Genomic Diagnostics
LEADING THE WAY TO IMPROVE HEALTH

Guardian Informed Consent

for SCN1A gene testing

1. Informed Consent for Genetic Testing: Genetic Epilepsy *SCN1A* gene

I _____ (Print Name), as parent/guardian of _____ (Print Patient's Name), hereby agree to have them participate in DNA testing for inherited and *de novo SCN1A* gene mutations. I understand that a sample will be removed from them using standard techniques that carry very little risk.

I understand that:

1. The mutation detection system used by Genomic Diagnostics (GD) for testing the *SCN1A* gene is among the most sensitive and specific yet developed. It is understood that *SCN1A* is now the most clinically relevant epilepsy gene and identification of mutations within this gene may assist clinicians in accurate diagnosis, appropriate treatment regimes and overall better patient management.

2. In other cases, the DNA test is unable to identify an abnormality, even though an abnormality may still exist in *SCN1A*. This event may be due to the current lack of knowledge in the scientific community of the complete gene structures, or a rare inability of the current technology to identify certain types of changes (mutations) in these genes. In addition, GD may not detect a mutation because the causative mutation may occur in an alternative gene.

3. Because of the complexity of genetic testing and the important implications of the test results, results will only be reported to me through a medical specialist, GP, or certified genetics professional. The result reports are kept strictly confidential. Participation in genetic testing is completely voluntary.

4. The test result may have implications for other members of my family. I will decide whether to advise them of this. I understand that the test could potentially affect my ability, and the ability of my family members, to obtain some types of insurance.

5. Genetic tests are being improved and expanded continuously. GD will store my sample and at my future request and with my consent, may in some cases, re-analyse the DNA by a new procedure(s). This additional testing may incur a further cost. My test results will be de-identified and may be used for statistical purposes. An anonymised sample of my DNA may be used in Institutional Ethics Committee approved research programmes to improve the diagnosis and management of epilepsy. If you don't want your sample to be kept for these purposes please tick here ☐

(Note: Genomic Diagnostics does not guarantee the integrity of the sample for future testing).

Signature of Guardian: _____ Guardian's Name: _____

Witnessed by: _____ Print Name: _____
(Signature)

Date: __ __ /__ __ / __ __ __ __

2. Physician's/Counsellor's Statement

I have determined that the above patient is a suitable candidate for genetic testing of the *SCNA1* gene. I have discussed genetic testing with their parents/guardian, presented the information outlined above, discussed the options available to them and have answered their questions. Results from these tests may be used to direct medical management.

Physician's Signature: _____ Print Name: _____

Date: __ __ /__ __ / __ __ __ __