Informed Consent for Next Generation Sequencing (NGS-Clinical Exome Sequencing, Whole Exome Sequencing and Targeted Panels) at the Cyprus Institute of Neurology and Genetics (Department of Biochemical Genetics)

I _____, request and authorise the Cyprus Institute of Neurology and Genetics (CING) to analyse the gene(s) stated on the test request form in: ◯ My sample ◯ My child's sample.

No testing apart from that which is ordered will be performed. Further testing requires my additional consent

I authorize CING to use my (or my child's) DNA sample and data for test validation or research purposes after the personal details are removed making the procedure anonymous ◯

I understand that:

- I have the right to refuse the above without having any consequence to the analysis. Samples and data will be stored for future use.
- I can withdraw my consent at any time.

For detailed information on data privacy policies at CING please refer to: https://www.cing.ac.cy/en/privacypolicies/cing-policy

I understand that:

1. The results of this DNA test could be:

a. Pathogenic/Likely pathogenic, and may possibly:

i. Relate to the diagnosis of the genetic condition in question.
ii. Reveal carrier status for the genetic condition in question.
iii. Be incidental findings such as a predisposition or an increased risk for developing a genetic disease in the future (please see point 9)
iv. Have consequences for other family members.

b. Benign/Likely benign, and may possibly:

i. Reduce but not eliminate the possibility that my condition has a genetic basis.
ii. Be uninformative.
iii. Not eliminate the need for additional testing.

c. Uncertain significance and may possibly:

   i. Lead to a suggestion that testing additional family members may be helpful.

   ii. Remain uncertain for the foreseeable future.

2. Current policy is for clinical interpretation and further investigation to be undertaken ONLY in those genes requested or related to the reason for referral**.**

3. Variants of uncertain significance (VOUS) are reported only when strongly related to phenotype.

4. There may be possible causes of error including, sample contamination, rare technical errors in the laboratory, rare DNA variants that compromise data analysis, inconsistent scientific classification systems, and inaccurate reporting of family relationships or clinical information.

5. The interpretation of the test results is based on the current scientific knowledge at the time of data analysis.

6. Due to the complexity of the test and the important implications of the results, the clinical reports are initially released only to the referring healthcare professional(s) listed on the test request form. It has been explained to me that my clinical report will be available to me after it has been released to my referring physician.

7. I can be offered genetic counselling with a geneticist, genetic counsellor or other qualified healthcare provider who can answer questions, provide information and advice about alternatives before and after having this test.

8. The data obtained in the analysis will be recorded, evaluated and stored in an anonymized form in scientific databases.

9. Incidental findings, which are variants not related to the reason for referral/genetic condition in question, and may be identified unintentionally within the process of analysis, are not reported.

BY SIGNING BELOW, I CONFIRM THE FOLLOWING:

1. I have been informed of the likelihood of finding a change in the gene(s) for which I am (or my child is) being tested and have received test-specific information.

2. I have read and understood the information provided on this form and have had an opportunity to have any questions answered by my healthcare provider.

Patient name: ……

Patient Signature: …….

Signature of parent/guardian, if patient is a minor: ……..

Parent/guardian's name: ……..

Date: ……

Statement by Healthcare Professional / Referring Physician:

By signing below, I confirm that I am the referring physician. I have clarified the purpose of the test described above. The patient has willingly decided to have this test performed by CING.

Healthcare provider name and signature: …….

Date: ……