



Prena LifePipe®

Prena LifePipe® is an efficient and customizable pipeline for non-invasive prenatal testing. It allows the screening of classic fetal karyotypic abnormalities and can be associated with other tools developed by Life&Soft.

Maternal plasma contains cell-free DNA from both mother and fetus. Next Generation Sequencing (NGS) of maternal plasma cell-free DNA allows for fast and safe non-invasive prenatal testing (NIPT) for the screening of fetal aneuploidies like trisomy 13, 18 or 21.

It is also possible to detect small copy number abnormalities (CNA) like deletions or duplications.

ANEUPLOIDY DETECTION

The detection of fetal aneuploidies is based on a combination of high quality alignments against the human genome and read counts to identify chromosomal gain. A first normalization is performed based on GC percentage to calculate a high-quality z-score.

A second normalization is performed in each sample to compute another z-score based on a chromosome not involved in the aneuploidy detection. The presence of a fetal aneuploidy is assessed by the value of the two z-score calculations.

SMALL CNA DETECTION

The detection of small CNA depends on a count and size-based analysis. Chromosomes are cut into bins and reads are counted. A z-score is computed to compare bin representation. A second z-score is computed to normalize the value against control samples in order to eliminate the background noise, and a deconvolution step is added to improve the detection of small CNA.

We use a statistical approach based on the read count and their size to determine the frequency of the abnormality in the sample and compare the value to the fetal fraction of the sample to determine whether the origin of the abnormality is fetal or maternal.

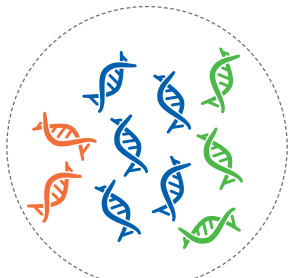
A MODULAR SOLUTION

Our bioinformatics pipeline can be easily integrated with other tools to improve the analysis experience. For example, Micro LifePipe® detects DNA viruses circulating in maternal plasma that would be important for a fetus' future.

The fetal fraction of plasma sample is of critical value for interpreting NIPT results. According to the sequencing method, the fetal fraction can be estimated by our solution. Our tool is also able to use an external fetal fraction estimation based on other experimental methods.

“ A trisomy is characterized by the presence of three instances of a particular chromosome, instead of the normal two. With NGS, it is defined by an increase of reads aligned on this particular chromosome compared to the other. ”

Reads obtained after sequencing of maternal plasma



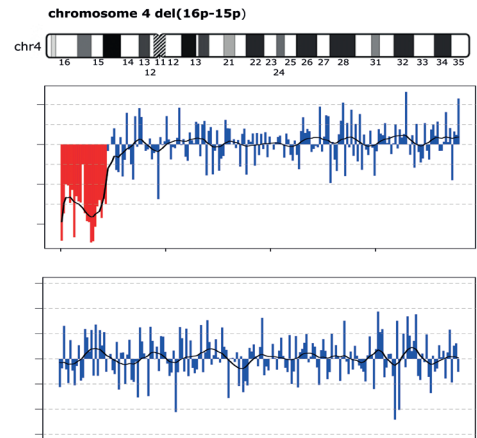
 Maternal reads  Fetal Reads


Aneuploidy detection
Copy number abnormality detection
 Example of the detection of a 4p deletion

Fetal 4p deletion



Control



 Exogenous reads Detection of a particular virus using **Micro LifePipe®**
 See our dedicated product sheet

WHY CHOOSE PRENA LIFEPIPE®?

Our bioinformatics pipeline has already been tested and validated for common fetal aneuploidies and CNA. Our tool is also designed to be used in conjunction with other Life&Soft products to improve the prenatal diagnosis.

Modular	Prena LifePipe® can be associated with different Life & Soft solutions to improve the diagnostic experience.
Built for diagnosis	You can analyze your sequencing results obtained by different sequencing technologies and platforms to detect karyotypic abnormalities and assess their fetal or maternal origins.
Quality	Our developments follow a strict quality process in accordance with ISO 62304 recommendations. The target date for CE marking is the end of 2018.
Supported	User can reach our support team anytime through our dedicated portal. Our tool is continuously improved with each new release.
Easy to deploy	Prena LifePipe® makes use of Docker® technology to allow quick and easy deployment onto your IT infrastructure.



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