

Greater sensitivity in detecting aneuploidies and MMSs in fetuses

GTx-NIPT

Non-invasive prenatal testing (NIPT) examines small fragments of fetal DNA that circulate in a pregnant woman's bloodstream.

This advanced screening method allows the detection of abnormal chromosome numbers, as well as microdeletion or microduplication syndromes (MMSs).

The significance of NIPT lies in its ability to contribute to comprehensive pregnancy care and informed planning, offering valuable insights for ensuring the well-being of both the mother and the developing fetus.

1 Improved performance

1

Our method has better precision, accuracy and recall compared to state-of-art alternatives for aneuploidy detection.

2 Real-world preparedness

2

This model is trained with real world and **virtual** samples to increase robustness and account for low fetal fractions and sequencing coverages.

3 Clear results

3

The microdeletion segment includes clear values of precision and recall for each probability of deletion, giving a new layer of information for physicians and patients.



Microdeletion

- Monosomy 1p36
- Wolf-Hirschhorn syndrome
- Cri-du-chat syndrome
- Langer-Giedion syndrome
- Jacobsen syndrome
- DiGeorge/velocardiofacial syndrome

Aneuploidies

- Down syndrome
- Patau syndrome
- Trisomy 16
- Edwards syndrome
- Trisomy 22
- Turner syndrome
- Klinefelter syndrome
- Triple X syndrome

