TORCH Syndrome

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Abstract: We present here a case report of a torch syndrome with significant findings. The baby had congenital cataract, blue berry muffin spots, atrial septal defect and supporting serological and radiological findings. A multidisciplinary approach of management was adopted.

Key words: Cataract, torch syndrome, blueberry muffin spots.

INTRODUCTION:

The word TORCH stands for Toxoplasma, Others (Parvovirus B19, Varicella-Zoster virus, Hepatitis B, Syphilis), Rubella, Cytomegalovirus, Herpes.

Major clinical features are jaundice with hepatosplenomegaly, cataract, blue berry muffin spots, congenital heart defects, cerebral calcification with supporting serological findings (positive IgG antibodies for different offending antigens). The diagnosis is mainly clinical and serological. A multidisciplinary approach of management is required.

CASE SUMMARY:

A 12 days old baby girl from Bhadoi district of Uttar Pradesh presented at our department with parents complaining of opacity and constant movement of both eyes since birth. Birth history revealed pre-term birth with birth weight of 2400 grams. She was born by the mother who had consanguineous marriage and had previous 3 abortions.

Ocular examination revealed normal anterior segment of both eyes. Bilateral cataractous lens was there (Figure 1). Both eyes fundus was not visible due to lenticular opacity.

The body of the baby had several reddish nodular lesions. Detailed pediatric examination revealed hepatosplenomegaly, bilaterally chest clear, heart murmurs present. Anterior fontanel was wide open. Moro’s and suckling reflex was present. Whole body had several bluish elevated dermal vesicles suggestive of extramedullary cutaneous erythropoiesis (Blueberry Muffin spots) (Figure 2-6).

Figure 1: Coloured photograph showing bilateral lenticular opacity

Figure 2: Coloured photograph showing Extramedullary cutaneous erythropoiesis (Blueberry Muffin spots)
Figure 3: Coloured photograph showing Extramedullary cutaneous erythropoiesis (Blueberry Muffin spots)

Figure 4: Coloured photograph showing Extramedullary cutaneous erythropoiesis (Blueberry Muffin spots)

Figure 5: Coloured photograph showing Extramedullary cutaneous erythropoiesis (Blueberry Muffin spots)

Figure 6: Coloured photograph showing Extramedullary erythropoiesis (Blueberry Muffin spots)

Serological examination of the mother revealed positive IgG antibody for Rubella, Cytomegalovirus, and Herpes simplex virus. IgG antibody for Toxoplasma gondii was in borderline range. IgM antibody for Rubella, Cytomegalovirus, Herpes simplex virus, Toxoplasma gondii was negative suggesting chronic maternal TORCH infection.

Serological examination of the baby revealed positive IgG antibody for Rubella, Cytomegalovirus, and Herpes simplex virus. IgG antibody for Toxoplasma gondii was negative. IgM antibody for Rubella, Cytomegalovirus, Herpes simplex virus, Toxoplasma gondii was negative suggesting maternal TORCH infection transmitted to the child.

Blood picture of the baby showed raised WBC with relative lymphocytosis suggesting infective pathology, raised Total and Indirect bilirubin, raised Alkaline phosphatase enzyme suggesting hepatic involvement.

2D Echo and colour Doppler report of the baby showed Congenital acyanotic heart disease with Ostium secundum type of Atrial septal defect.

USG Cranium of the baby (Figure 7,8) revealed B/L mildly dilated 3rd ventricles with dilated frontal and temporal horns. Multiple echogenic foci (calcifications) are scattered throughout the brain parenchyma predominantly at periventricular region. Few cystic lesions are also present in bilateral frontal and temporal lobes. These findings supported TORCH etiology.
Finally the diagnosis of TORCH Syndrome was made and a multidisciplinary approach of management was adopted.

**DISCUSSION:**

The word TORCH stands for Toxoplasma, Others (Parvovirus B19, Varicella-Zoster virus, Hepatitis B, Syphilis), Rubella, Cytomegalovirus, Herpes.

**Toxoplasmosis**

It is caused by the infection with the protozoan Toxoplasma gondii, an obligate intracellular parasite. T. gondii is transmitted through the fecal matter of cat, eating raw meat, contaminated water and soil, and unpasteurized goat milk [1]. Clinical features of toxoplasmosis are jaundice with hepatosplenomegaly, microcephaly or hydrocephalus, cerebral calcifications, retinochoroiditis, microphthalmia, convulsions, mental retardation, deafness. Congenital toxoplasm
osis can lead to repeated abortions. Fetuses infected in the third trimester are often asymptomatic at birth. At early detection, the mother is treated with spiramycin (1500 mg BID) to prevent fetal infection. If the fetus is found to be infected, the treatment is combination of pyrimethamine and sulfadiazine (classic therapy). It is given along with supplements of folinic acid as it prevents the bone marrow suppression caused by pyrimethamine and sulfadiazine [2].

Rubella or German Measles

It is transmitted through direct contact or airborne droplets from the respiratory system. Rubella virus enters into mother’s body, then spreads via blood, placenta, and infects the fetus. Maximum chances of infection to the baby are mainly in the first and last trimester of pregnancy. Congenital rubella infection can present as 1. Classical congenital rubella syndrome (congenital heart diseases particularly patent ductus arteriosus or pulmonic stenosis and ocular defects mainly cataracts, glaucoma or microphthalmia, microcephaly or deafness). 2. Expanded congenital rubella syndrome (intrauterine growth retardation, thrombocytopenic purpura, encephalitis and myocarditis, blueberry muffin baby, jaundice with hepatosplenomegaly, purpura or dermal erythropoiesis. Other manifestations may include a large anterior fontanelle, transient radiolucencies in longitudinal bones, failure to grow well and dental enamel defects. 3. Late onset rubella syndrome (acute severe multisystem disease, with interstitial pneumonia, skin rash, diarrhea, hypogammaglobulinemia, circulating immune complexes, aseptic meningitis, hepatosplenomegaly, thrombocytopenia, and Pneumocystis pneumonia).

Cytomegalovirus Infection

It is transmitted to an infant during pregnancy or by ingestion of infected human milk or direct contact with urine and saliva. Infants show various clinical features such as optic atrophy, microcephaly, intracranial calcifications, hypotonia, decreased hearing, thrombocytopenic purpura. If the mother has a primary infection during pregnancy, fetal morbidity rate is high [3].

Herpes Simplex Virus

It is found in two forms HSV 1 and 2. HSV1 mainly affects upper body parts and leads to infections as gingivostomatitis, pharyngitis and HSV2 is mainly involved in the genital herpes. Neonatal HSV infection most commonly presents within 1 to 3 weeks of birth. Localized disease presents as vesicular eruptions on skin, mouth or eyes. If it is left untreated, more than 70% of cases involves whole body and becomes disseminated disease. Clinical features of disseminated disease include poor feeding, fever, lethargy, apnea, convulsion, respiratory distress, hepatomegaly, jaundice, and disseminated intravascular coagulation. Despite systemic treatment, disseminated disease is associated with mortality rates between 50% (HSV-2) and 70% (HSV-1) [4]. Features associated with poor prognosis are hemorrhagic pneumonitis, meningoencephalitis, severe coagulopathy and liver failure.

So, a multidisciplinary approach of management is required.

# CONFLICTS OF INTEREST: There are no conflicts of interest.

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REFERENCES:

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FIGURE LEGENDS:

Figure 1: Bilateral lenticular opacity
Figure 2-6: Extramedullary erythropoiesis (Blueberry Muffin spots)
Figure 7, 8: B/L mildly dilated 3rd ventricles with dilated frontal and temporal horns. Multiple echogenic foci (calcifications) are scattered throughout the brain parenchyma predominantly at periventricular region. Few cystic lesions are also present in bilateral frontal and temporal lobes.