Breaking the Limit of Non-Invasive Prenatal Testing (NIPT): From a Single Genetic Disorder to Many (Ramot)

code: 5-2018-1198

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A computational method for identifying the fetus DNA in mothers' blood sample. The method utilizes Bayesian algorithms for assigning a probability to a DNA read being of fetus origin. In addition, application of AI methods enables to account for additional features, such that separation of fetus DNA sequences are at high accuracy.

BACKGROUND

NIPT is currently used for: (i) chromosomal abnormalities (for example, trisomy 21); (i) fetal sex determination; (iii) Rhesus D genotyping; and, (iv) single-gene disorders (SGDs) of paternal origin. However, for SGDs of maternal origin, sensitivity poses a challenge that limits the testing to one genetic disorder at a time. Our developed method, termed Hoobari, increases the resolution and the sensitivity of fetus DNA identification, enabling, for the first time to predict multiple maternal single gene disorders as well as inherited insertions–deletions (indels).

STAGE OF DEVELOPMENT

The researchers have verified the Hoobari method against multiple genetic disorders of several families. The gold standard was a comparison to fetus umbilical cord blood. In addition, they have compared the Hoobari method to current state-of-the-art methods.

INTELLECTUAL PROPERTY

Ramot has filed a provisional US patent application protecting the Hoobari method in 2018, which is titled: "Method and system for identifying gene disorder in maternal blood".

REFERENCES

Rabinowitz et al. Bayesian-based non-invasive prenatal diagnosis of single-gene disorders. Genome research 2019: 29 (3), 428-438.

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