What is already known on this topic

Measurement of fetal nuchal translucency (NT) thickness provides effective first trimester screening for trisomy 21

Subsequent analysis of those at high risk by full karyotyping of chorionic villous samples detects all clinically significant chromosomal abnormalities

Analysis by quantitative fluorescent polymerase chain reaction (qf-PCR) is cheaper and faster but does not detect all significant abnormalities

What this study adds

Analysis of samples by qf-PCR in all cases and full karyotyping only in the 10% of cases with fetal NT of 4 mm or more identifies 99% of significant chromosomal abnormalities

This strategy costs 60% less than full karyotyping for all

observation of failed diagnosis in about 1% of cases may be an overestimate because about half of these cases either miscarried or had major abnormalities detected by the routine second trimester scan.

In summary, a policy of initially doing rapid aneuploidy exclusion for chromosomes 13, 18, 21, X, and Y in the first trimester, with full karyotyping only if the fetal NT thickness is increased, would reduce the economic costs of a first trimester screening programme for trisomy 21, allowing for rapid delivery of results and minimising parental anxiety and the risk of unnecessary termination of pregnancy.

Contributors: See bmj.com.

Funding: This study was supported by a grant from the Fetal Medicine Foundation (Charity No 1037116). The Fetal Medicine Foundation did not have any role in study design, in collection, analysis, and interpretation of data, in the writing of the report, or in the decision to submit the paper for publication.

Competing interests: None declared.

Ethical approval: King's College Hospital Ethical Committee approved the study.

- 1 Pertl B, Yau SC, Sherlock J, Davies AF, Mathew CG, Adinolfi M. Rapid molecular method for prenatal detection of Down's syndrome. *Lancet* 1994;343:1197-8.
- 2 Verma L, Macdonald F, Leedham P, McConachie M, Dhanjal S, Hulten M. Rapid and simple prenatal DNA diagnosis of Down's syndrome. *Lancet* 1998;352:9-12.
- 3 Mann K, Fox SP, Abbs SJ, Yau SC, Scriven PN, Docherty Z, et al. Development and implementation of a new rapid aneuploidy diagnostic service within the UK National Health Service and implications for the future of prenatal diagnosis. *Lancet* 2001;358:1057-61.
- 4 Levett LJ, Liddle S, Meredith R. A large-scale evaluation of amnio-PCR for the rapid prenatal diagnosis of fetal trisomy. *Ultrasound Obstet Gynecol* 2001;17:115-8.
- 5 Evans MI, Henry GP, Miller WA, Bui TH, Snijders RJ, Wapner RJ, et al. International, collaborative assessment of 146,000 prenatal karyotypes: expected limitations if only chromosome-specific probes and fluorescent in-situ hybridization are used. *Hum Reprod* 1999;14:1213-6.
- 6 Department of Health. *Our inheritance, our future: realising the potential of genetics in the NHS*. London: Stationery Office, 2003.
- 7 Adinolfi M, Sherlock J. Prenatal detection of chromosome disorders by QF-PCR. *Lancet* 2001;358:1030-1.
- 8 Snijders RJM, Noble P, Sebire N, Souka A, Nicolaides KH. UK multicentre project on assessment of risk of trisomy 21 by maternal age and fetal nuchal translucency thickness at 10-14 weeks of gestation. *Lancet* 1998;351:343-6.
- 9 Souka AP, Von Kaisenberg CS, Hyett JA, Sonek JD, Nicolaides KH. Increased nuchal translucency with normal karyotype. *Am J Obstet Gynecol* 2005;192:1005-21.
- 10 Grimshaw GM, Szczepura A, Hulten M, MacDonald F, Nevin NC, Sutton F, et al. Evaluation of molecular tests for prenatal diagnosis of chromosome abnormalities. *Health Technol Assess* 2003;7(10):1-146.
- 11 Leung WC, Lam YH, Wong Y, Lau ET, Tang MHY. The effect of fast reporting by amnio-PCR on anxiety levels in women with positive biochemical screening for Down syndrome—a randomized controlled trial. *Prenat Diagn* 2002;22:256-9.
- 12 Daniel A, Hook EB, Wulf G. Collaborative US data on prenatal diagnosis for parental carriers of chromosomal rearrangements: risks of unbalanced progeny. In: Daniel A, ed. *The cytogenetics of mammalian autosomal rearrangements*. New York: Alan R Liss, 1988:73-162.

(Accepted 18 November 2005)

doi 10.1136/bmj.38730.655197.AE

bmjlearning.com

Learning for consultants

BMJ Learning offers online learning resources to help doctors stay up to date in a variety of clinical and non-clinical topics. So far we have concentrated on general practitioners and junior doctors, but we have now launched a new group of modules for consultants.

Hospital consultants are a disparate group, so how can we provide learning for everyone? We feel that there are generic topics that nearly everyone will be interested in. For example, the new module on how to set up a course offers practical guidance for would-be educators. According to Ben Turney, who runs courses frequently, "any course will have its own special features, but there are some underlying principles that will ease the organisation of any course." The key seems to be organisation, and the module gives detailed advice on everything from catering to costs.

We have also had many requests for "bite sized" clinical material and so have a growing number of learning resources on important clinical topics such as emerging resistance to antibiotics in *Helicobacter pylori*. If standard treatment has failed to eradicate the bug in one of your patients then you will probably first think of concordance. But another common reason for failure of treatment is resistance to one of the antibiotics used—mainly clarithromycin and metronidazole. Resistance to

tetracycline and fluoroquinolones is also emerging. We are delighted to publish this module in collaboration with the journal *Gut*.

When you have successfully worked through a learning module you can print out a certificate of completion. You can gain personal credits for continuous professional development by completing our new learning modules. If you have any other burning learning needs or desires then please let us know. Meanwhile, have a look at the current content—just click on www.bmjlearning.com, and off you go.

Kieran Walsh *clinical editor, BMJ Learning*
(bmjlearning@bmjgroup.com)

We welcome articles up to 600 words on topics such as *A memorable patient, A paper that changed my practice, My most unfortunate mistake*, or any other piece conveying instruction, pathos, or humour. Please submit the article on <http://submit.bmj.com>. Permission is needed from the patient or a relative if an identifiable patient is referred to. We also welcome contributions for "Endpieces," consisting of quotations of up to 80 words (but most are considerably shorter) from any source, ancient or modern, which have appealed to the reader.