Prenatal AMA diagnosis has traditionally been performed by cytogenetic analysis of cultured fetal cells, which is time-consuming (7 - 21 days) and expensive (R2 400, NHLS rate). Quantitative fluorescent polymerase chain reaction (QF-PCR) analysis is now recognised as a viable alternative to cytogenetic analysis for the rapid detection of autosomal aneuploides.[1]

QF-PCR is a molecular (DNA)-based test performed on uncultured fetal cells. Used in the Laboratory of the Division of Human Genetics it is able to detect the common numerical chromosomal abnormalities of chromosomes 21, 18, 13, X and Y. Trisomies 13, 18 and 21 are detected with about 99% accuracy, usually within 48 - 72 hours and at a cost of R1 197 (NHLS rate).

QF-PCR has limitations. It cannot yield accurate results when there is maternal contamination of amniotic fluid or, less commonly, when chromosomal imbalances such as low-level mosaicism (<30%), some types of polyploidy, and structural chromosomal abnormalities (deletions, translocations and ring chromosomes) are present. It detects extra chromosome 21 material present in cells with unbalanced translocations of chromosome 21, but does not identify the problem as a translocation. These shortcomings are, however, put into perspective by a multi-centre audit of 23 genetic laboratories, which found that, in over 98 000 amniocenteses performed after Down syndrome screening, only about 1% of autosomal chromosome abnormalities were not detectable by QF-PCR.[2]

In industrialised countries, newer technologies such as QF-PCR are considered as optional extras to routine cytogenetic analysis for prenatal diagnosis.[1] However, in resource-limited developing nations such as South Africa, we believe that QF-PCR can be the standard diagnostic technique.

Therefore, at the Division of Human Genetics, QF-PCR will now be done instead of conventional cytogenetic analysis in cases where AMA is the only indication. Should abnormalities be detected on sonar or other specific indications, full karyotyping can still be requested and performed. Pretest counselling of AMA women should take this policy change into consideration; a *pro forma* consent form for amniocentesis that does so, is available from the Division of Human Genetics.

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Of HIV, grief and TOP

To the Editor: People living with HIV are frequently people living with very high levels of grief. They grieve over the loss of life expectancy and of their own dreams for the future. They are often grieving over the loss of a spouse or a child. In our practice, they are frequently grieving over the loss of other close relatives. This makes them emotionally very vulnerable people.

At first sight, the offer of TOP to such people in early pregnancy may seem to be a compassionate way to avoid further grief from infant losses, and to avoid increasing the number of orphans in our nation. Yet there is strong evidence that a decision for TOP may precipitate a severe grief reaction of its own. Such grief has been associated with a 7-fold increase in suicide and homicide,[1] and a 180% increase in psychiatric illness, in the year following TOP in first-world countries with excellent access to health care.[2,3] In South Africa, with the generally poor access to psychiatric care and high levels of violence, the effects can be expected to be far greater. The effects of maternal depression, of resorting to substance abuse or of the development of self-destructive behaviour, may completely negate the advantages to the family of not having another baby to nurture.

This question raises the need for very skilled and careful counselling of women with HIV before TOP is offered, and very careful follow-up and emotional support following TOP, should they choose that option. It also makes even stronger the case for ready access to HAART.

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