

## Performance evaluation of the combined screening of congenital and hereditary diseases of the fetus

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### Objective

To evaluate the effectiveness of combined screening of congenital and hereditary diseases of the fetus.

### Methods

We conducted a retrospective analysis of prenatal examinations in pregnant women in the Republic from 2008 to 2012. This included 18947 patients in the at risk group who were screened at the Republican Center "Mother and Child Screening" between 16-20 weeks until 2012. The age of the women ranged between 16-45 years.

### Results

Over the period 2008-2012, an ultrasound examination was performed on 670, 860 pregnant women. 21. 8% of women were identified to have markers of chromosomal abnormalities. The detection rate of fetuses with congenital and hereditary diseases among pregnant women at risk was 1. 9%. The results of biochemical screening for variations in the serum markers were detected in 2341 women. In 269 of the cases, fetal malformations were found. Obstetric pathology such as a non-developing pregnancy, fetal death and other pathologies were identified in 202 cases. In 16. 2% of cases, isolated defects were observed, and in 39. 8% of cases there were a combination of several factors. Biochemical screening analysis showed that out of 7230 surveyed women, the high risk group made up 142 (1. 96 %), the intermediate-risk group 796 (11%), and the low risk group had 6292 (87%) of the pregnant women.

### Conclusion

The results of the analysis showed that for the period from 2008 to 2012, 23% of women were screened. The data obtained indicate the significant efficacy of the combined screening test, including ultrasound examination in conjunction with the definition of marker proteins and calculation of individual risk of having a child with malformations or chromosomal diseases.

Biomarker	Monitoring Group					
	Normal, n=11,022		Malformations, n=828		Down's syndrome, n=457	
	Mean value	Dispersion	Mean value	Dispersion	Mean value	Dispersion
AFP, MoM	1.03	0.17	2.04	0.51	0.91	0.38
Hcg, MoM	0.96	0.34	1.6	0.29	2.32	1.41

