GTPIM109

Invasive Prenatal Diagnostic Procedures

Patient Information Material

What is prenatal diagnosis?

Diagnosis of a disorder or deformity of a baby in the womb before birth is called prenatal diagnosis.

Why does a doctor request for invasive prenatal sampling tests?

The prenatal sampling procedures are done to diagnose genetic disorders or deformities in the unborn baby that cannot be diagnosed by ultrasound alone.

At what time of pregnancy these tests are done?

A doctor can request any of the following three procedures depending on gestation (weeks) of the pregnancy:

- Chorionic Villus Sampling (CVS): Biopsy of small chorionic villi / placental tissue at 10-12 week gestation.
- Amniocentesis: Removal of 20-30ml amniotic fluid at 16 week gestation
- Cordocentesis: Sampling of fetal cord blood at 18 weeks of gestation

Who does the procedure?

Experienced Sonologists / Radiologists or Obstetricians perform the procedure under aseptic conditions.

How is the procedure done?

All procedures are done under sterile conditions with ultrasound guidance. A sterile needle is inserted transabdominally and sample (amniotic fluid / CVS / Cordblood) is removed without disturbing the baby in the womb.

Is the procedure painful?

No normally, it does not hurt anymore than having an injection or a shot. The procedure is performed under a local anesthesia.

What are the pre-procedure instructions for patients?

There are no special instructions or restrictions for patients before the procedure.

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How long does the procedure take? Does it require hospitalization?

The sampling procedure usually takes 10mins. There is no need for hospitalization. However, the patient is advised to rest for15-30 mins after the procedure.

What care should the pregnant woman take after the procedure?

Rest for the day is recommended. Lifting heavy weights and long distance travel should be avoided immediately after theprocedure.

Is there any risk for pregnancy loss?

Rarely, a pregnant woman might experience spotting, cramping, pain and leakage of amniotic fluid within 1-3 days after sampling. It is strongly advised to call your doctor or near by hospital in such a rare situation.

How accurate are prenatal genetic tests?

Genetic tests (Chromosomal / DNA) have more than 98% sensitivity and accuracy.

What are the limitations of prenatal genetic tests?

Genetic tests are very specific in diagnosis. They do not diagnose fetal defects that are non-genetic in nature or that are sure to other untested genetic defects or mutations. Limitations are best explained by genetic counselors.

What happens in a genetic counseling session?

Counseling done by qualified genetic counselor focuses on gathering medical and family history explaining causes of genetic defects. Prognosis, modes of inheritance, prenatal testing details, including their benefits, risks and limitations are discussed with the family.

Can we know the sex of our baby?

NO. As per the Indian prenatal diagnostic act, fetal sex is not disclosed in genetic test reports.

When does an obstetrician request prenatal testing?

- There is a close relative or a previous child with a serious genetic condition
- One of the partners in a couple has a serious condition that may be passed on to a baby
- One or both parents are known 'carriers' of a particular faulty gene
- The mother is in her mid-30s or older (not necessarily her first pregnancy) as there is an increased risk for having a baby with chromosomal problems due to more or less than the usual number of chromosomes; eg. Down syndrome
- There has been exposure to some chemical or other environmental agent
- The results of screening tests such as ultrasound or first or second trimester screening may determine that the baby is at increased risk for a particular genetic condition in the pregnancy

GeneTech is a pioneer in medical genetics testing in the country and offers the most comprehensive testing and screening services for genetic disorders in India

GeneTech, 8-2-293/17/A25, Opposite KBR Park, Road No. 14, Banjara Hills, Hyderabad, TS - 500 034 (INDIA).
Tel: 040-23400932, 23406178 **Website:** www.genetech.co.in **Customer Care: 98480 41127**