Ultrasound Diagnosis of Fetal Malformations

A. Kurjak and Lj. Zergollern-Cupak

In perinatal medicine, probably no single assessment will ever be more important than the early detection of malformed babies. The results obtained by ultrasound in many centres have been remarkable in this particular area. Table 1 presents fetal malformations detected over an 8-year period using a combination of ultrasonic and cytogenetic diagnosis.

Table 1. Fetal Malformations Detected during an 8-year Period (1974-1981)

Anencephaly	56
Hydrocephaly	31
Hydranencephaly	1
Spina bifida	6
Microcephaly	6
Intrathoracic cyst	3
Liver cyst	2
Duodenal atresia	5
Urinary tract obstruction	4
Potter's syndrome	3
Polycystic kidney disease	3
Omphalocele	2
Thanatophoric dwarfism	1
Sacrococcygeal teratoma	1
Short-limbed dysplasia	3
Miscellaneous multiple anomalies	25
Adenoma of fetal thyroid	1
Hygroma colli	2
Tunner syndrome	2
TOTAL	162

This report is based on the study of over 2700 women who are at high risk for having an abnormal fetus. The ultrasonic centre in Zagreb is specialised in the detection or exclusion of fetal malformations and patients from all over the country come for this examination. In most of the detected cases early amniocenteis was performed and the results analysed, together with ultrasonic findings, whenever it was necessary.

As may be seen from Table 1, the most frequently occurring malformations are those of cranial defects. Interestingly enough spina bifida is a very rare abnormality in our lab. material. It has to be stressed however that this Table shows only isolated spinal defects and not spi-

na bifida associated with other serious cranial defects. This group of malformations was detected among 60,000 pregnant women examined at our centre in Zagreb. A total of 7 doctors participated in the detection but 80% were detected by the most experienced among them.

Cranial defects

Here, ultrasound is the most successful method. It is now generally accepted that not a single anencephalic baby should be missed even by a beginner. The early diagnosis of hydrocephaly is important for the further prognosis and treatment of the affected baby. Early diagnosis is made upon the finding of dilated ventricles. To achieve this high-risk women should be given at least two examinations prior to 24 weeks of gestation. Unlike hydrocephaly and anencephaly, microcephaly is still surrounded by a lot of controversy. Diagnosis of microcephaly may be made more confidently with the serial measurement of fetal head and abdomen and early recognition of abnormal growth patterns. There are a variety of structures which are connected with the head but are clearly extracranial. In our centre, fetal thyroid tumour and cystic hygroma were successfully diagnosed.

Abdominal masses

These masses originate from the gastro-intestinal tract, the renal system, the adrenal gland, the lymphatic system, the reproductive system and the vascular system.The most common cause of distention of the fetal abdomen is ascites which is easily recognisable. The distending loops of fetal gut are highly suspect of fetal bowel obstruction. The "double bubble" sign is highly indicative of duodenal atresia while "triple bubble" is diagnostic of jejunal atresia. In the case of omphalocele it is important to identify the structures within the omphalocele because of the possibility of reconstructive surgery.

Urinary tract

There is no problem in the visualisation of the fetal kidney and urinary bladder. In high-risk patients this will relieve the mother of a great emotional burden. There are a number of reports of the successful diagnosis of renal agenesis, polycystic kidneys, and isolated renal cyst. The most frequent cases found in our material are those of obstructive uropathy which can involve the urethra or ureters. Apart from the typical ultrasonic findings of these conditions there is a very strong association of oligohydramnios.

Limb deformities

All four fetal extremities can be studied with real-time ultrasound. The ultrasound study of the fetal limbs has potential use in the identification of various types of short-limbed dwarfs and severe phocomelia. In a pregnancy at risk for one of these conditions it can be a great confort to the parents to know that the limb length is normal. Conversely, parents may not wish to continue with an affected pregnancy. However, these measurements require significant experience on the part of the examiner and should by no means be entrusted to the beginner or to a person who only occasionally uses ultrasound techniques.

Oligohydramnios and polyhydramnios

Both conditions are highly suspect for fetal anomalies. Oligohydramnios is typical for urinary tract anomalies while polyhydramnios is highly associated with bowel obstruction. A recent analysis of works from our own department and from 12 other centres of 2351 patients with polyhydramnios, showed a range of twenty to fifty-one per cent of associated congenital malformations, indicating the variability in the clinical diagnosis of polyhydramnios rather than the true incidence of the malformation. It would be reasonable to say that the association of malformation occurs in approximately one third of cases of polyhydramnios. In about half of these, the polyhydramnios itself may require treatment so an accurate diagnosis of the related abnormality is necessary to distinguish the hopeless cases from those for which some reparative procedure is possible in the neonatal period.

Conclusion

It is now generally accepted that ultrasound should be used as the initial test in the complex antenatal diagnosis of fetal malformations. Most of the gross abnormalities can be diagnosed directly and diagnostic results will be significantly improved by the combined use of cytogenetic examination.

References :

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Prof. dr. A. Kurjak
Ultrasonic Centre
Pavleka Miskina 64
41000 Zagreb, Yugoslavia