NAME:
DOB:
UMRN:

MULTI-GENE SEQUENCING: INFORMATION FOR PATIENTS

Why are you being offered this test?

Your doctor would like to assess for a genetic cause of your/your child's condition. Finding a genetic cause may help health professionals treat or manage the condition. It could also help you and your family better understand the condition and plan for the future. Genetic testing may inform families about the chance of having another child with the same condition.

What is multi-gene sequencing?

Our genes carry important information to make our bodies grow and function properly. This information is stored in the DNA. Previously, scientists and doctors were only able to test one gene at a time. In recent years, advances in technology have allowed us to look at many genes at the same time.

Everyone's DNA contains lots of genetic differences (variants). Most of these variants don't change how a gene works and are considered harmless (benign variants). The purpose of sequencing is to look for variants that do change how a gene works and are the cause of a genetic condition (pathogenic variants).

What are the possible results?

No abnormality found	Pathogenic variant(s)	Unclear result	Unexpected result
The test has not found a cause of your/your child's condition. **Please note:** This does not exclude the possibility of a genetic cause for the condition.*	A cause of your/your child's condition has been found.	A change is identified but its consequence is unknown. Testing of family members may clarify the result. Future knowledge may also provide better understanding of such result. Genetic counselling is advised if such findings are reported.	This result may not be relevant to your/your child's condition but may cause health problems in the future and may be relevant to other family members. **Please note:** The chance of this type of finding is very small as only genes related to your/your child's condition are tested. Genetic counselling will be helpful in this scenario.

*Not all variants can be detected by sequencing, for example complex structural variation and repetitive sequences. In addition, variants of uncertain clinical significance may be detected but may not be reported.

What happens to the samples and the results?

DNA samples are stored long-term. They may be used in the future if additional testing is required. De-identified samples may be used for ethically-approved research, quality improvement, or education. You can advise at any point if you do not want your/your child's sample to be used in these scenarios or if you would like the sample to be destroyed.

Results are reported to the doctor ordering the test and any approved recipient(s). They are also stored according to government regulations. De-identified results may be submitted to secure international clinical databases helping in continuous improvement of result interpretation.

CONSENT

- I understand this test may detect a genetic abnormality that could provide important medical information for me/my child or indirectly, for my family.
- I understand that a normal result does not rule out a genetic abnormality being present but not detected/reported.
- I understand that testing of other family members may be requested to help clarify findings.
- I can advise at any point to stop testing if I have concerns about the information which can be revealed.
- I can advise at any point to destroy my/my child's sample(s), or to refuse to allow my/my child's de-identified sample(s) being used for ethically approved research, quality improvement, or education.
- I have been provided a copy of this information sheet.

Please contact your doctor if you wish to advise on any of the points above.

Full Name (patient/guardian) _____

Signature _____ Date _____

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