into question the presence of a uterine malformation. Insufficient gonadothropinic stimulation in fact, as occurs in uterine malformations which have decreased miometrial capacity to expand, may lead to thereatened abortion and consequently to a periodic expulsion of decidual material from the horn devoid of embryo ([6]).

The right correlation between clinical and histological aspects of an expulsion of polypoid material in a pregnant woman with a history of threathened abortion, may lead to the discovery of uterine malformation.

The main differential diagnosis is that with ectopic pregnancy whose clinical picture in the early stages is similar. Previously described clinical examinations ([4]) in the early stages of pregnancy are useless.

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DETAILED ULTRASOUND AS A SCREENING METHOD FOR CRANIOSPINAL ABNORMALITIES

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Summary: High resolution diagnostic ultrasound was assessed as a screening method for craniospinal anomalies during the second trimester of pregnancy in a population at low risk for neural tube defects (83,403 mothers). The effectiveness of the test was about 60% and the failure rate mainly due to late attendance. In a subgroup (9325) where the screening purposes were satisfactorily fulfilled, the detection rate (87%) was substantially greater. The significance of the results and the cost/benefit ratio, especially compared with serum alpha-feto protein screening services, are then discussed.

INTRODUCTION

Craniospinal abnormalities are among the most common congenital defects. Since 95% of these cases occur in pregnancies without recognizable risk factors ([14]), the need for an effective screening method is obviously mandatory. Regular screening programmes with measurement of the alpha-fetoprotein concentration in maternal serum (MS-AFP) have been ini-

tiated in the United Kingdom and several other countries for early antenatal detection of neural tube defects NTDs), because of the relatively high incidence of this pathology (about 4-5/1000 births) ([13]). Ultrasound is currently considered a necessary ancillary technique for MS-AFP screening and, with improved training and increased confidence, also an alternative to amniocentesis for mothers with serum results.

For the past 4 years a screening programme, based on detailed ultrasonic examination of all pregnant women, has been our policy ([4]), which has been extended to 15 District Hospitals. The results of this period are the object of the present study.

MATERIAL AND METHODS

Between January 1981 and December 1984, all the patients attending the Antenatal Clinic of the II Department of Obstetrics and Gynaecology (University of Bari) underwent sonographic examination early in the second trimester (18-22 gestational weeks). Two ultrasonic apparatuses were employed (Aloka 256, linear array; CGR sonal 400, mechanical sector scanner). The number of patients was 9325, including 8930 routine scans (to asses gestational age and to rule out major structural defects) and 395 high risk mothers (table 1). The average attendance of the patients was about 97%.

The same screening purposes were followed by 15 District Hospitals in our Region (11 Antenatal Clinics with high resolution scanning facilities; number of pregnancies = 74078). The percentage of attendance was 57.5% (42595 mothers screened; an uptake ranging from 41.3% to 73.7%).

Table 1. — *Patients included in the group with high risk for a craniospinal defect.*

Previous affected baby with	
N.T.D.	60
isolated hydrocephaly	10
microcephaly	2
Family history of N.T.D.	125
Raised M.S.A.F.P.	120
Suspicious routine ultrasound performed elsewhere	78
Total	395

RESULTS

Table 2 shows number and types of craniospinal defects which were found in the population. The anomalies are divided into 2 groups on the grounds of correct or failed early ultrasonic diagnosis (before 24 weeks gestation). Only second trimester diagnoses were considered successfully screened. The prevalence was 1.58/1000 births (open NTDs = 0.98/1000), which is consistent with a low risk population ([3]).

The overall detection rate of the screening was 53% (table 3), ranging from 78.7% for Anencephaly to 52.8% for Hydrocephaly and to 40% for Spina bifida. Where the screening was correctly attended by the women (>95%), the figures are definitely higher (table 4), with sensitivity of 100% for Anencephaly, 80% for both Hydrocephaly and Spina bifida (overall detection rate = 87%).

Table 2. — *Number and types of craniospinal defects, which were found in the population.*

	Detected 24 weeks	Undetected
Anencephaly	37	10
Spina bifida	10	19
Encephalocele	4	2
Hydrocephaly	19	17
Microcephaly	/	5
Others	/	9

Table 3. — *Levels of effectiveness of the screening over the whole population.*
Sensitivity (true positive / true positive + false positive)

Anencephaly	78.7%
Hydrocephaly	52.8%
Spina bifida	40%

Table 4. — *Sensitivity and specificity of the test over the correctly screened group.*

	True positive	False positive	True negative	False negative
Anencephaly	19	0	9306	0
Spina bifida	4	1	3319	1
Hydrocephaly	4	0	9320	1

Table 5. — *Sensitivity, specificity and predictive values of the method used to detect craniospinal defects.*

	Anencephaly	Spina bifida	Hydrocephaly	Overall
Sensitivity	100%	80%	80%	87%
Specificity	100%	99.1%	100%	99.9%
Predictive value of negative test	100%	99.1%	99.9%	99.9%
Predictive value of positive test	100%	80%	100%	96.4%

After a wrong diagnosis of meningocele, one fetus was terminated, whereas the autopsy revealed a parotideal cyst (false positive rate = 0.02%, related to the latter subgroup). In table 5 the efficacy levels of the method for the correctly examined group are analyzed.

DISCUSSION

Congenital defects occur in 5-7% of all newborns. Approximately 2% are classified as major abnormalities, and 4-5% as minor ([5]). With high resolution scanning equipment and trained personnel, it is theoretically possible to recognize the majority of fetal anatomical defects during the second trimester of pregnancy. A sensitivity of 95%, with a false positive rate of 0.3%, for major defects, was obtained from an experienced, high risk referral centre ([6]). Detection rates of 80% for Spina bifida and 100% for Anencephaly, with specificity of 99%, were recorded when detailed ultrasound was offered to mothers at risk for NTDs ([10]). The same high efficacy has not been sufficiently proved when applying the screening to the entire obstetric population. Contradictory rates of detection by routine ultrasound have been reported ([1, 2, 7, 8]).

Large scale MS-AFP screening should be able to detect more than 90% of anencephalic fetuses, 70% of fetuses with Spina bifida, about half of cases with abdominal wall defects and nearly all fetuses with congenital nephrosis of the Finnish type ([12]). A percentage of 56.1% open NTDs were terminated as a direct result of serum screening in the South Wales experience ([9]) and real concern was raised whether overall efficacy levels above 65% for open Spina bifida could be achieved under normal service conditions. Similar results were obtained in this study by ultrasound alone (53% of craniospinal defects detected and consequently managed). Since 37.7% of the women failed to fulfill our criteria mainly because of late attendance, an improvement can be expected with earlier booking and wider facilities of equipment in all the antenatal clinics. A higher detection rate was indeed achieved in the subgroup of mothers, where the screening was thoroughly performed. The rate of false positive terminations (0.02%) was comparable with other Authors' experience ([1]) and similar to the one expected for an established AFP screening ([12]).

It is very questionable whether these effectiveness levels (87%) could be raised or even sustained under standard conditions ([11]). The association of serum and amniotic fluid AFP assays and ultrasonography showed a greater sensitivity than each method alone ([8]). Yet the main limit of MS-AFP results interpretation is the requirement of a precisely assessed gestational age, and maximum of efficacy of the screening is between 16 and 18 weeks of pregnancy ([14]). An ultrasonic examination would therefore be necessary before the sample, whereas it is generally agreed

to postpone the first scan beyond 18 gestational weeks, as this is the most advantageous period for detection of fetal anatomy. It has to be assessed whether a combination of the two methods would improve the current results of diagnostic ultrasound and whether its cost/benefit ratio would apply to a population at low risk for NTDs.

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