

# Cheatsheet for our Patients

## What I should ask my physician about NIPT

- › What are the common chromosome conditions screened for via NIPT?

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### **Why does this matter?**

*Aneuploidy is a presence of an abnormal number of chromosomes in the cell, for example trisomy (presence of 3 instead of 2 chromosomes) or monosomy (presence of only 1 instead of 2 chromosomes). 70 % of syndromic congenital abnormalities are contributed by Trisomy T21, T18 or T13 and 10 % by Turner syndrome (Monosomy X).*

- › Am I at risk for giving birth to a baby with a chromosomal conditions that can be diagnosed using NIPT?

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*The risk for aneuploidies is mainly dependent on the age of the mother. However there is a certain risk at every maternal age. To allow a better understanding of the individual risk, NIPT can make the difference on top of the prenatal screenigs.*

*Considering that NIPT genetic testing is noninvasive and brings no risk for pregnant mothers and their developing babies (fetuses) the NIPT testing could be recommended to all pregnant women. It is important to discuss this issue with your doctor*

- › Why is it important to consult my family before I decide to get a NIPT genetic test?

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*The test might give results showing that the fetus has a high risk beeing affected by a severe disease. The test and eventual consequences should be better discussed before between you and your partner or involved family.*

**Why does this matter?**

› What can the results of a NIPT genetic test tell me? What effects will they have?

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*NIPT test will inform you on the occurrence of the most common and relevant chromosomal aberrations of the fetus. It does not assess the complete health status and can therefore not exclude that the baby is affected by another disease. If the test turns out positive the results should be confirmed by invasive prenatal analysis. If negative these most common fetal abnormalities are most likely ruled out. In both cases the result should be discussed individually with respect to your family history and pregnancy*

› I or a family member gave birth to a baby affected with chromosomal abnormality. Does that mean my/her next baby will get the associated disease?

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*In the case of a chromosomal aberration a risk for your pregnancy might be present as well. To rule this out NIPT might be useful, however this is dependent on the individual aberration. Genetic counseling for the familiar chromosomal abnormality should be done before any tests are initiated.*

› I have a child affected with chromosomal condition. Are there any lifestyle changes that can lower my chances of having another affected baby?

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*Lifestyle changes are very unlikely to affect the recurrence risk of a genetic condition. However as other risks for the baby are dependent on the parental lifestyle you should discuss this individually with your obstetrician.*

› What are my options if I find out that my developing baby (fetus) has a chromosomal anomaly?

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*The knowledge on an existing chromosomal abnormality allows you to make informed decision how to prepare for a baby which will have certain handicaps and his very specific needs. Dependent on the timepoint of the pregnancy and legal regulations in the individual country you are residing also a termination of the pregnancy might be a possible decision. As these are very difficult decisions they should be discussed with your clinician and also with your family.*

› Will my sample and results be secure and kept private?

**Why does this matter?**

*The data as all data of genetic analyses have to be treated and archived with greatest care also with regard to data security. This can be assured for CENTOGENE NIPT data and should also be asked from other providers.*

› How much does NIPT cost? Does insurance pay for it?

*The individual price needs to be discussed with your clinician. Insurance coverage might be available but is of course dependent on your individual coverage and needs to be clarified with your insurance company.*

› **More questions**

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