Screening for Down syndrome

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Objective

To study the strategies of prenatal screening for Trisomy 21.

Methods

Retrospective study was conducted at the service "A" of the Maternity and Neonatology Center of Tunis during a period of 10 years. We included all cases of Trisomy 21 diagnosed prenatally that were either had pregnancy termination or resulted in live births. We searched screening methods used and we evaluated their performance.

Results

The overall prevalence of Trisomy 21 was 1. 67 per 1000 pregnancies. Prenatal screening was carried out in 46. 5% of patients. The diagnosis of Trisomy 21 was made prenatally in 84. 9% of cases. The mean maternal age was 36. 1 years and the mean gestational age at diagnosis was 17 weeks. In the first trimester ultrasound screening led to the diagnosis in 54. 5% of cases with a sensitivity of 61. 1%. Maternal serum markers (free β hCG and PAPP-A) contributed to the diagnosis in 9. 1% of the cases. Second trimester screening by free β hCG, AFP and uE3 was carried out in 24. 3% of the cases, with a sensitivity of 75%. Second trimester ultrasound scan detected 33% of fetuses with T21.

Conclusion

Prenatal screening for Trisomy 21 is constantly improving. Ultrasound screening (especially that of the first trimester) is an important pillar in screening strategies. This fact encourages us to improve its quality by training professionals involved. We also should ensure the availability of biomarkers for all women.