NIPT good clinical practice guidelines
prepared by the BeSHG Prenatal Committee on 26.01.2017
approved by the College for Medical Genetics on 03.02.2017

Introduction

To assist women and their partners in making reproductive choices, prenatal screening for Down syndrome and other common autosomal aneuploidies is offered to pregnant women in Belgium. Non-invasive testing (NIPT) for Down syndrome (trisomy 21) and other common autosomal aneuploidies (trisomy 18 and 13) based on sequencing of cell-free DNA (cfDNA) in maternal plasma is a valuable technique for prenatal screening of high and low risk populations.1,2,3

Application of NIPT

The use of NIPT for prenatal screening in a general Belgian obstetric population results in the smallest number of missed diagnoses of fetal trisomy 21.4,5 Moreover, the number of invasive tests that are performed as a result of a positive screening test is much lower than using the combined first trimester screening (cFTS) as the primary screening instrument. Therefore, NIPT is currently the best choice as a first tier prenatal screening tool for trisomy 13, 18 and 21.

Good clinical practice with NIPT as a screening tool

- NIPT is the first tier screening tool for prenatal screening for fetal trisomy 13, 18 and 21.
- Pre-test counselling with information about the different screening options and their possibilities and limitations is required.
- Informed consent has to be obtained.
- NIPT does not replace the first trimester fetal ultrasound for measurement of the nuchal translucency (NT) and identification of fetal malformations; fetal ultrasound should be performed before NIPT screening to ascertain whether there is an indication for another prenatal test or for additional genetic counselling.
- In case of ultrasound abnormalities, including NT >95 percentile, invasive techniques (chorionic villus sampling or amniocentesis) are indicated.
- Acquiring pre-NIPT family history by means of pedigree information is standard practice to make sure that no other prenatal test is indicated.
- Referral of a patient with a positive NIPT for invasive prenatal diagnosis by amniocentesis is necessary.
- Accreditation of genetic labs offering NIPT and regular peer review on a national level (Prenatal Working group of the Belgian Society of Human Genetics) is required.
- If NIPT is used beyond the scope of trisomy 13, 18 and 21, appropriate genetic counselling is required.
• The validity and clinical utility of NIPT as a screening tool for fetal sex chromosome abnormalities is not established, therefore they are not included in the report.

• NIPT should be performed with caution:
  - in case of a multiple pregnancy or a pregnancy with a vanishing twin
  - if the patient has (had) cancer
  - if the patient recently had heparin therapy or a blood transfusion
  - if the patient has had immunotherapy, a stem cell transplant or an organ transplantation

• Incidental findings (= findings which are not directly related to the indication for which the NIPT was performed, e.g. an fetal aneuploidy of a chromosome other than 13, 18 and 21 or a genetic anomaly in the mother) should be handled according to the “Belgian guidelines for managing incidental findings detected by NIPT”.6 In case of incidental findings that are likely to be valid and have obvious clinical utility, referral for genetic counselling is required.

• The fetal fraction (= proportion of fetal cell-free DNA) is determined as a standard quality control parameter that is taken into account while interpreting all NIPT results.

References


5. PUBLICATIE VAN DE HOGE GEZONDHEIDSRAAD nr. 8912 Implementatie van niet-invasieve prenatale genetische screening van trisomie 21 (Syndroom van Down) in de Belgische zorgpraktijk 07.05.2014