



PRENATAL DIAGNOSIS OF HEMOGLOBINOPATHIES: EXPERIENCE FROM A SINGLE HOSPITAL IN EASTERN INDIA

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INTRODUCTION

- ❖ Hemoglobinopathies are monogenic disorder.
- ❖ Beta thalassemia is more clinically relevant - point mutation in Ch 11, require lifelong treatment.
- ❖ Alpha thalassemia- deletion in Ch 16, mostly incompatible with post natal life (hydrops fetalis).
- ❖ Sickle cell disease- single nucleotide mutation in the HbA gene, Glutamic acid > valine at the 6th position.
- ❖ Chorionic villus sampling (CVS) is the gold standard invasive procedure for prenatal diagnosis.
- ❖ Experienced hands CVS is a safe procedure with overall foetal loss rate of 0.5-1.5%.

AIMS & OBJECTIVES

- ❖ To detect sickle cell disease and thalassemia major of the fetus.
- ❖ To analyse the results and outcome of pregnancies following Chorionic Villous Sampling.

MATERIALS & METHODS

- ❖ It is an ongoing, descriptive study of all women who had undergone CVS at SCB Medical College & Hospital, Cuttack from 1.6.2022 to 15.10.23 to rule out Sickle cell disease and Thalassemia major in the fetus.
- ❖ The procedure was done by USG guided trans abdominal route and retrieved chorionic tissue was sent for genetic study.

INCLUSION CRITERIA:

- ❖ Pregnant women with h/o previous child suffering from Sickle Cell Disease or Thalassemia Major.
- ❖ When Both Couples are either Sickle Cell Trait or Thalassemia Minor.
- ❖ When one of the partner is suffering from disease or trait.
- ❖ Obstetric history, family history, Indication for the procedure, gestational age at the time of CVS, complications and outcomes of the procedure is noted in all cases.

RESULTS: Maternal Characteristics

Variables		Frequency	Percentage
Indication	Thalassemia Trait	25	83.33%
	Sickle Cell Trait	4	13.33%
	Sickle cell dis.	1	3.33%
Socio-economical Class	Middle	11	36.67%
	Lower	19	63.33%
Background	Rural	21	70%
	Urban	9	30%
Parity	Para 0	3	10%
	1-2	25	83.33%
	3 or more	2	6.67%



RESULTS: Clinical parameters

Parameter	Number (n=30)	Percentage
Age (yrs)		
21-25	8	26.67%
26-30	10	33.33%
>30	12	40%
Gestational Age (Weeks)		
11-14 th	8	26.67%
15-18 th	19	63.33%
>=19	3	10%
Parameter	Number	Percentage
Successful in first attempt	27	90%
Failed	1	3.33%
Maternal tissue contamination > Repeat > Successful	2	6.67%

Out of 30 cases. 12 have delivered 6-
Vaginal delivery and 6 -LSCS

In 2 cases, termination was done as fetus was homozygous for thalassemia major.

1 case was presented as incomplete abortion at 24 weeks, after 10 wks of CVS.

1 case - CVS could not be done due to posterior placenta 14 patients continuing their pregnancy

CONCLUSION

- ❖ Transabdominal CVS is safe and reliable procedure for prenatal diagnosis in early pregnancy and should be considered as procedure of choice.
- ❖ In experienced hands, the miscarriage rate is very low.
- ❖ CVS offers options of management if pregnancy is affected.