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# Prenatal diagnostic method

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**Abstract:** Prenatal diagnostic methods are becoming very popular these days. This method is based on the early detection of pregnancy- related problems, including potential genetic diseases. Prenatal diagnosis identifies all problems before birth, anatomical and physiological problems related to the health of the zygote, embryo or fetus. Prenatal diagnosis refers to the testing and evaluation of a fetus before birth to detect genetic, chromosomal, structural, or metabolic disorders. It helps in early identification of potential health issues, allowing for informed decision-making and possible interventions.

**Keywords:** Prenatal, scrining, amniotic liquid, ultrasound.

**Introduction:** Ethical and Psychological Considerations: Prenatal diagnosis raises ethical concerns, particularly regarding pregnancy termination, potential anxiety for parents, and the right to know versus not know. Genetic counseling is essential for informed decision-making.

Indications for Prenatal Diagnosis-Advanced maternal age (≥35 years)

-Family history of genetic disorders

-Previous child with a congenital anomaly

-Abnormal ultrasound findings

-Carrier status of genetic diseases (e.g., thalassemia, cystic fibrosis)

**Types of Prenatal Diagnosis**

1.Non-Invasive Techniques

-Ultrasound (USG): Used to detect structural abnormalities, fetal growth, and placental position.

-Maternal Serum Screening: Measures specific proteins and hormones in maternal blood to assess the risk of chromosomal disorders (e.g., Down syndrome).

-Cell-Free Fetal DNA Testing (NIPT): Analyzes fetal DNA in maternal blood to screen for chromosomal abnormalities like trisomy 21, 18, and 13.

2.Invasive Techniques

-Chorionic Villus Sampling (CVS) (10–13 weeks): Biopsy of placental tissue to diagnose genetic conditions.

-Amniocentesis (15–20 weeks): Extraction of amniotic fluid for genetic and chromosomal analysis.

-Percutaneous Umbilical Blood Sampling (PUBS): Direct sampling of fetal blood from the umbilical cord to diagnose blood disorders and infections.

Ultrasound Findings and Interpretation: Ultrasound is the most commonly used non-invasive prenatal diagnostic tool. It helps in: First-Trimester Ultrasound (6–13 weeks): Confirms pregnancy and gestational age, Detects multiple pregnancies (twins, triplets, etc.). Identifies major structural defects (e.g., anencephaly),Nuchal translucency (NT) scan (11–13 weeks): Measures fluid at the back of the fetal neck. Increased thickness suggests Down syndrome (Trisomy 21) or congenital heart defects

Second-Trimester Ultrasound (18–22 weeks): Anomaly scan (Level II scan): Detects congenital malformations (e.g., cleft lip, heart defects, neural tube defects like spina bifida). Evaluates placental position (e.g., placenta previa) and amniotic fluid levels.

Third-Trimester Ultrasound (28+ weeks): Monitors fetal

growth restriction (IUGR).Assesses fetal well-being (Doppler studies, biophysical profile)

In Uzbekistan Case Study: Prenatal Diagnosis of Aicardi and Andermann Syndromes: At the Republican Center "Screening and the Child" in Uzbekistan, a retrospective analysis was conducted on 21 pregnant women whose fetuses were diagnosed with agenesis of the corpus callosum. The study aimed to facilitate the early diagnosis of rare conditions such as Aicardi and Andermann syndromes through comprehensive dynamic examinations, including ultrasound and laboratory studies. This approach highlighted the significance of detailed prenatal assessments in detecting rare congenital anomalies. Aicardi syndrome and Andermann syndrome are rare genetic disorders, with Aicardi syndrome characterized by a specific triad of symptoms (chorioretinal lacunae, infantile spasms, and agenesis of the corpus callosum), while Andermann syndrome is associated with congenital heart defects, kidney anomalies, and diaphragmatic hernia.

In India Case Study 1: Prenatal Invasive Testing at a Tertiary Referral Center

A study conducted at a tertiary referral center in India evaluated 42 fetuses with increased nuchal translucency (NT) measurements (>95th percentile). Among these, 12 cases exhibited additional abnormalities. The study underscored the importance of comprehensive assessments, including chromosomal microarray analysis (CMA), to detect genetic abnormalities. However, limitations such as cost constraints affected the availability of CMA in all cases. Notably, in fetuses with increased NT but normal karyotype and anomaly scans, the survival rate was 95.6% (22 out of 23 cases). PMC

Case Study 2: Awareness of Prenatal Genetic Screening: A cross-sectional study assessed the awareness of prenatal genetic screening tests among pregnant women in India. The findings revealed significant disparities in knowledge, emphasizing the need for improved educational initiatives to ensure informed decision-making regarding prenatal testing.

## CONCLUSION

Prenatal diagnosis plays a crucial role in early detection, risk assessment, and management of fetal abnormalities. While non-invasive tests (USG, NIPT) are safe and widely used, invasive tests carry risks but provide definitive diagnosis. Ethical considerations highlight the importance of genetic counseling to help parents make informed choices. The prenatal diagnostic method is currently being carried out at an excellent level at the Mother and Screening Center of Uzbekistan, which is equipped with the latest medical equipment. We know that different populations live in all countries of the world. If we could widely use prenatal diagnosis in all populations, we would be able to prevent hereditary diseases and gene disorders that are likely to be observed in future generations.

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