Ultrasound screening program for detection of fetal malformations. Past and current status.

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In the period 1970 to 1980 over 30.000 patients were examined. The total number of examinations was more than 10.000, which comes to an average of 3.000 with 11.000 examinations per year.

The indicence of fetal malformations detected by ultrasound screening program until 1979 was 2-3/1.000 deliveries. In the last 2 years the rate of detected fetal abnormalities and malformations has increased to 5-6/1.000 deliveries. Besides the fluctuation in incidence of fetal malformations, the introduction of the new generation of high resolution real time scanner played an important part in improvement of intrauterine diagnosis.

One can distinguish 5 groups of diagnosis obtained by ultrasound:

1. Positive findings: Prospective diagnosis obtained by ultrasound with corresponding clinical consequences.
2. Suspicious findings: Diagnosis uncertain, but important hints for clinical management obtained (early growth retardation, Oligohydramnios, Hydramnios).
3. Failures: Malformations were not seen by ultrasound.
4. False positive findings: Normal fetuses with "abnormal" ultrasound findings with clinical consequences (termination of pregnancy).
5. Undetectable" cases: At present time the ultrasound diagnosis is not possible with conventional investigations, complementary techniques are necessary.

The rate of positive and suspicious findings in the first half of pregnancy was constant between 1970 and 1979 at approximately 50%. It has to be stressed that the accuracy of ultrasound diagnosis was higher with screening in both, second and third trimester of pregnancy. The failure rate varied between 0-25% of all malformated fetuses. The rate of "undetectable" cases decreased from 50% in 1970, to 40% in 1975 and 7% in 1980. It is too early to make any conclusions whether the significant increase in 1980 was due to better screening methods and apparatus. Until now, there were no termination of pregnancy because of false positive findings (abnormal ultrasound findings with normal pregnancy outcome).

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The list of positive findings obstained by us is shown in Tab.I.:

Anencephalus	12	Arthrogryptosis multiplex cong.	2
Hydrocephalus	5	Splanchnocystic dysencephaly	1
Iniencephalus	3	Teratoma sacrococcygeal	1
Microcephalus	1	Potter-Syndrome I	3
Epignathus	2	Duodenal/Oesophageal atresia	4
Encephalocele	2	Fetoumbilical dysplasia	1
Meningocele	9	Comp. stenosis of urethra	1
Hydrops fetus	6	Hamartoma	1
Omphalocele	3	Renal cyst	1
Phocomelia	1		

In all those cases clear ultrasound diagnose led to corresponding clinical management to termination of pregnancy with exception of cases with duodenal/oesophageal atresia.

Analysing the failures one can distinguish two major problems:

1. inadequat presentation and interpretation of fetal anatomy step by step,
2. atypical or unfavourable fetal presentation associated with oligohydramnios.

The "undetectable" cases are listed on Tab.II.:

Trisomy 8 - 21	23	Diaphragmatic hernia	3
Fetal heart malformations	17	Osteogenesis imperfecta	2
(left heart hypoplasia 3)		Ichtyosis congenital	1
(translocation of great vessels 3)		Vacterl-Syndrome	1
Microcephalus	4		

The second major group of "undetectable" cases are cardiac malformations. We believe that now some cases such as left heart hypoplasia and translocation of great vessels could be detected with creening program. The failure rate was inconstant and depended on several factors:

- WHO was the screener, qualification and experience of the screener
- WHAT scanners were used
- HOW the screening was done, only by biometry or by presentation of fetal anatomy combined with biometry
- WHEN the screening was performed, early of late pregnancy or both.

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