Medical genetics: advances in brief

Effect of detailed fetal echocardiography as part of routine prenatal ultrasonographic screening on detection of congenital heart disease Stumpflen I, Stumpflen A, Wimmer M, Bernaschek G. *Lancet* 1996;348:854-7.

Anomaly scanning has become a standard investigation in many obstetric centres during the second trimester of uncomplicated pregnancies. Scanning of the fetal heart requires additional time and expertise and is not routinely included. Those women thought to be at higher risk of fetal cardiac anomalies are frequently offered additional fetal echocardiography. Stumpflen and colleagues have examined the value of offering detailed fetal echocardiography to all pregnant women. A total of 3085 women were screened including 540 who would have been offered fetal echocardiography on the basis of being at high risk because of a family history of congenital cardiac disease or coexisting maternal disease. A further 364 cases would also have been considered at high risk because of other detected fetal anomalies. Forty six fetuses were noted to have congenital cardiac disease, 15 (33% of the total) in pregnancies without specific risk factors. Three cases were found in the group with maternal risk factors and 28 in fetuses with other abnormalities. Six minor cardiac abnormalities were detected postnatally. Fetal karyotyping was undertaken in all but one of the sonographically detected abnormalities and in 17 (37%)

a chromosome abnormality was diagnosed. Four cases of chromosome abnormality were detected only because of the cardiac findings. The incidence of cardiac defects in the group with maternal risk factors was lower than expected (5.6 per 1000 compared to general population risk of 8.0 per 1000), which may be explained by small numbers. The authors suggest that because of this and the large proportion of abnormalities detected in pregnancies without risk factors (about half of which would have been missed on a four chamber anomaly scan view) fetal echocardiography should be considered as a possible screening test for all pregnancies. The possibility of offering such a test to all pregnant women would have considerable resource implications.

ANGELA BARNICOAT

Molecular genetic tests as a guide to surgical management of familial adenomatous polyposis

Vasen HFA, van der Luijt RB, Slors JFM, *et al.* *Lancet* 1996;348:433-5.

Surgical treatment of familial adenomatous polyposis (FAP) may be by restorative proctocolectomy or alternatively by colectomy with ileorectal anastomosis. The former approach has a higher morbidity but the advantage of removing all the mucosa, avoiding later recurrence, the latter treatment is associated with a risk of later development of

rectal polyps and carcinomas, but usually has satisfactory functional results. These authors review experience in The Netherlands of 225 patients who had an ileorectal anastomosis for FAP following a colectomy and have been followed up for at least a year. They propose that the location of the mutation within the adenomatous polyposis coli (APC) gene could be important in decisions about surgical management. Mutations in the APC gene were detected in 87 of the 225 patients studied; 72 were before codon 1250 and 15 after. Mutations before codon 1250 have been associated, by other authors, with a less severe phenotype, with a later onset of disease and fewer polyps, than those after this codon. Vasen *et al* found that the risk of having further surgery increased with time after a primary procedure, rising to 42% of patients 20 years after colectomy with ileorectal anastomosis requiring rectal excision. The risk of further surgery was clearly higher in the group with downstream mutations to codon 1250, and significantly different from those with mutations before codon 1250. Nonetheless there was a risk of nearly 40% of patients at 20 years requiring rectal excision even in the latter group. Information about molecular pathology may be important in making decisions about surgical management in FAP. However, there are other factors which are likely to be important in this decision making, as even in the group with a lower risk based on the genetic findings a significant proportion will require further surgery.

ANGELA BARNICOAT