What’s in an “increased risk of NTD” report?

A raised maternal serum alpha-fetoprotein (AFP) value in mid-pregnancy is always a matter for concern. The association between raised AFP values and fetal neural tube defects is perhaps the most immediate which comes to mind. However, a number of other conditions may also be indicated by a raised maternal serum AFP.

Some of these are relatively benign; others are more serious. In all cases, the appropriate next step is a detailed ultrasound examination of the pregnancy. In around 9% (1 in 11) of cases there will be something visible on ultrasound to account for the raised AFP result. In the five years to 31st December 1999, a maternal serum AFP more than twice the median value for the population (i.e. more than 2.0 MoM) was found in 1976/67965 (2.9%) of pregnancies screened by the SAMSAS Programme. Sixty one of these (3.1%, or 1 in 32) were affected by a fetal neural tube defect; 32 anencephaly, 27 meningomyelocoele and 2 encephalocoele. (For the characteristic maternal serum analyte patterns associated with these conditions see the SAMSAS booklet “Information for Health Professionals”. Call (08) 8204 7285 if you have not received this).

In 48 pregnancies (2.4%, or 1 in 41) fetal death had occurred and in a further 19 pregnancies (0.96%, or 1 in 104) the fetus had another identifiable and serious condition; exomphalos (5), gastroschisis (9), triploidy (2), trisomy 13 (2), Turner syndrome (1). Overall in second trimester, 1 in 16 pregnancies in which there was a high maternal serum AFP had a fetal death in utero or a fetus with a serious abnormality at the time of screening. The combination of biochemical and ultrasound screening effectively detected these. Less serious were 47 (2.4%, or 1 in 42) unsuspected twin pregnancies. Finally, there were 75 pregnancies (3.8%, or 1 in 26) in which the raised maternal serum AFP was associated with a later fetal death in utero, missed abortion, premature rupture of membranes, or stillbirth. Without ultrasound signs, however, there was nothing to distinguish these pregnancies from the majority singleton pregnancies (1726/1976, or 87.4%) with a raised maternal serum AFP who went on to deliver a normal infant.
In summary, a report from the SAMSAS Programme stating “increased risk of NTD (1:30)” means there is a 1:30 chance the fetus has an NTD. However, it also means there is a 1:11 chance that ultrasound examination will reveal either an unsuspected multiple pregnancy or a significant problem for which intervention and counselling will be necessary.

For those of you who practice within postcodes 5000-5999, the SAMSAS request form can be used with your Medical Director software. Click on under ‘Obstetrics’, enter the details as you normally do, and insert a SAMSAS request form in your printer. All necessary details will be transferred. If you need more SAMSAS request forms, and their accompanying pre-test counselling booklets, call us on (08) 8204 7285. We will dispatch them to you directly.

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