How does NIPT work?

It consists of several steps

1.sample preparation:

-Maternal blood gets drawn

-The blood plasma gets isolated

-CfDNA is used to prepare sequence libraries

barcodes are attached to each cfDNA fragment, this allows numerous samples to be analyzed at the same time

-In addition each fragment is labeled with unique adapters at each end

-The barcodes distinguish the reads from each sample

2. Sequencing

-Both ends from each fragment are sequenced using short 36base-pair reads which correspond to the adapters ends

-The ends are sequenced to determine the length of the cfDNA fragment

-Each pair of sequenced reads is aligned to the genome

The length of cfDNA fragment is inferred from the distance between the two ends

3. Analysis:

-One notable metric uses a dynamic threshold for each sample to detect aneuploidy

-The reads are sorted by chromosomes to look for deviations from each the expected distribution

What is NIPT

It is an optional blood test which screens for certain genetic conditions. It started for trisomy 21 but at this time it works as well for trisomy 18, trisomy 13 and sex chromosomal abnormalities. The screen gives answers wether the sample as a high or increased chance/ risk for such condition. The screen doesn't give a 100% answer wether the baby has a disease. Many mothers do the test due to their 'higher' age in which they're pregnant or due to medical/ family history

Compare NIPT to FTS:

Both analyze for trisomy 21, trisomy 18, trisomy 13.In FTS the mothers blood gets analyzed for certain protein markers. In addition to that a NT (nuchal translucency) fest performed by which the a pocket fluid gets measured at the baby's neck.

Even though both procedures give information on higher or increased chances for chromosomal abnormalities the NIPT is more accurate because the blood sample has placental origin.

Modern diagnostic system

Poster on NIBTS by Lara Wolf, first year