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Comparing ultrasound findings to prenatal invasive test diagnosis in a regional hospital in south africa

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Objective

Invasive prenatal testing is the gold standard of prenatal diagnosis of chromosomal abnormalities. In the South African public sector biochemical screening and non-invasive prenatal testing is not available, methods of screening mainly rely on advanced maternal age and antenatal ultrasound. The aim of this study was to compare the accuracy of ultrasound screening with the cytogenetic diagnosis found at prenatal invasive testing at Rahima Moosa Mother and Child Hospital (RMMCH) from January 2014 to May 2016. Secondary objectives included the evaluation of invasive prenatal testing in terms of indications, complications and pregnancy outcome.

Methods

The study took place at RMMCH, a regional academic hospital in Johannesburg which performs approximately 12 000 deliveries annually. Charts were reviewed retrospectively for patients who underwent invasive prenatal testing.

Results

Ninety-seven patients were identified, and 96 results obtained. The main indication for invasive prenatal testing was abnormal ultrasound findings. In total, 12. 5% of the test results were abnormal, including two patients with Trisomy 13, two with Trisomy 18, two with Trisomy 21, two with Klinefelter syndrome, one with a balanced translocation, one with cystic fibrosis, one with spinal muscular atrophy and one with Wolf-Hirschorn syndrome. In this sample eight patients (67%) had abnormal ultrasound findings which included several soft and hard markers. In the sample of 97 patients, there were thirteen patients with major structural anomalies at ultrasound alluding to a possible outcome of structural abnormality at birth. Six of these patients had fetuses with chromosomal abnormalities confirmed at karyotyping.

Conclusion

It is expected that in a resource restricted area where biochemical screening is not available that advanced maternal age and ultrasound findings are the main reasons to lead to invasive prenatal testing. The rate of abnormalities found is higher than internationally quoted. The study shows that an invasive testing service can be successfully run in a resource restricted setting with mainly ultrasound as screening test but ongoing education of the availability of the service in the public sector is needed.