AICOG 2025
Mumbai
67th All India Congress of Obstetrics & Gynaecology
FOGSI celebrates 75 years - Diamond Jubliee
8-12 January, 2025 | JIO World Convention Centre

Poster Number: EP **162** Presenter Name: **SRINIVASA K.C.** Post-graduate Student

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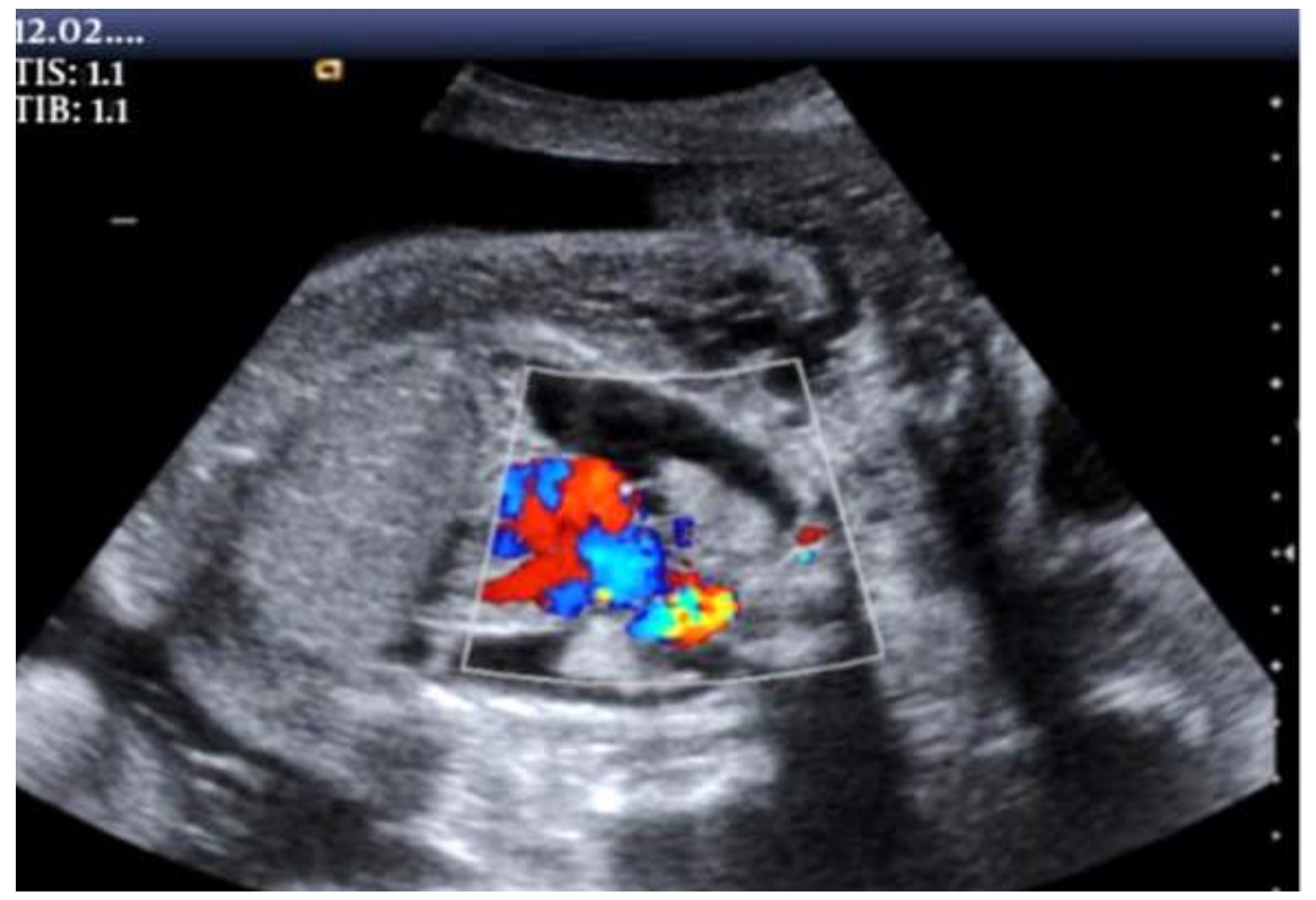
Title: TOXOPLASMOSIS IN PREGNANCY

INTRODUCTION:

Toxoplasmosis is caused by ubiquitous protozoan parasite Toxoplasma Gondii It is generally asymptomatic disease. Primary Maternal infection during Pregnancy can be transmitted to the foetus and results in serious sequelae and major congenital malformations, intrauterine death and severe genetic abnormalities in the newborn.

CASE OPERATION PROCEDURE:

- ❖ 23 years old G3P1L0A1 with 34 weeks of gestation with known case of hypothyroidism admitted to our hospital i/v/o decreased perception of foetal movements, unbooked case.
- ❖ On examination – Vitals stable no pallor, no icterus no pedal oedema CVS /RS – NAD, Per-abdomen uterus 28 to 30 week size clinical oligo present breech at lower pole. Routine antenatal investigations were normal.
- ❖ Growth scan : It shows dilatation of 3rd and 4th ventricles with periventricular calcifications f/s/o CMV infection with Stage-1 IUGR, Pleural effusion and pericardial effusion.
- ❖ Torch screening test: IgM antibodies Positive for toxoplasmosis.



Patient Name **Mrs DEVIKA**
Age : 23 Year(s) Gender : Female
Sample ID : 19264602
Sample Type : Serum
Patient ID : 2128071
Ref. Doctor : SNEHA LAB
Ref. Customer :

Lab Code : CPL-KA-053
Sample Collection Date : 2024-06-09 14:51
Registration Date : 2024-06-09 14:51
Approved Date : 2024-06-09 18:41

IMMUNOLOGY/SEROLOGY

Test Description	Result	Units	Biological Reference Ranges
TORCH 10 Profile IgG IgM			
Toxoplasma gondii IgG Antibody (Method: ELISA)	**1.46**	S/Co	< 0.9 : Negative 0.9-1.1 Doubtful > 1.1 : Positive
Toxoplasma gondii IgM Antibody (Method: ELISA)	0.28	S/Co	< 0.9 : Negative 0.9-1.1 Doubtful > 1.1 : Positive

INTERPRETATION

Toxoplasma IgG	Toxoplasma IgM	Remarks
Negative	Negative	No infection or very early infection; no previous exposure
Negative	Positive	Acute infection
Positive	Positive	Acute infection; Chronic infection; could indicate re-activation; IgM may be positive for several months after the infection resolves. Toxoplasma IgG avidity test will help differentiate between acute & chronic infection.
Positive	Negative	Past infection

Note:
- This assay is used for quantitative detection of specific IgM antibodies to Toxoplasma gondii in serum samples.
- Positive result for Toxoplasma IgM indicates possible acute infection with Toxoplasma. False positive reaction due to rheumatoid factor and persistence of positive IgM for upto 2 years is not uncommon.
- Equivocal results should be re-tested in 10-14 days.
- Negative result indicates no serological evidence of infection with Toxoplasma. False negative can be due to immunosuppression or due to low/undetectable level of IgM antibodies. A suspected diagnosis of acute toxoplasmosis should be confirmed by detection of Toxoplasma gondii DNA by PCR analysis or repeat test after 2 weeks.
- The diagnosis should not be established on the basis of single test and the results should be interpreted in conjunction with clinical findings. The magnitude of the measured result is not indicative of the amount of antibody present.

Comments:
Toxoplasma gondii is an obligate intracellular parasite capable of infecting a wide variety of intermediate hosts including man. Toxoplasmosis is acquired by humans through ingestion of food or water contaminated with cat feces or through eating undercooked meat containing viable oocysts. Vertical transmission of the parasite through the placenta can also occur, leading to congenital toxoplasmosis. Infection in man is usually asymptomatic. Following primary infection, Toxoplasma gondii can remain latent for the life of the host; the risk for reactivation is highest among immunosuppressed individuals. The most common symptomatic presentation in man is lymphadenopathy, encephalitis, myocarditis and pneumonitis. Demonstration of Toxoplasma IgG in the serum of person with eye lesion helps in diagnosing ocular toxoplasmosis. In addition, antibody levels and demonstration of parasite DNA in aqueous humor confirms the diagnosis of ocular toxoplasmosis. Persistent or increasing IgG antibody levels in the infant compared with the mother and /or positive result of Toxoplasma specific IgM or IgA are diagnostic of Congenital toxoplasmosis. Demonstration of Toxoplasma specific IgG in CSF can help confirm the diagnosis of Congenital toxoplasmosis when the results are compared to serum Toxoplasma specific IgG levels. Congenital toxoplasmosis occurs when a woman passes the infection to her fetus after acquiring a primary infection during pregnancy or more rarely, when a pregnant woman is immunocompromised and a previously acquired infection is reactivated with an overall transmission rate varying between 30% to 50 % depending on the stage of pregnancy. Demonstrating Toxoplasmic specific IgM & IgA antibodies in fetal serum or isolating Toxoplasma from fetal leucocytes is a definitive diagnosis of fetal infection.

Test Description	Result	Units	Biological Reference Ranges
Rubella Virus IgG Antibody (Method: ELISA)	0.39	S/Co	< 0.9 : Negative 0.9-1.1 Doubtful > 1.1 : Positive

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Manager Technical

Dr. H. Sridhar MD
Consultant Pathologist

Common imprinting disorders associated with UPD

Imprinting disorder	Location	Type of mutation	UPD	Genes involved	Major clinical features
Transient neonatal diabetes mellitus	6q24	Pat, duplication, Methylation defect	Pat	*PLAG1, HYMAI*	IUGR, transient diabetes, hyperglycemia without ketoacidosis, macroglossia, omphalocele, heart & renal defects
Russell-Silver syndrome	7p11–p13, 7q31–qter, 11p15	Mat duplication, hypomethylation, genome-wide UPD, point mutations	Mat	*IGF2, H19, CDKN1C, KCNQ1, KCNQ1OT1*	IUGR/PNGR, macrocephaly, hemihypotrophy, prominent forehead, triangular face
Familial nonchromaffin paraganglioma	11q13, 11q23	Aberrant methylation	Mat	*SDHD*	Tumor of the paraganglia
Beckwith-Wiedemann syndrome	11p15	Hypermethylation, Chromosomal aberration, hypomethylation, genome-wide UPD, point mutations	Pat	*IGF2, H19, CDKN1C, KCNQ1, KCNQ1OT1*	Prenatal & postnatal overgrowth, organomegaly, macroglossia, omphalocele, neonatal hypoglycemia, hemihypertrophy, increased tumour risk
Kagami-Ogata syndrome	14q32	Mat deletion, aberrant methylation	Pat	*RTL1, DLK1*	IUGR, polyhydramnion, abdominal and thoracal wall defects, bell-shaped thorax, coat-hanger ribs
Temple syndrome	14q32	Pat deletion, aberrant methylation	Mat	*RTL1, DLK1*	IUGR/PNGR, neonatal hypotonia, feeding difficulties in infancy, truncal obesity, scoliosis, precocious puberty
Prader-Willi syndrome	15q11-q13	Pat deletion, aberrant methylation	Mat	*SNRPN, MKRN, MAGEL2, NDN, U5snoRNAs*	PNGR, mental retardation, neonatal hypotonia, hypogenitalism, hypopigmentation, obesity/hyperphagia
Angelman syndrome	15q11-q13	Mat deletion, aberrant methylation, point mutations	Pat	*UBE3A*	Mental retardation, microcephaly, no speech, unmotivated laughing, ataxia, seizures, scoliosis
Pseudo-hypoparathyroidism type 1b	20q13	Mat deletion, aberrant methylation, epimutation, Pat & Mat loss of function mutation	Pat & Mat	*GNAS*	Resistance to PTH and other hormones; Albright hereditary osteodystrophy, subcutaneous ossifications, abnormal growth patterns,Cushing syndrome, precocious puberty, fibrous dysplasia
Precocious puberty syndrome	15q11.2	Point mutations	Pat	MKRN3	Early activation of the hypothalamic-pituitary-gonadal axis resulting in gonadotropin-dependent precocious puberty
UPD 20 syndrome	Chr20	Unknown	Mat	Unknown	Growth failure, feeding difficulties

UPD 6 syndrome and UPD16 syndrome are still under debate. Pat – Paternal; Mat – Maternal

MANAGEMENT AND RESULTS:

- ❖ Patient was planned for induction of labour was induced with Tab. Mifepristone 200MG STAT followed by Foley's bulb and Cervi prime gel to deliver a S/L/PT Male baby of b.wt 1.56 kg @5.15pm on 12/06/2024, baby was resuscitated following delivery i/v/o of poor APGAR score but baby couldn't be reviewed even after 10mins of resuscitation and declared dead.
- ❖ Due to previous history of 1st trimester abortion following a fever episode and history of still birth in previous pregnancy, products of conception/dead fetus was sent to pathological autopsy examination and chromosomal micro array analysis.
- ❖ Pathological autopsic examination of dead neonate shows pericardial effusion and dilated chambers of the heart and unilateral undescended testes.
- ❖ Chromosomal microarray analysis of dead neonate shows Uniparental Disomy common imprinting disorders associated with this genetic abnormality include Rusell-silver syndrome, familial non chromartin ganglioma, temple syndrome, precocious puberty syndrome, Angelmann syndrome.

CONCLUSION: Diagnosis of maternal toxoplasmosis during pregnancy is based on seroconversion in pregnancy and anti toxoplasma gondii IgM and IgG and amniotic fluid survey for toxoplasma gondii specific DNA by PCR method for females planning for pregnancy it is recommended to avoid risky behaviours such as eating raw or under cooked meet.

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