**Supplement 1: Information to be included in the NIPT informed consent**

**advantages of NIPT**

* NIPT identifies common chromosomal aneuploidies: Down syndrome (trisomy 21), Edwards syndrome (trisomy 18) and Patau syndrome (trisomy 13).
* NIPT can also detect other conditions that are not targeted in current screening methods, such as fetal sex and sex chromosome abnormalities. Compared with invasive methods, NIPT allows earlier identification of these syndromes.
* Compared with other screening methods, NIPT grants increased detection rates, and decreased false positive rates (for the syndromes it tests for). Lower false positive rates preclude unnecessary invasive procedures thereby decreasing the rate of iatrogenic pregnancy loss.

**Limitations of NIPT**

* NIPT is a highly reliable screening test and is currently not considered a diagnostic test.
* The false negative rate (normal result despite a chromosomally abnormal fetus) is as high as 1.4% and the false positive rate (abnormal result despite chromosomally normal fetus) is as high as 2.1%. These rates are higher than those reported for invasive tests [3–6]. Thus, an abnormal result, should be validated by invasive testing (CVS or amniocentesis).
* In some instances, due to insufficient amount of fetal cfDNA, subject to technical/biologic factors, the test is uninformative and must be repeated. The risk of an uninformative result ranges from 0 to 13% in different reports [10].
* Prenatal detection of sex chromosome anomalies and fetal gender raise serious ethical issues pertaining to early fetal sex determination and termination of pregnancies affected by sex chromosome aneuploidies.
* NIPT is currently limited to specific aneuploides. Other cytogenetic abnormalities that are routinely identified by invasive procedures (such as additional chromosomal aneuploidies, translocations and duplications/deletions) will not be detected. At this time, NIPT is not designed to detect a wide range of microdeletions/duplications that are detected by chromosomal microarray (CMA).
* NIPT does not substitute a medically indicated invasive procedure, particularly in the presence of sonographically visualized fetal structural abnormalities. In such cases fetal karyotyping and CMA are recommended.
* NIPT does not test for single-gene mutations or syndromes attributed to such mutations. Therefore, when at risk for conditions that are not detected by NIPT (according to personal or familial history, or abnormal laboratory or sonographic parameters), the patient must be referred to genetic consultation.
* NIPT has not been validated extensively in pregnancies at low risk for chromosomal abnormalities and as such its cost-effectiveness in such patients remains unclear. With this in mind our current opinion addresses NIPT for high-risk pregnancies.
* Limited data is available on use of NIPT in multiple order pregnancies, including cases of ‘vanishing twin’. Nonetheless, in twin gestations, where first- and second-trimester screening is less accurate and where invasive procedures are associated with a relatively higher risk of pregnancy loss, NIPT may prove useful.

**Additional Comments**

It is recommended that detection rates and false positive rates for the syndromes tested, as reported in scientific publications, be declared in the result form.