

## Patient information

### Panorama test and the microdeletion screening

#### Purpose of the test

The purpose of the Panorama Non-Invasive Prenatal Test (NIPT) is to screen pregnancies to determine which ones are at high risk for the fetus to be affected by the trisomy 21, 18, 13 as well as monosomy X or triploidy. Panorama is a non-invasive test which means that it is performed on a simple maternal blood sample. This sample contains DNA from both the mother and the fetus, a property that allows to analyze the genetic changes of the child. It is available for women who are at least 10 weeks pregnant ( $\geq 9+0$ ). The test results give information only about the chromosomal state of the child. No information is transmitted about the DNA of the parents.

The microdeletion screening can be performed only additionally to the Panorama test. It screens with the same sample for the microdeletion syndromes 22q11.2/DiGeorge, Prader-Willi, Angelman, Cri-du-Chat and deletion 1p36.

#### Test results

The Panorama test like every NIPT is not a diagnostic test. It will only provide information about the risk of these conditions in your current pregnancy: high or low for each syndrome analyzed. The results are delivered usually within 7 working days to your physician with a detailed interpretation. There is a chance that the sample submitted will not return any result at the first blood drawing (in about 3% of the cases). In such a situation, it is recommended for most of the cases to repeat the test (for free).

#### Test Results Follow-up

If a high risk for a syndrome is reported and that there is a high chance that this is the case, it is not possible to conclude this definitively at this stage. In this situation, it is highly recommended to perform an invasive diagnosis (chorionic villus sampling or amniocentesis).

Your health care provider will explain you the test results and the recommended follow-up steps, which may include a referral to a genetic counselor.

#### Test limitations

Although this screening test will detect the vast majority of the analyzed syndromes, it is however not 100% of them. A 'low risk' result reduces highly the chances that your fetus has an anomaly, but a minimal risk cannot be excluded.

All other anomalies not listed in the section "purpose of the test" and mosaic trisomies cannot be detected by this test. Inaccurate test results or a failure to obtain test results may occur due to one or more of the following rare occurrences: courier/shipping delay; human error; biological factors such as a too low DNA level from the fetus in the maternal blood sample, mosaicism in the fetus, placenta or mother.

This test cannot be performed on patients who are carrying multiple babies (twins, triplets, etc.), on pregnancies that used an egg donor or surrogate, on pregnancies in which the mother had a bone marrow transplant or if the parents are closely related by blood.

The microdeletion screening cannot be performed on a pregnancy with a mother carrying herself a microdeletion.

#### Costs

Panorama test: CHF 800.-, there may be additional costs for blood withdrawal and for counseling

Microdeletion screening: CHF 230.-

The invoice will be issued to the patient after arrival of the results.