Prenatal diagnosis: What to tell the parents?

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The authors of the article “Prenatal Diagnosis of Facial Clefts: Evaluation of a Specialised Counselling” [1] present an elegant study documenting the favourable psychological effect of prenatal diagnosis of this defect. Parents valued the counselling, however unpleasant the initial message. They welcomed the fact that they were prepared at the birth for what was going to come and the security of an already arranged treatment pathway.

In general it can be stated that the beneficial effects of this type of prenatal diagnosis of non-acute disease are felt in a wide spectrum of congenital diseases, such as omphalocele, gastroschisis, renal disease, adenomatoid cystic disease of the lung, and brain malformations. Forewarned parents are prepared at birth and the acute shock of discovery is cushioned; they are already acquainted with the attending specialists and can enjoy the relative comfort of a well designed plan of campaign for their child. The knowledge that there is treatment available, and that all is not lost, is a highly important factor in acceptance of the newborn's medical problem.

Prenatal diagnosis plays an even greater role in the diagnosis of acute life-threatening disease such as severe congenital heart disease and brain malformation. In this category there is always the option of termination of the pregnancy, but, more importantly, prenatal diagnosis may contribute to a better overall outcome for the patient if the pregnancy is continued. Bonnet et al. have shown that survival in transposition of the great arteries is higher in the group with prenatal diagnosis, and we have shown that a planned delivery in a centre geared to the treatment of patients with congenital heart disease may ensure a better long-term neurological outcome. Both facts can be attributed to the better general preoperative condition of these babies, and hypoxic lactacidosis can be prevented by an immediate start with low-dose prostaglandin and ventilatory support. This means that the neonates arrive at surgical correction in better condition and preoperative neurological damage is minimised. This, and the obstetricians’ increased awareness of the anatomy of the 4-chamber and long axis view of the foetal heart, has resulted, in the Lausanne region, in prenatal referral of 30% of all foetuses with congenital heart disease.

An encouraging sign is that the growing experience of obstetricians and perinatologists is reflected in the larger number of referrals for prenatal congenital abnormalities. In congenital heart disease this is not the result of an increase in maternal indications (mother or sibling with congenital heart disease, mother with diabetes or SLE, toxic exposure of the mother), but of widening of the foetal indications (presence of an organ or chromosomal abnormality, foetal arrhythmia or foetal hydrops, or hydramnion of unknown origin) with the increased risk in the presence of a nuchal translucency.

Finally, a remarkable fact in the article of Hohlfeld et al. is that in two cases the referring obstetrician did not tell the parents about the diagnosis of the congenital defect (facial cleft). In this day and age one wonders whether this is a desirable policy. We feel strongly that if parents accept and pay for prenatal evaluation of the foetus they have the right to be informed and the attending physician is under an obligation to inform them, whether the news is good or bad. Hopefully this article, which highlights the positive aspects of prenatal diagnosis and information, may contribute to full briefing of all parents of foetuses undergoing prenatal ultrasonographic evaluation.

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