Characteristics of pregnancies undergoing NIPT or amniocentesis at fetal-medicine center: A review

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ABSTRACT

Introduction: Non-invasive prenatal testing (NIPT) is increasingly accepted as a second-tier aneuploidy screening method, while amniocentesis retains its role in the era of non-invasive testing. Understanding the patterns and characteristics of both tests in a local context can help integrate NIPT into standard screening practices. **Materials and Methods:** This retrospective study analyzed mother and sonogram characteristics of pregnant women who underwent NIPT or amniocentesis at Hospital Sultan Idris Shah, Serdang over two years (2022-2023). Anonymous data were collected from electronic medical records and MFM clinic records. **Results:** There were 2,274 new cases in 2022 and 1,970 in 2023, with 125 NIPTs and 162 amniocentesis performed over two years. NIPT uptake increased. Mean maternal age was 34.59±5.64 years for NIPT and 31.13±5.36 years for amniocentesis. Gestation at the first visit was 18.27±6.24 weeks for NIPT and 24.38±5.64 weeks for amniocentesis. In the NIPT group, the reasons for referral were abnormal scan (26.6%), maternal age (23.4%) and history of fetal anomaly (14.1%). In the amniocentesis group, the reasons were abnormal scan (80.4%), maternal disease (4.3%) and multiple pregnancy (3.7%). In the NIPT group, 87.5% delivered at term, 6.3% preterm and 4.7% had miscarriage/IUD. In the amniocentesis group, 46.7% delivered at term, 13.3% preterm, 17.0% had miscarriage/IUD and 15.2% had termination of pregnancy. Main sonogram abnormalities in the NIPT group included brain (30.43%), kidneys/bladder (26.09%), and long bones (17.39%). In the amniocentesis group, the main abnormalities were cardiac (43.42%), brain (26.31%), and neck (12.5%). One out of 128 NIPT cases had low fetal fraction (0.78%). Fetal fraction positively correlated with gestational age and negatively with maternal age and BMI. No correlation was found with parity. **Conclusion:** NIPT uptake increased, serving as an adjunct stratification for advanced maternal age and soft markers with low relative risk. Fetal fraction in NIPT correlates with maternal age, BMI and gestation.