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# CYTOGENETIC STUDIES IN ABORTUS FOETUSES OF TORCH POSITIVE MOTHERS EXPERIENCING BAD OBSTETRIC HISTORY



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# **ABSTRACT**

During gestation, the microorganisms causing severe birth defects and the resulting clinical syndrome have been categorized as TORCH infections which includes Toxoplasmosis, others (syphilis, varicellla-zoster, parvovirus B19), Rubella, Cytomegalovirus (CMV) and Herpes infections. Hence the present study was undertaken to investigate the role of maternal infections (TORCH) and the incidence of foetal chromosome anomalies. Aborted fetuses of T0RCH positive mothers were selected for this study. The foetal blood samples were collected from all aborted fetuses of mothers with TORCH IgM/IgG antibodies. Genetic study was carried out in aborted fetuses of fifty five TORCH positive mothers. Among 55 aborted cases 45 (82%) showed normal karyotype and the rest 10(18%) showed abnormal karyotype. The study revealed that the incidences of foetal chromosomal abnormalities were increased with increased age of women. Among TORCH positive cases maximum abnormal karyotype were observed among CMV infected cases followed by Rubella. Karyotype abnormality was found to be elevated if the mother was positive for CMV and any other infections. Least abnormalities were observed among Toxoplasma positive cases. The increased incidences of foetal chromosome abnormalities among the TORCH positive subjects strongly recommend for prenatal screening and genetic counseling to the parents before the next pregnancy.

# **KEYWORDS**

TORCH, Karyotype, Chromosomal abnormalities, Bad obstetric history

#### Introduction

During gestation many microorganisms can infect the fetus, causing severe birth defects. Such organisms and the resulting clinical syndrome have been categorized as TORCH infections (Franca and Mugayar, 2004).TORCH, includes Toxoplasmosis, other (syphilis, varicellla-zoster, parvovirus B19), Rubella, Cytomegalovirus (CMV), and Herpes infections. These are some of the most common infections associated with congenital anomalies (Stegmann and Carey, 2002).

It is absolutely necessary to screen TORCH infection for women who had the previous history of abnormal pregnancies in order to prevent birth defects and perinatal complications (Cao et al, 1999). Primary TORCH infections in the mother can lead to severe fetal anomalies or even fetal loss (Kaur et al, 1999).

It is becoming clear that viruses have evolved elaborate interactions with the cellular repair, recombinant machinery in order to create an environment conducive to their own replication. The host cell's DNA damage machinery is alert for pertubations in DNA which could lead to genetic instability. After an infection, some of this machinery is inactivated by the virus in attempt to remove obstacles to productive infections: however, other components are utilized by the virus to promote viral DNA replication (Wilkinson and weller, 2004).

The genome of DNA viruses that replicate in the nucleus interact with and cause the redistribution of many cellular proteins. Larger scale rearrangements of nuclear structures can start early during infection and become extreme as complexes of viral proteins and replicating DNA molecules, known as replication compartments expand to fill the nucleus , infection by several DNA viruses induces a DNA damage (Roger, 2006). Hence the present study was undertaken to investigate the role of maternal infections (TORCH) and the incidence of foetal chromosome anomalies.

## Materials and methods

Aborted fetuses of TORCH positive mothers were selected for this study. The inclusion criteria's were ;women of child bearing age ,couples having early pregnancy with congenital anomalies/dysmorphic features and /or couples experiencing multiple pregnancy loss or conceived after a long period of infertility. Detailed clinical history and other relevant information's were collected by using proforma .The laboratory reports of TORCH screening of all these subjects were recorded. The foetal blood samples were collected from all aborted fetuses of mothers with TORCH IgM/IgG antibodies, satisfying above said criteria.

#### Observations and results

One hundred and ten women with a history of recurrent abortion or women conceived after a long period of fertility treatment, who were TORCH positive were selected for this study .Postmortem karyotyping was performed in all aborted fetuses of these 110 TORCH positive mothers.

When foetal karyotypes analyzed, 90 (82%) showed normal karyotype and the rest 20 (18%) showed abnormal karyotype. Generally the incidences of foetal chromosomal abnormalities increased with increased age of women.

Based on the duration of married life it is found that when duration increases the chromosomal abnormalities also increased.

Among the abnormal foetal karyotype 4 showed sex chromosome anomalies and the rest 16 were autosomal. The abnormal karyotype includes 60% of numerical abnormalities and 40% of structural abnormalities.

Toxoplasma IgG/IgM positive cases were observed in 32(32.72%) subjects. Rubella IgG/IgM positive cases were observed in 38 (34.5%) subjects. CMV IgG/IgM positive cases were observed in 28(25.5%) subjects. HSV IgG/IgM positive cases were observed in 42(38%) subjects.

Among 32 Toxoplasma positive women, foetal chromosomal anomalies were found in 6 cases. In the case of Rubella infection, 12 abnormal foetal karyotype were observed among 38 subjects with Rubella IgG/IgM positive. Foetal chromosome anomalies were observed in 10 of the 28 subjects with CMV IgG/IgM positive and 10 foetal chromosome anomalies were observed in 42 subjects with HSV IgG/IgM positive.

## Summary and conclusions

The increased incidence of foetal chromosome abnormalities among these subjects with TORCH infections strongly recommends the need for genetic screening to the next pregnancies and to extend genetic counseling. Pre-pregnancy or routine antenatal screening for presence of, or susceptibility to, some of these infections and appropriate management can prevent adverse fetal or perinatal outcomes; screening should include Rubella IgG, Hepatitis B surface antigen and serological tests for syphilis and HSV antibody. Keeping consideration of the high cost of the test panel, selected tests (of the whole panel) are recommended on an individual case basis. Incorporation of rubella

immunization into the national immunization schedule is recommended. Toxoplasma-associated infection can be prevented by educating the public about avoidance of ingestion of raw or insufficiently-cooked meat and poultry and keeping proper hygiene.

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