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Fetuses with increased nuchal translucency and normal karyotype

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Objective

To identify the causes of increased nuchal translucency thickness within chromosomally normal fetuses.

Methods

We reviewed the outcome of all pregnancies presenting at the department of gynecology and obstetrics A in the hospital of Charles Nicolle in Tunis-Tunisia with increased NT between October 2019 and October 2022. Fetal karyotyping and ultrasound investigation at 11-14 weeks gestation were offered in all cases. The nuchal translucency was defined as the thin translucent area lying between the inner surface of the skin and the soft tissue interface overlying the cervical spine. The maximal nuchal translucency thickness was measured on a sagittal section of the fetus and the measurement was considered as enlarged when it was ≥ 95th. Cystic hygromas were not included under the definition of nuchal translucency An adverse outcome was defined as miscarriage, intrauterine death, termination of pregnancy at parental request or the finding of one or more structural defects or genetic disorders.

Results

A total of 29 fetuses with increased NT were observed in this period. A chromosomal anomaly was detected in 19 (66%) fetuses. In 34% (10/29) fetuses, the karyotype was normal. The average maternal age was 30.6 years ±5.7. Anomalies were detected, at the time of ultrasound or after birth of these fetuses: cardiac defects (3 cases), diaphragmatic hernia (2cases), 1 Megacystis (1case), body stalk anomaly(1case), congenital adrenal hyperplasia(1case), Omphalocele (1case) and spina bifida (1case). A spontaneous abortion or an intrauterine death occurred in 2% and in 1% of these fetuses, respectively.

Conclusion

Even in the absence of aneuploidy, increased fetal nuchal translucency has proven to be a marker of fetal cardiac malformations, many other fetal malformations and genetic syndromes. Increased nuchal translucency could constitute an indication for a specialized fetal echocardiography.