

## Comment by the ACC on Rapid Prenatal Detection of Aneuploidy

April 29, 1999

### Introduction

by Lorraine Gaunt, ACC Secretary

*Dear Colleague,*

Re: Rapid prenatal detection of aneuploidy

As a consequence of advertising by companies supplying reagents and by private laboratories, targeted directly at Obstetricians, several Cytogenetics centres have been asked regarding the current availability of prenatal aneuploidy screening using FISH and PCR based techniques, and have sought guidance from the ACC Council. Although Council has no direct influence on provision of services, it is aware that these techniques are still undergoing formal evaluation in the UK and that most centres are not funded to provide them. As such, it was felt that it could be beneficial if there was some degree of uniformity in response to approaches from Obstetricians and others.

Although individual laboratories will need to react in accordance with their own particular priorities and circumstances they may find it helpful to incorporate parts or all of the enclosed document, as a useful form of wording describing the current situation, in any response to outside enquiries.

**Please note that this is not intended to be an ACC Council Statement on the efficacy or desirability of these techniques.**

Furthermore, it is not intended to pre-empt or comment on ongoing evaluations, or directly influence decisions regarding provision of services, responsibility for which remains at the local purchaser/provider level.

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## Rapid prenatal detection of aneuploidy

### A comment from the ACC Council

Many years' experience has shown conventional karyotyping of cultured amniocytes to be a reliable, cost effective and highly accurate means of prenatal diagnosis of a wide range of chromosome abnormalities, its only disadvantage being the period during which cells need to be cultured prior to analysis.

FISH and PCR based strategies offer the prospect of more rapid prenatal diagnosis, avoiding the need for in vitro cell culture, clearly a potential benefit to patients and all others concerned. Several commercial kits are now available, but presently all have the disadvantage that inherent technical limitations, combined with the need to contain their price within acceptable limits, restrict their scope to the accurate detection of non-mosaic forms of the common aneuploidies, trisomies 13, 18 and 21 and numerical sex chromosome abnormalities.

As this group represents only 70-75% of abnormalities detected by routine karyotyping, the manufacturers of these kits emphasise their use as adjuncts to conventional analysis, not as replacements. Indeed, these products are licensed for use in the United States only under these specific conditions.

In addition, unlike conventional karyotyping, these techniques are not recommended by the manufacturers for use with small or blood stained samples, often with such sub-optimal cases being excluded from the apparent high accuracy figures quoted in advertising literature.

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The value of these rapid screening techniques is currently under assessment [NHS Executive Research and Development Programme Health Technology Assessment (HTA)], the recommendations of which are awaited by both purchasers and providers of cytogenetic facilities within the UK. In the interim, driven by marketing considerations, several manufacturers and private laboratories are targeting obstetricians and cytogeneticists directly, encouraging them to offer these screening tests to patients prior to the outcome of this HTA evaluation.

On occasions, this extends to suggesting, by default, their use as replacements for conventional karyotyping, an option which falls outside all current and likely acceptable standards of practice.

Used as an adjunct to conventional karyotyping, such rapid screening techniques would add approximately 40-50% to the laboratory cost of processing amniotic fluid samples. Selective use of rapid screening for certain categories of high risk patients would disproportionately increase these additional laboratory unit costs due, among other factors, to the high initial capital investment in equipment which will be required for their introduction.

Beyond this, there may be further cost implications in relation to extra counselling time and any additional antenatal clinic appointments required for results to be delivered in two stages.

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Although rapid testing for common aneuploidies clearly offers advantages to some groups of patients, its widespread applicability and its desirability as a screening technique for use for all current prenatal diagnostic referrals, remains under review. At the moment, until appropriate HTA recommendations and follow-up funding are in place, these tests are unlikely to be generally

available in NHS Diagnostic Cytogenetic Laboratories.