Sinai Health | Mount Sinai Hospital
Joseph & Wolf Lebovic Health Complex

PATIENT INFORMATION (PLACE LABEL HERE or TYPE)
Last Name: _____
First Name: _____
MRN: _____
Date of Birth: _____ Prov: _____
Ontario Health Card & Version Code _____
Sex (OHIP): Male ☐ Female ☐
Sex assigned at birth: Male ☐ Female ☐
Visit #/Patient location: _____

Consent for Low Pass Genome Sequencing (LP-GS) Analysis of Fetal Tissue

Your physician has offered you a fetal diagnostic test (LP-GS) for chromosomal copy number variant analysis to identify potential fetal chromosomal abnormalities as part of your prenatal or perinatal care.

Low pass genome sequencing (LP-GS) is a test used to look for extra or missing pieces of chromosomes (DNA) called copy number variant (CNV). Having extra or missing pieces of chromosomes could cause birth differences, developmental delay, or other health problems.

How is LG-PS performed? ? This genetic test is performed by the Mount Sinai Cytogenomic Diagnostic Laboratory using DNA extracted from a fetal sample. The laboratory looks at any changes in the chromosomes and uses medical information, family history, and current knowledge to understand whether these changes might cause health problems.

Sinai Health uses a third-party specialized online software service to assist with accurately interpreting the fetal LP-GS data/ sequencing results, which will require sharing your genetic and clinical data with the software service. While Sinai Health cannot guarantee unauthorized access to the data or use or disclosure of your data, our contract with the software service provider ensures that your data will be processed at a secured data center that is physically located in Canada and treated with industry appropriate data privacy and security safeguards. Please note that the software service uses artificial intelligence (AI) technology that may automatically utilize the data to increase the performance capacity of the software.

What might I learn from this diagnostic testing? The laboratory will report the following information:

- The finding of missing or extra pieces of chromosomes that are expected to cause a well-described condition related to the reason for testing (i.e. chromosome deletions or duplications that are termed “pathogenic” or “likely pathogenic” variants).
- Chromosomal changes (missing or extra pieces of DNA) that may be linked to learning difficulties, serious health problems in childhood, or conditions that could affect care during pregnancy, at birth, or in early childhood — even if they are not related to the original reason for testing.
- Chromosomal “variants of uncertain significance” that are related to the reason for referral, but the impact of the finding is not well understood at this time.

It is important to note that the interpretation of LP-GS results is based on current knowledge and this may change over time due to new scientific knowledge.

What are the risks and limitations of this test? When chromosomal changes are identified in a fetus, follow-up testing in family members, typically the parents, is often required. This may reveal that one of them also carries the same chromosomal variant.
(LP-GS) may reveal that the biological relationships within a family differ from what was reported to the healthcare provider, this includes non-paternity and consanguinity (i.e. the parents are related by blood). Incorrect information about biological relationships and health status may prevent accurate interpretation of LP-GS results, and it may be necessary to report these findings to the healthcare provider who ordered your test. In addition, LP-GS does not detect all types of chromosome variants, and does not analyze individual genes. If LP-GS does not identify a variant that may explain the reason for referral, it does not rule out the possibility that a chromosome variant may still be responsible. Finally, as with all laboratory tests, there is a small possibility of error or sample failure.

What happens to my sample after testing? After LP-GS is completed, the sample(s) from the LP-GS analysis will be stored at the Mount Sinai Diagnostic Medical Genetics Laboratory. The remaining sample may be used for additional clinical genetic testing [as indicated/requested by your healthcare provider(s)] only with your express consent). It may also be used for test development/validation and quality assurance procedures in the laboratory after it has been de-identified (personal identification information is removed).

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How will I learn my results? LP-GS results will only be reported to the healthcare provider(s) who ordered the test. Your genetic counsellor, nurse, or physician will inform you of your test results, and the written report will become part of your medical record at Mount Sinai Hospital. It is recommended that you receive genetic counselling before proceeding with testing and after testing once final results are available.

BY SIGNING BELOW, I CONFIRM THAT: (1) I have read (or had read to me) and I understand the information provided in this consent form; (2) I understand that the LP-GS test is voluntary, and I may choose not to have my sample tested; (3) I have received a copy of this consent form; (4) All my questions have been satisfactorily answered; and (5) I hereby consent to proceeding with the LP-GS test.

Signature of Patient/Substitute Decision Maker (SDM)
Print name of Patient/Substitute Decision Make (SDM)
Date (MM/DD/YYYY)

HEALTHCARE PROVIDER STATEMENT: By signing below, I attest that: (1) I am the genetic counsellor or authorized healthcare provider; (2) I have explained the purpose of test described above; (3) The patient has had the opportunity to ask questions regarding this test and to seek genetic counseling; and (4) The patient has voluntarily decided to have this test performed by Mount Sinai Cytogenomic Diagnostic Laboratory.

Signature of Health Care Provider
Print name of Health Care Provider (For external provider, print CPSO number and facility name)
Date (MM/DD/YYYY)