NIPT

Non-Invasive Prenatal Genetic Testing

出生前検査認証制度等
運営委員会

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You have probably heard of prenatal tests called “amniocentesis” and “chorionic villus sampling” (CVS), which are both conventional methods used to detect chromosomal diseases in babies before birth. These tests are definitive, but invasive (including needle insertion into the uterus); therefore, the risk of miscarriage cannot be ruled out.

NIPT has been developed as a non-invasive screening test — i.e., without any invasion of the pregnancy — and offers a means to test for the risks of a baby having a chromosomal disorder.

This instruction contains important information for expectant mothers and their families considering whether to undergo NIPT.

Overview of NIPT

- NIPT is a test used to estimate the possibility of fetus having chromosomal anomalies called trisomies — such as trisomy 21, trisomy 18, and trisomy 13.
- The test is available from 9-10 weeks of pregnancy. It only requires 10-20 ml of blood from the mother.
- NIPT is a screening test that examines small fragments of DNA (cell-free DNA, cfDNA) circulating in the blood of pregnant women.
- Approximately 10% of the cfDNA floating in the mother’s blood is of placental origin. As it does not always match the DNA of the fetus itself, the results may be slightly less accurate. Therefore, the NIPT is considered a highly accurate test; however, it can only tell you how likely it is that your baby has a chromosomal difference.
- The results are reported as “positive,” “negative,” or “inconclusive.” “Positive” indicates a high probability of chromosomal disorders.
- If you test “positive” for NIPT, it is necessary to have a definitive test (amniocentesis or CVS) performed, while in the case of “inconclusive,” you need to consult again on the possible subsequent actions that can be taken.

What are Chromosomes?

- Most individuals have 46 chromosomes, consisting of 22 pairs of autosomes (numbered 1-22) and a pair of sex chromosomes (XX or XY).
- Each chromosome contains a large number of genes that provide a blueprint for the body’s structure and function.
- Changes in the number or structure of chromosomes can affect fetal growth and development, which can result in birth defects and physical characteristics.
- Chromosomes normally occur in pairs. Trisomy means that a person has three chromosomes instead of two. For example, trisomy 21 indicates that a baby has 3 copies of chromosome 21. (“Tri”- is Greek for “three”)

What NIPT can and cannot tell us

- NIPT is a test performed to estimate the possibility of trisomy 21, 18, and 13. No other abnormalities are detected.
  - ▸About 3-5 out of every 100 babies are born with congenital diseases.
  - ▸Of these, chromosomal abnormalities account for approximately 25% of all the diseases. Moreover, 70% of these abnormalities are trisomy 21, 18, or 13. According to the calculation, 0.7 out of 100 babies have one of those trisomies.

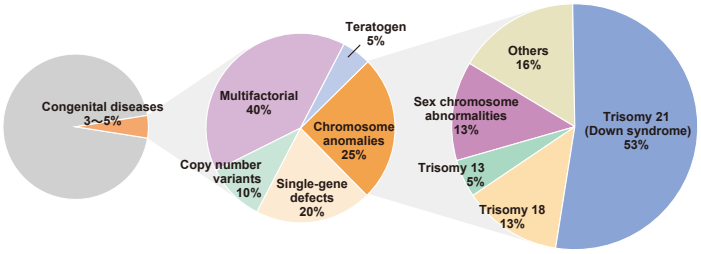

Figure. Frequency of congenital diseases and chromosome anomalies.

(Thompson & Thompson Genetics in Medicine 8th Edition Saunders 2016; Wellesley D, et al. Eur J Hum Genet 2012: 20:521)

Maternal age and trisomy

- It has been reported that the risk for trisomy increases with maternal age.
- Not every kind of chromosomal anomaly shows a clear correlation with maternal age.

Maternal age at delivery	Trisomy 21	Trisomy 18	Trisomy 13
20	1/1441	1/10000	1/14300
25	1/1383	1/8300	1/12500
30	1/959	1/7200	1/11100
35	1/338	1/3600	1/5300
40	1/84	1/740	1/1400
45	1/30		

Table. The frequency of trisomy in relation to maternal age

Congenital disorders as diversity

- Any baby can be born with a congenital disorder that is sometimes caused by chromosomal anomalies. Congenital disorders may give the baby unique characteristics which can be regarded as a part of their individuality, and what is more, human diversity as a whole.
- It would be a challenge to live with some congenital abnormalities, but this is not a critical factor in deciding whether or not you are happy.
- Whatever situation you are in, don’t forget that you are never alone. The government has attempted to build a mutually supportive and acceptable society that provides public services.
- Individual choice should be respected, including the decision to terminate or continue pregnancy. Some groups provide peer support to mothers and their families to help them make more informed decisions.

NPO for Family and Baby Wellness
https://fetalhotline.fab-support.org/

For prenatal testing, we are in collaboration with the facility (core hospital) below.

Facility Name:
Physician in charge:
Facility Address:
Contact:

Pediatricians could also be consulted before and after the test.
We collaborate with a “prenatal consult pediatrician” below.

Physician Name:
Facility Name:
Contact:

Health Labour Science Research Grant Project in FY2021 (Basic Research on Raising Next-Generation Children, including Overcoming Children’s Diseases)
“Research on the Establishment of a System for Providing Prenatal Diagnoses”