

# Methods of Prenatal Diagnosis

| Methods                               | Procedure                                                                                                                                                                                      | Advantages and Risks                                                                       |
|---------------------------------------|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|--------------------------------------------------------------------------------------------|
| Chorionic villous sampling            | Transcervical or tranabdominal aspiration of trophoblastic tissue for karyotyping and genetic studies in pregnancies less than nine weeks                                                      | Early diagnosis and timely termination of pregnancy. 3% abortion risk.                     |
| Genetic amniocentesis                 | Aspiration of amniotic fluid under ultrasound guidance and culture and karyotyping of amniotic cells. Early - < 11 weeks; Late – 16-18 weeks                                                   | Early diagnosis and timely termination of pregnancy. 3% abortion risk. RH Isoimmunization. |
| Routine ultrasonography               | Performed at 16-18 weeks or earlier to diagnose structural anomalies of the early embryo-fetus                                                                                                 | Allows second trimester termination of pregnancy                                           |
| In vitro fertilization                | Cell taken from the earliest morula for karyotyping                                                                                                                                            | Allows termination of the implantation if abnormal                                         |
| Percutaneous umbilical blood sampling | PUBS refers to transabdominal aspiration of fetal blood from the umbilical vein for detailed analysis of fetal diseases by performing hematological, genetic, immunological etc fetal diseases | Allows the diagnosis of numerous fetal diseases                                            |

# Studies In Inherited Metabolic Diseases Prenatal And Perinatal Diagnosis

**Michael Hoelscher**



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