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Use of target sequencing in noninvasive prenatal testing for beta thalassemia

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Objective

To investigate and improve noninvasive prenatal testing for β-thalassemia and its clinical application Taking full advantage of this special geographical advantages of Li Nationality in Hainan Province, to conduct further exploration and improvement noninvasive prenatal testing of β-thalassemia and investigate the clinical application value that maternal peripheral blood plasma free fetal DNA on fetal beta thalassemia non-invasive prenatal diagnosis.

Methods

Two women with a heterozygous mutation of β-thalassemia who underwent a routine obstetric outpatient prenatal examination in the First Affiliated Hospital of Hainan Medical University between January 2015 and March 2017 and their partners were offered to be part in the study. The procedures were explained and written informed consent was obtained. Initially, an amniocentesis followed by a genetic examination using PCR combined with RBD technology was performed. Subsequently, blood of the couples' forearm was extracted using EDTA K2 anticoagulation blood sampling. Finally, the fetal thalassemia gene type was examined using free fetal DNA in maternal peripheral blood for applying AMP combined with second generation sequencing technology and a series of technical operations. The results were compared to the results of the amniocentesis. The results of the noninvasive prenatal testing demonstrated a good accuracy.

Results

The detection of fetal thalassemia using the noninvasive prenatal testing with target sequencing is possible and may be comparable to invasive testing.

Conclusion

The discovery of cff-DNA opened a new chapter of noninvasive prenatal diagnosis, and NIPT technology based on the two generation sequencing platform has been widely used in clinical diagnosis of Down syndrome with an accuracy of up to 99%. Therefore, new ideas for research using the NIPT should be developed.