Scholars Journal of Medical Case Reports **ISSN 2347-6559 (Online)**
Sch J Med Case Rep 2014; 2(7):470-472 **ISSN 2347-9507 (Print)**

(An International Publisher for Academic and Scientific Resources)
www.saspublishers.com DOI: 10.36347/sjmcr.2014.v02i07.015

Congenital Rubella Syndrome with Basal ganglia calcification and Bilateral Nuclear Cataracts in a Neonate: A Rare Entity

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Abstract: In-utero transmission of rubella virus during pregnancy leads to congenital Rubella syndrome (CRS), which is characterized by cataracts, deafness and sensorineural hearing loss. While the nuclear cataract is the most frequently occurring abnormality, basal ganglia calcification is a rarer manifestation. The Risk of fetal transmission of rubella virus is highest during the first 12 weeks of gestation and decreases afterwards. A 36weeks, 2600grams male neonate was born by vaginal delivery with normal APGARS. The neonate was admitted on third day of life with complaints of yellowish discoloration of the skin and sclera. On examination, the neonate had mild hypotonia, icterus up to the legs and systolic murmur on cardiac examination. Interestingly, the ophthalmological examination revealed bilateral nuclear cataracts. Chest radiograph showed cardiomegaly and ECHO revealed PDA (Patent Ductus Arteriosus). Hence, CRS was suspected and further evaluation was done. There was thrombocytopenia, mild unconjugated hyperbilirubinemia with mild elevation of transaminases. Another rare finding was basal ganglia calcification on neurosonogram. TORCH profile in both mother and baby showed elevated rubella IgM levels confirming CRS. The neonate received supportive and symptomatic treatment. We report a preterm, male appropriate for gestational age neonate with congenital rubella syndrome and its rare manifestation of basal ganglia calcification.

Keywords: Congenital rubella syndrome (CRS), neonate, basal ganglia calcification, congenital cataract, congenital heart disease (CHD), TORCH infection

INTRODUCTION

Veale, a Scottish physician described Rubella as a contagious, benign disease in 1866 [1]. However, the classic triad of congenital rubella syndrome (CRS) was found by Sir Norman Gregg, consisting congenital cataracts, deafness and heart defects [2]. CRS occurs due to maternal primary infection with rubella virus during first trimester. It has a wide spectrum of presentation which ranges from silent viremia to spontaneous abortions, still births and severe congenital malformations of the fetus. Similarly, gestational age at the time of viremia also determines the severity and type of congenital defects [3]. Both deafness and cardiac manifestations occur if viremia takes place before 11weeks and viremia between 13-16 weeks results in isolated deafness. However, the developmental anomalies are infrequent, when the infection strikes beyond 16weeks of gestation. This principle is common to majority of the intrauterine infections [3]. Though, the exact teratogenic mechanism of in-utero rubella infection remains unidentified, viral replication in the cells during fetal organogenesis seems to be accountable for CRS [4]. While the reported incidence of cataracts and microphthalmia in CRS is 60-70%, congenital glaucoma and intra cranial calcifications are rare, more so in the neonate [5-8]. The most common outcome is sensory neural or central deafness, observed in 80% of the cases [6]. Though, CRS is a rare entity, it is the second most common cause of non-traumatic childhood cataracts and an important preventable disease leading to blindness in children.

CASE REPORT

A 36weeks out born, appropriate for gestational age (birth weight-2600grams) male neonate was born to a second gravida by spontaneous vaginal delivery.

There was history of upper respiratory tract infection during the first trimester, however the presence of rash was unnoticed. There were no other maternal complications. The neonate cried soon after birth with normal APGARs. The first pregnancy was a miscarriage, hence mother's TORCH profile was done, which showed elevated rubella IgM levels. The neonate was admitted on third day of life with yellowish discoloration of the skin and sclera. On examination there was mild hypotonia, icterus and bilateral nuclear cataracts were also seen (Fig. 1). Systemic examination revealed pre-cardial activity, pan systolic murmur (suggestive of congenital heart defect-PDA) with hepatosplenomegaly. Hence, congenital rubella syndrome (CRS) was diagnosed and the neonate was further investigated. Sepsis screen was negative but thrombocytopenia was evident in complete blood picture, along with unconjugated hyperbilirubinemia and elevated transaminases. Chest radiograph showed mild cardiomegaly and echocardiogram revealed PDA (3mm) with left to right shunt and patent foramen ovale with left to right shunt. Basal ganglia calcification was noted on neurosonogram (Fig. 2). However, there were no intra-cranial calcifications on skull radiograph. Oto-Acoustic emission (OAE) examination was normal in both ears, ruling out the possibility of cochlear deafness. The neonate's rubella IgM levels were elevated, confirming *CRS*. Supportive and symptomatic treatment was given to the baby and it was discharged on decongestive treatment for PDA. B scan of the eyes was normal and the infant is planned for cataract surgery at 3 months of age.

Fig. 1: Bilateral nuclear cataracts, as shown with arrows

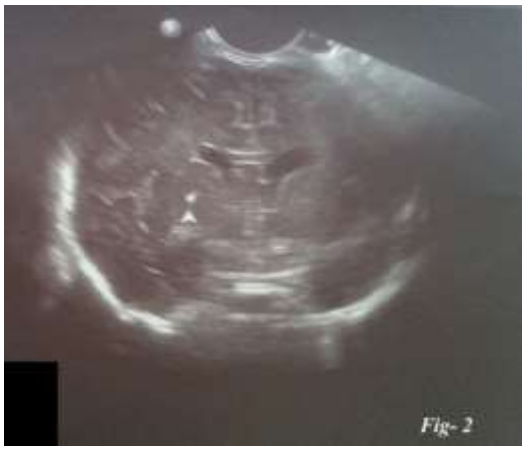

Fig. 2: Ultrasound image revealing right basal ganglia calcification

DISCUSSION

Congenital rubella syndrome (CRS) results from chronic infection of the fetus with rubella virus with progressive damage to the various organs. It leads to a wide array of systemic manifestations ranging from acute multi organ involvement in the neonatal period to deafness and progressive mental retardation in infancy [6]. Although, there is a considerabe decline in the frequency and prevalence of fetal damage due to rubella virus with widespread vaccination against rubella, CRS is not completely exterminated. The virulence of the organism and the gestational period of exposure determine the severity of damage [6]. The outcome is worse with exposure in early gestation. The frequency of defects in CRS is higher with early exposure, (in the first 12 weeks- more than 80%, 13-14 weeks-54%, and at the end of second trimester-25%). The spectrum and severity of fetal involvement has two peaks, one before 12 weeks of gestation and the other at 36 weeks and beyond. Formation of cytotrophoblast during second trimester leads to increased resistance of the placenta to viral invasion, but thinning of the trophoblast during third trimester enables viral transmission. The frequency of ocular and cardiac defects wanes off, while that of hearing defects amplifies with increasing gestation. As the fetus is physiologically immune-deficient, transplacentally transmitted virus spreads in the rapidly growing fetus leading to interruption in organogenesis and major congenital defects [6].

Intra uterine growth retardation and prematurity frequently manifest in CRS. The commonest defect is central hearing loss [6]. An interesting observation is that severe hearing loss is much more frequent than milder forms. Isolated hearing loss can denote CRS [6]. Among the ocular defects, nuclear cataract is the commonest one, while congenital glaucoma is a rare [7, 9, 10]. The index neonate had bilateral nuclear cataracts. The nuclear cataract results from viral invasion of the developing lens. Congenital glaucoma results from either failure of absorption of

mesoderm at the angle of anterior chamber or from failure of differentiation of canal of Schlemm. The increased intraocular pressure leads to corneal clouding. Often, it is seen at a later age than in the neonatal period. Other ocular abnormalities like microphthalmia and salt pepper retinopathy can also manifest in CRS. Characteristic cardiac defects that occur in *CRS* include patent ductus arteriosus (PDA), pulmonary stenosis and ventricular septal defect (VSD). Cardiac lesions are more frequently prevalent in neonates with ocular lesions [6, 9, 10]. Index case had PDA along with ocular manifestations.

Other infrequent manifestations include intra cranial calcifications [8], meningo-encephalitis, hepatosplenomegaly, bone lucencies, thrombocytopenia, anemia, rubelliform rash, cryptorchidism and diabetes mellitus. Microcephaly with mental retardation, hypogammaglobulinemia, thymic hypoplasia, thyroid abnormalities and polycystic kidney disease were also described in CRS. Basal ganglia calcification is a very rare presentation, which is present in the index case [8]. Some of the defects can present months to years later in CRS. Neonatal rubella can manifest, when the maternal infection takes place near term due to insufficient maternal antibodies along with transplacental viremia. The impact of passive immunization is uncertain in the prevention of progressive disease [11].

Isolating rubella virus from oropharynx, urine or detecting rubella specific IgM in the cord blood or neonatal blood along with elevated rubella IgG titers over time are diagnostic modalities of CRS [12]. In the index case, IgM levels were elevated, confirming the diagnosis of CRS. There is no specific treatment for congenital rubella. However, preventing CRS by immunizing all susceptible adults plays a paramount role in eliminating or reducing the incidence of this dreadful disease. Conception should be delayed for three months after rubella vaccination, to avoid the risk of fetal infection.

CONCLUSION

It is a case of a late preterm male neonate with congenital rubella syndrome and its rare manifestations of congenital nuclear cataracts and congenital intra cranial calcification (basal ganglia calcification). Coexistence of many other less frequent systemic manifestations and positive serology makes this case unique.

ACKNOWLEDGMENTS

We are thankful to Mr. Muzahid for formatting and thanks to the nursing staff for their help in managing this rare case.

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