SMART STUDY PATIENT INFORMATION LEAFLET

Protocol Title: **S**NP-based **M**icrodeletion and **A**neuploidy **R**egis**T**ry (SMART)
Protocol #: 14-024-NPT
Sponsor: Natera Inc.

Principal Investigator: Prof Fergal Malone
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INTRODUCTION

You are being asked to participate in a research study because you have chosen to receive Panorama prenatal screening as part of your antenatal care. Before you decide to be a part of this study, you need to know why the research is being done, what it will involve and the risks and benefits. Please take time to read this information carefully.

DISCLOSURE OF FINANCIAL INTERESTS

Natera Inc. is providing funds to the Rotunda Hospital to support this research study. Natera Inc. is the genetic testing company, based in California USA that developed the Panorama prenatal screening test.

PURPOSE OF THE STUDY

The purpose of this study is to evaluate the performance of the Panorama screening (SNP based Non – Invasive Prenatal Testing), in a large group of pregnant women, by collecting the birth outcomes of the pregnancy and comparing the outcomes with the prenatal screening results. Panorama is used in clinical practice at the Rotunda to screen pregnancies in order to determine which ones are at higher risk for the fetus having chromosomal abnormalities. For the purpose of this study an additional microdeletion [22q11.2] is being added to the screening. This additional microdeletion is not currently offered as part of routine clinical practice in the Rotunda and the purpose of this study is to verify whether it is sufficiently sensitive to offer it as part of routine screening in the future.

NUMBER OF SUBJECTS AND LENGTH OF STUDY

This is a multi centre study. About 10,000 subjects will participate in this study at research sites around the world. This study site is expected to enroll 1200 subjects at the Rotunda Hospital. We expect recruitment to the study to take 24 months. In terms of your time input we expect this to last approximately 15 minutes.

STUDY PROCEDURES

- Natera will receive your original Panorama screening blood sample and test result as part of your routine clinical care and seeks your consent for the study team to access this data and use the sample for the purpose of this research.
- In this study, the study team will receive coded data in respect of you and your baby.
- Natera are also asking you to provide a follow up sample from your baby. In some cases a sample will be indicated as part of routine clinical care and sent to Natera for analysis and Natera is asking that it would be allowed to use this sample and result for the purpose of this research.
- Where there is no clinical indication for genetic testing following delivery of your baby we are asking you to consent to provide a DNA sample (buccal swab or leftover newborn blood spot (if available)). This sample (buccal swab or blood spot) will be sent to Natera and used to test your baby's DNA for the presence of the same genetic microdeletions for which you were screened prenatally using Panorama.

Before any follow up samples are sent to the Natera laboratory, you will be asked to read and sign the informed consent confirming that you wish to participate in this study.

Pregnancy outcomes

If you choose to have amniocentesis or other diagnostic testing, the results of that testing will be collected. Other pregnancy follow-up data and sample collection will happen in one or more of the following ways:

1. Birth record review, including delivery records and review of the newborn examination and any genetic testing results from your child that are done by your pediatrician.

2. A newborn genetic sample may be collected and sent to a Natera research laboratory. If possible, a leftover dried blood spot from the state newborn genetic screening program will be sent to the research laboratory. If not, a follow up cheek swab will be requested for research follow up testing.

3. You may be contacted by a member of the study team in the Rotunda after the birth of your baby to confirm details of your pregnancy and outcome.

FUTURE RESEARCH

- If you consent for the use of your or your child's study samples for future research, fully anonymized and de-identified samples may be kept and used for future maternal fetal health research projects, and for validation and improvement of diagnostic testing platforms. These samples will be stored at the sponsor, Natera Inc. in California
- You are not required to consent to samples being used for future research in order to participate in this study. If you do not consent to the use of your samples or your child's samples for future research, your samples will be discarded at the end of the study. If you do consent to the use of your samples or your child's samples for future research, the samples will be discarded no more than 10 years after the close of the study

RISKS AND DISCOMFORTS

There is a risk of loss of confidentiality by participating in research; however, stringent precautions are taken to keep all patient information secure.

BENEFITS

Although you will not directly benefit from this study, the research results may lead to the development of a better non-invasive test to evaluate the health of pregnancies in the future. If a genetic abnormality is identified in your baby that would otherwise not have been found, this could result in improved health for your baby.

COSTS OF PARTICIPATION

If you choose to participate, you will not be billed for the 22q deletion screening test. Follow up sample analysis will be performed at no additional costs to you for participating.

CONFIDENTIALITY

The medical information collected on you for this research study will come from information you give study staff. The information collected for this research study will be held securely at the data coordinating center-DCC (George Washington University Biostatistics Center in Rockville, Maryland) in a database consisting of information from all of the participants in this study. In the consent form you are asked to consent to the transfer of data collected on you to the USA for to be retained at the DCC and for its use for the purpose of this research. The information held at the DCC does not include your name, address, or hospital number. The database will contain your date of birth, Panorama case number and newborn blood lab screening number which could identify you. The DCC will use a unique code of numbers (study number allocated at enrollment) to identify each person. The key to the code linking the data to you will be kept in the Rotunda in a secure locked file. Only the research study staff employed for this study at this hospital will have access to the key to the code and this key will be destroyed after five years.

To the extent allowed by law, every effort will be made to keep your personal information confidential. However, information from this study will be submitted to the study sponsor (Natera Inc.) and to the appropriate regulatory authorities as required for safety and study monitoring purposes only. The results of this research project may be presented at meetings or in publications; however, you will not be identified in these presentations and/ or publications. Participation is entirely voluntary and you are free to decide not to participate in the study or to withdraw your participation and that of your baby at any time without having to give any reason.

QUESTIONS/COMPLAINTS/CONCERNS

If you have any questions or requests for information relating to this research study or your participation in it, or if you want to voice a complaint or concern about this research, you may contact Prof Fergal Malone, Rotunda Hospital, Parnell Sq. Dublin 1.