

EP 11.20 Effectiveness of a chromosomal abnormality screening program using maternal age and nuchal translucency measurement in a maternal-fetal medicine unit

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Objective

The aim of this study was to report the performance of a screening strategy based solely on maternal age and nuchal translucency measurement in a maternal-fetal medicine unit.

Methods

We reviewed the first-trimester screening results (between 11 and 13+6 weeks gestation) of women evaluated at the maternal-fetal medicine department over a 12-year period. Risk calculation was performed using the Fetal Medicine Foundation (FMF) risk calculator, taking into account maternal age and nuchal translucency measurements. NT was measured according to FMF standardized guidelines. Fig 1.

Results

A total of 9,730 patients were analyzed. Among them, 278 patients were identified as having a risk greater than 1 in 100 and had complete follow-up at the hospital. The prevalence of high-risk cases was 2.86%. In this high-risk group prenatal diagnosis was offered, 66 fetuses were diagnosed with chromosomal abnormalities. Additionally, 4 cases were detected in the low-risk group. Fig 2 The screening showed a sensitivity of 94.3%, specificity of 97.8%, a positive predictive value (PPV) of 23.7%, and a negative predictive value (NPV) of 99.9%. Of the 70 fetuses with chromosomal abnormalities, 35 were diagnosed with Trisomy 21, 14 with Trisomy 18, three with Trisomy 13, and 18 with Monosomy X. Among the 35 cases of Trisomy 21, 31 were classified as high risk, yielding a sensitivity of 88.6%, with 4 cases falling into the low-risk group. The specificity remained at 97.8%.

Conclusions

The combination of maternal age and standardized nuchal translucency measurement provides high sensitivity and specificity for identifying fetuses at increased risk for chromosomal abnormalities. This screening strategy can serve as an effective option in settings where economic constraints limit access to more advanced testing, such as the combined serum screening (dual marker test) or cell-free DNA testing.

Figure 1. NT was measured according to FMF standardized guidelines.

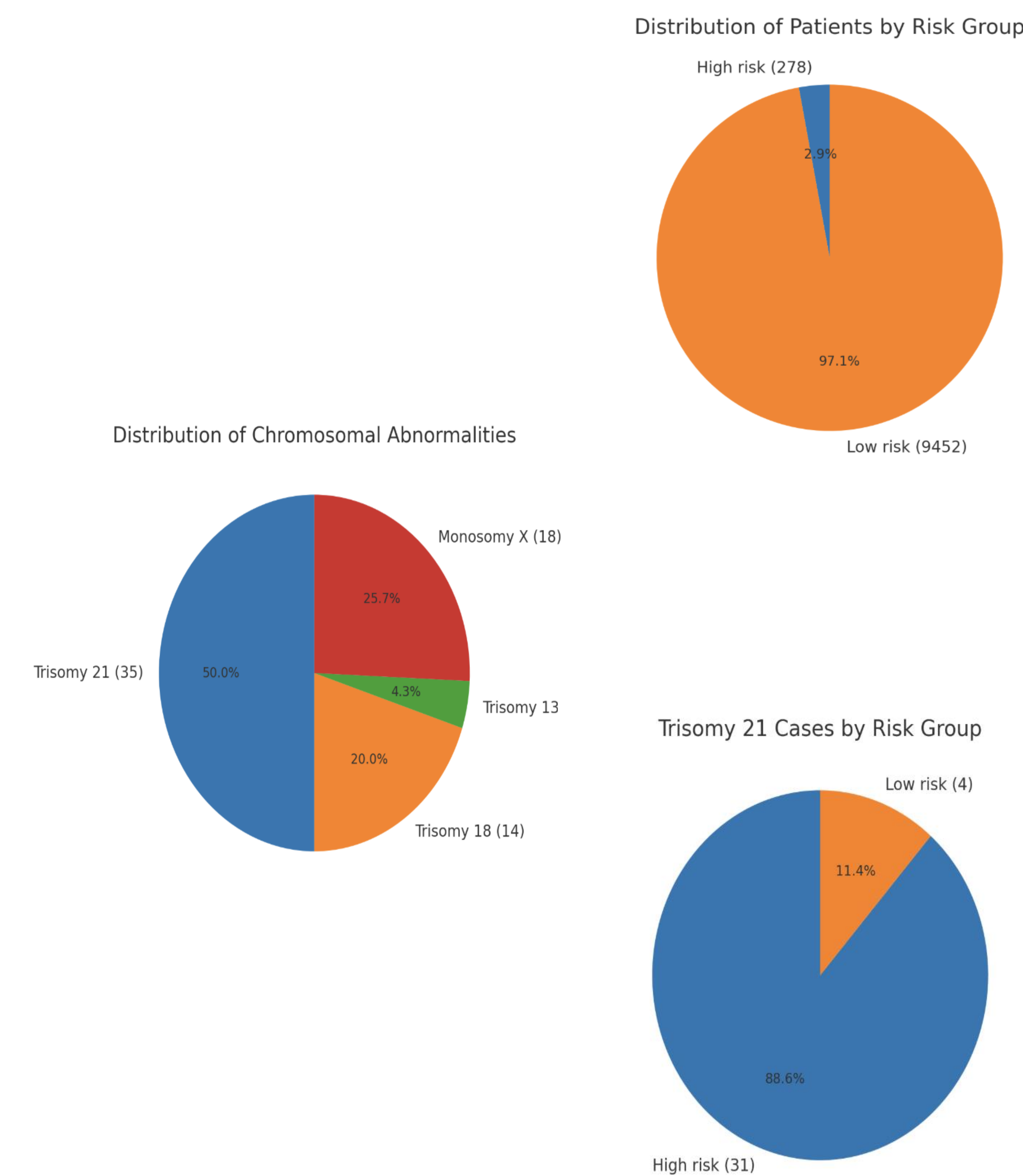


Fig. 2 Results in high and low risk screening