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Aneuploidy screening in women of advanced age in a limited resource public health care setting

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Objective

Prenatal screening in women of advanced maternal age has been shown to be ineffective in Cape Town. This study determines the uptake rate of prenatal screening and invasive testing by pregnant women over the age of 37, as well as factors which influence this.

Methods

This is a retrospective audit of prospectively collected data from all women over 37 years seen at the Tygerberg Hospital fetal medicine unit in 2016. Invasive genetic testing was offered for age over 40 years, for a high ultrasound-based risk for aneuploidy or a fetal anomaly on ultrasound. Women over 40 received formal pre-screen counseling when feasible. Pregnancy termination (TOP) was offered for severe structural anomalies or confirmed genetic disorders.

Results

Of 1196 older women, 640 (53.5%) were offered invasive testing according to our protocol. Only 114 (9.5% of all women, 17.8% of those offered an invasive procedure) accepted this and an additional 10 women opted for TOP without invasive testing. Women older than 40 years (75.8 vs 59.6%), women who had previous first trimester losses and women who received pre-screen counselling (21.2% vs 7.9%) were more likely to decline invasive testing. The most frequent reason for declining was the parents' decision that they would not consider TOP (75%). A higher adjusted risk for trisomy 21 and a less favourable risk adjustment had a significant effect on the acceptance rate of testing in both first and second trimester assessments. Acceptance was highest when a fetal anomaly was detected (54%). Ultrasound-based risk reduction was more effective in the first compared to the second trimester assessment (92% versus 64 % becoming low risk) but most assessments were performed, and most aneuploidies and anomalies were detected in the second trimester.

Conclusion

The uptake of invasive testing in this population was low (17.8%). It was somewhat influenced by maternal characteristics (age, previous miscarriage) and the ultrasound-based risk result, but more so by the provision of pre-screen counselling (45% opted out) and predominantly by the women's pre-existing attitude towards the service, towards trisomy 21 or TOP in general. The cost of routine serum screening does not seem justified in this population but the expansion of pre-screen counselling services and first trimester ultrasound screening can improve the cost effectiveness of this screening program. Second trimester ultrasound screening will continue to be the mainstay of prenatal genetic screening in this population as it provides access to screening for women who are referred or who initiate antenatal care after the first trimester and makes a significant contribution to the detection of aneuploidies and structural anomalies.