First trimester ultrasonography screening for fetal abnormalities: 34 years experience of study

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Objective

To evaluate the effectiveness of routine ultrasound examination to detect different fetal abnormalities in first trimester of pregnancy in daily clinical practice and to determine the incidence of abnormalities in this population.

Methods

85, 325 pregnant women were examined between 1980 and 2013 in first trimester of pregnancy without preliminary clinical/genetic counseling. The age of patients ranged from 14 to 47 years (mean=30. 5). Nulliparus women were 50, 740 (59. 5%), multiparous women with two previous pregnancies were 22, 732 (26. 6 %) and multiparous with more than two previous pregnancies were 11, 853 (13. 9%). For the ultrasound examination standard ultrasound machines with 3. 5 MHz transabdominal transducer were used and transvaginal transducer was added in 1994. The routine ultrasound examination was as part of the system of special integrated program for fetal and maternal status complete evaluation. After 1994 the nuchal translucency measurement was included in the study program. The statistical analysis was performed by statistical package program for social science (SPSS).

Results

Fetal abnormalities were detected in 331 cases (0. 39%). Chromosomal abnormalities in 211 (0. 25%) and 156 (0. 78%) of them were diagnosed prenatally and 55 (0. 22%) postnatally. From the cases prenatally detected, 81 (51. 7%) were determined in the second trimester because of false-negative results in the first trimester. Overall detection rate, positive predictive value and specificity were 55%, 11% and 74%, respectively.

Conclusion

Comparatively low rate of detected structural/chromosomal abnormalities in first trimester using routine ultrasound examination between 8 and 14 weeks of pregnancy was evident. The success of prenatally detection can be achievable only using good machines ultrasound and people well trained.

Table. Detection of major structural defects prenatally and postnatally

Detection rate (n (%))				
Fetal abnormality	**n**	**1st trimester**	**2nd trimester**	**Postnatally**
Central nervous system	**96**	**65 (67)**	**27 (28)**	**4 (5)**
Hydrocephaly	18	10(56)	8 (44)	0 (0)
Acrania-exencephaly	26	21 (81)	5 (19)	0 (0)
Holoprosencephaly (alobar)	1	0 (0)	0 (0)	1 (100)
Spina bifida	32	25 (78)	6 (19)	1 (3)
Encephalocele	17	9 (53)	6 (35)	2 (12)
Microcephaly	1	0 (0)	1 (100)	0 (0)
Agenesis of CC	1	0 (0)	1 (100)	0 (0)
Face and neck	**13**	**4 (31)**	**2 (38)**	**7 (31)**
Facial clefting	7	1 (14)	1 (14)	5 (72)
Cystic hygroma	2	2 (100)	0 (0)	0 (0)
Other	4	1 (25)	1 (25)	2 (50)
Thoracic	**10**	**0 (0)**	**3 (30)**	**7 (70)**
Tetralogy of Fallot	2	0 (0)	2 (100)	0 (0)
Atrioventricular canal	1	0 (0)	0 (0)	1 (100)
Left heart defects	1	0 (0)	0 (0)	1 (100)
Right heart defects	2	0 (0)	0 (0)	2 (100)
Great artery defects	1	0 (0)	0 (0)	1 (100)
Other cardiac	2	0 (0)	0 (0)	2 (100)
Lung	1	0 (0)	1 (100)	0 (0)
Gastrointestinal	**43**	**31 (72)**	**9 (21)**	**3 (7)**
Gastroschisis	15	14 (98)	1 (2)	0 (0)
Omphalocele	21	16 (76)	3 (14)	2 (10)
Diaphragmatic hernia	2	0 (0)	1 (50)	1 (50)
Large wall defects	2	1 (50)	1 (50)	0 (0)
Other	3	0 (0)	1 (100)	0(0)
Urological	**73**	**49 (67)**	**15 (21)**	**9 (12)**
Renal agenesis	4	3 (75)	1 (25)	0 (0)
Hydronephrosis	19	17 (89)	2 (11)	0 (0)
Cystic kidney	13	10 (77)	2 (15)	1 (8)
Megacystis	17	16 (94)	1 (6)	0 (0)
Other	20	3 (15)	9 (45)	8 (40)
Skeletal	**32**	**19 (65)**	**7 (22)**	**6 (13)**
Limb reduction defects	8	3 (37)	3 (37)	2 (26)
Osteochondrodysplasias	20	16 (80)	3 (15)	1 (5)
Arthrogryposis	4	0 (0)	1 (25)	3 (75)
Other	**64**	**14 (22)**	**44 (69)**	**6 (9)**
Polymalformation	33	6 (18)	25 (76)	2 (6)
Tumors	2	0 (0)	1 (50)	1 (50)
Hydrops	29	8 (28)	18 (63)	3 (9)
Total	**331**	**182 (55)**	**107 (32)**	**42 (13)**
Chromosomal abnormalities	211	55 (26)	101 (48)	55 (26)