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Retrospective analysis of the outcomes of patients presenting for genetic counselling with fetal abnormalities

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ABSTRACT

Fetal abnormalities are congenital abnormalities that are identified prenatally, which may be structural or functional in nature. Genetic counselling is a non-directive and non-judgmental process of information-giving, at the same time as providing psychosocial support. It is offered to women and their partners who have a fetal abnormality detected during pregnancy. When a fetal abnormality is detected, the patient can sometimes be offered a termination of pregnancy, and the decision of whether or not to continue the pregnancy is made by the patient.

The first aim of this research was to conduct an audit of the genetic counselling service provided by the Division of Human Genetics, NHLS and WITS, in order to assess the level of service being offered to patients with diagnosed fetal abnormalities. The second aim of the research was to determine what factors, if any, influenced the decision patients made regarding whether to continue or interrupt their pregnancy.

One hundred and seventy one files of women, who received genetic counselling for an identified fetal abnormality during pregnancy from the division between 2002 and 2006, were included in the retrospective clinical audit.

The patients seen for genetic counselling represent 1.1 % of the estimated number of women in Johannesburg who could have had abnormalities detected prenatally, based on the prevalence of congenital disorders in the area and an ultrasound prenatal detection rate of 56.2 %. Two thirds of patients who were offered TOP chose to terminate their pregnancy. The most clinically significant predictor of the decision to terminate an affected pregnancy was found to be an earlier gestation at offer of TOP, which suggests that earlier detection and diagnosis of abnormalities is beneficial to patients. Overall, 62 % of patients were not offered genetic counselling follow-up appointments after conclusion of their pregnancy. The genetic counselling service offered to patients thus needs to be improved, in particular, the follow-up service patients receive after TOP or delivery is not adequate.