A structurally normal fetus at the 11- to 14-week ultrasound does not guarantee a newborn without congenital anomalies: a cohort study

Sarti Junior AM, Oliveira Junior RE, Duarte G, Berezowski AT, Okido MM, Marcolin AC.

Background

- Congenital anomaly (CA) is an important cause of infant mortality and a significant reason for long-term loss of human potential among survivors worldwide.
- Even in fetuses with normal karyotypes and structures, abnormal first-trimester sonographic markers raise suspicion of a wide range of fetal defects and increase perinatal morbidity and mortality.
- Therefore, the first trimester assessment of aneuploidy risk can facilitate the prediction of adverse perinatal outcomes, including CA.

Objectives

- To analyze the influence of maternal variables and abnormal first-trimester sonographic markers on CA at birth when the fetus is structurally normal at the 11- to 14-week ultrasound.

Metodologia

- This cohort study involved a first-trimester ultrasound scan in 310 high-risk women undergoing routine antenatal care. All of them were admitted to the University Hospital of the Ribeirão Preto Medical School, State of São Paulo, Brazil.
- Detailed assessments of fetal anatomy and first-trimester sonographic markers were performed at 11-14 weeks of pregnancy by three sonographers who had the appropriate Fetal Medicine Foundation Certificate of Competence in this assessment.
- Nomograms for the 5th, 50th, and 95th percentiles were created for the nuchal translucency (NT) thickness and ductus venosus (DV) PIV values.
- Multilevel regression analysis was used to determine the effects of maternal variables and abnormal first trimester sonographic markers on the incidence of CA at birth.

Resultados

- Three hundred ten patients were evaluated, and 41 patients (13.2%) had an anomalous newborn.
- Congenital heart defects were the most common type of CA (36.4%), followed by: urinary system (12.1%), central nervous system (12.1%), multiple malformations (9%), musculoskeletal anomalies (6.1%), lung defects (6.1%), gastrointestinal tract (3%), and others (15.2%).
- The mean NT thickness of the study population was 1.73±0.72 mm (95th percentile= 2.5 mm). The mean NT of the newborns with CA was 2.0 mm (0.7 – 8.9 mm).
- The mean DV PIV was 0.98±0.15 (95th percentile= 1.22).
- Table 1 shows the effects of maternal characteristics and abnormal first trimester sonographic markers on the incidence of CA at birth.

Conclusions

- Increased NT and/or high adjusted risks for trisomy 21, 18, and 13 increase the risk of CA at birth, even in fetuses with a structurally normal 11- to 14-week ultrasound scan.