of fetal life while they are being actively formed, the so-called critical period for these structures. The ductus arteriosus, on the other hand, is present for many months of fetal life and for a short time after birth, and whatever the mechanism of final closure it is conceivable that it could be damaged at any stage. The greater incidence of patent ductus arteriosus in these children may thus merely reflect the longer time that this structure is at risk." 7. Agarwal et al in 2001 reported a 32-year-old, fifth gravida woman who was detected to be strongly positive for rubella at 15 weeks of gestation (IgM titers: patient - 0.973 vs control - 0.428) delivered a term, IUGR male baby with cyanosis and respiratory distress. Echocardiography revealed severe Ebstein's anomaly with a patent ductus and a small ventricular septal defect. This is the only case of CRS with multi-aneurysmal which has been reported so far 8.

REFERENCES:
4. Harrison's principles of internal medicine. 18th edition.
5. Surveillance Guidelines for Measles, Rubella and Congenital...