

Prenatal diagnosis of aneuploidies in the Amur region using an electronic patient referral system

Samochvalov AV

Amur Regional Clinical Hospital, Blagovetshensk, Russia

Objective

To present the results of the introduction of the monitoring of pregnant women in combination with prenatal screening in the first trimester in the Amur region, which is a large territory of 360,000 km with the population of 217,600 women of reproductive age.

Methods

There was an established and implemented program for the electronic referral of pregnant women in all remote areas. Genetic counseling was also available. The patient demographics were recorded as well as the gestation age. Following their registration, the patient was given a date for prenatal screening. Ultrasound examination, blood collection and transportation to the screening laboratory was carried out for each patient. There were 3 ultrasound rooms, a biochemical laboratory (using Delfia Express and Astraia software), a clinical geneticist, and the invasive procedures room. All pregnant women who had their examination in the center received their results of prenatal screening in the same day.

Results

In 2013, a total of 8499 patients were seen for first trimester screening. There were 138 cases (1.6%) in the high risk group and in 118 of them underwent an invasive procedure. We found 23 (19.4%) cases of aneuploidies in this group. Down syndrome was confirmed in 16 cases, Patau syndrome in 2 cases, Edwards syndrome in 2 cases, Turner syndrome in 1 case and other chromosomal abnormalities in 2 cases. The coverage of first trimester combined screening was 89% for 2013. In comparison with 2012 combined screening was performed in 6389 pregnant women and diagnosed 15 cases of chromosomal abnormalities, 9 of them with Down syndrome. In 2011, 6874 women had the combined screening which discovered 6 cases of chromosomal abnormalities, 4 of them with Down syndrome.

Conclusion

Organization of the combined screening in the first trimester with using of electronic registration of pregnant women in this remote region allowed us to examine 89% of pregnant women. Based on the fact that the number of births within the last three years (2011-2013) was static, the detection rate of prenatal screening increased in 2013 by 35% compared to 2012 and by 74% in comparison with 2011.