EFFECTIVE AND CLINICALLY APPLICABLE NON-INVASIVE ASSESSMENT OF KEL1 POSITIVE FETUSES IN KEL1 NEGATIVE “K” ALLOIMMUNIZED PREGNANT WOMEN

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OBJECTIVE

The clinical importance of assessing the fetal KEL genotype is to diagnose “K” positive fetuses (genotype KEL1/KEL2) in “K” alloimmunized pregnant women (genotype KEL2/KEL2). Just these fetuses (only 5%) are at risk of hemolytic disease and the fetal anemia should be diagnosed by Doppler ultrasound measurement of Middle Cerebral Artery Peak Systolic Velocity (MCA-PSV). Non-invasive assessment of the fetal KEL genotype is not standardly available to date. The aim of this study is to assess the fetal KEL1/KEL2 genotype from cell free fetal DNA in plasma of KEL2/KEL2 pregnant women.

MATERIALS AND METHODS

The fetal genotype was assessed by TaqMan Real-Time PCR and minisequencing (dilution series and control samples were used). A total of 138 pregnant women (between the 8th and the 23rd gestational week) were tested by minisequencing. Fetal genotype was further verified by buccal swab of the newborn.

RESULTS

TaqMan probes showed fluorescence background and the method was not able to discriminate between the background and the fetal DNA admixture. The minisequencing proved to be a reliable method. In 2.2% of the examined women (3/138) testing of plasma samples failed, 94.8% of women (128/135) were KEL1 negative and a total of 3.1% fetuses (4/128) were KEL1 positive. Sensitivity and specificity reached 100%, p <0.001.

CONCLUSION

Minisequencing is a reliable method for the assessment of the fetal KEL1 allele from plasma of KEL2/KEL2 pregnant women.