INFORMATION AND CONSENT TO NON-INVASIVE PRENATAL GENETIC TESTING

1. *Description of the purpose of sampling and genetic testing*

The purpose of the proposed testing is a non-invasive prenatal diagnostic testing of defects of fetus Trisomy 21, 18, 13, 16, 22 , sex chromosome aneuploidies and microdeletions Syndrome DiGeorge syndrom (deletion 22q11.2), Syndrome Cri-du-chat, Syndrome Prader-Willi/Angelman, 1p36 deletion syndrome. Trisomy 21, trisomy 18 and Trisomy 13, are top three of the common chromosomal abnormalities, usually due to the presence of one extra copy of chromosome number 21, 18 or 13. Affected individuals are characterized by special facial features and mental retardation, often accompanied by multiple organ abnormalities. At present, there is no curative treatment for these diseases.

2. *Description of the proposed methods and procedures*

The object of the procedure is blood sampling of 2x 10 ml. Blood sampling is performed by using a special needle after disinfecting of the injection place.

The blood samples will be packed in Streck´s Cell – Free DNA BCT and mixed by gentle inversion 8 to 10 times. After this procedure the samples will be sent to Sequenom Laboratories, 3595 John Hopkins Court, San Diego CA 92121, US. The Non-invasive Prenatal Genetic Testing will provide an estimation of the fetal risk being affected by "Chromosomal Aneuploidies", by detecting fetal chromosomal materials with the new generation of high-throughput sequencing technology, coupled with advanced bioinformatics' analysis. This method is non-invasive, with no risk of causing miscarriage and intrauterine infection, and is highly sensitive, with accuracy over 99.9%

The report will be available within 9 business days from the time when the laboratory receives the sample.

3. *Risks of the medical procedure, and of unexpected findings for the patient and genetically related people*

The sampling of blood could rarely cause bruising or exceptionally inflammation in the place of injection.

Although the latest research suggested that this test is highly accurate, with detection rate of fetal Trisomy 21, Trisomy 18 and Trisomy 13 are 99.6%, 99.9% and 91.7%. For chromosome Y 99.4%, sex aneuploidies 96.2% and microdeletions syndromes 94%.
Therefore, a positive result should still be confirmed with conventional karyotyping procedure, while a normal result cannot totally exclude the possibility of an affected fetus. This is the limitation of the current technology.

A repeat blood sampling may be required due to a low concentration of fetal DNA in about 0,9% cases.

The following situations are limited in our test: mothers with chromosomal aneuploidies, very early pregnancy, chimera, chromosome microdeletion and microduplication. If the pregnant women have received allergenic blood transfusion, transplantation or stem cell therapy, there will be a possibility of false-result because of exogenous DNA.

4. *Alternatives of the medical procedure*

There are these alternatives of the medical procedures:
Invasive tests: chorionic villi (CVS) of amniotic fluid (AMC) sampling

5. *Information on potential limitations in the usual way of life and work skills, information about treatment regime and appropriate preventive measures*

In the case of blood sampling there are no restrictions after the procedure. The testing is non-invasive and so it does not limit you in the usual way of your life in any way.

6. *Consent with the testing*

I hereby expressively grant my consent to the company IMALAB s.r.o., ID Number 63468387, residing in Zlin, U Lomu 638, Postcode 760 01, Czech republic, for the above specified sampling of my blood, for subsequent centrifugation and extraction of plasma or DNA and for sending the samples to the company Sequenom CMM, 3595 John Hopkins Court, San Diego CA 92121, US.I also grant my consent to the company Sequenom Laboratories to the non-invasive genetic tests of Chromosomal Aneuploidies.

I proclaim that I was informed by the doctor fully and in a satisfied way about reasons leading to the planned testing. The doctor explained me detail and clearly the methods of the testing, with it´ s advantages and success rate. I further proclaim that I was informed by the doctor about the consequences and potential risks of the method, about its alternatives and about other important circumstances. The doctor also informed me about possible restrictions in the way of life and working ability after performance of this method and about my right to decide about the testing freely. I had chance to ask the doctor about everything what I did not fully understand. The doctor answered all my questions clearly and in a satisfying way. I confirm that I understood all answers to my questions.

I further proclaim that I informed the doctor about all facts important to consider my health. I accept the warning that in case of untrue of this statement neither the companies IMALAB s.r.o. and Sequenom Laboratories nor the doctor are not responsible for the caused consequences. I undertake the obligation to inform the company IMALAB s.r.o. in case of any change in a written form.

I proclaim that I was informed about the price of the test in the amount of 1490 USD and I undertake to pay it properly and on time.

I agree that IMALAB s.r.o. transfers my personal data to Sequenom Laboratories, and to the use my clinical data by my physician and/or the laboratory for the purposes of test performation.

Patient	
Name:	
Date of Birth:	
IVF:	yes – no
Date:	
Signature:	

I, the undersigned MUDr. …….

hereby declare that I have instructed the pacient with the content of this consent in anappropriate way as listed above.

Date:

Doctor´s signature: