the children's hospital at Westmead

think. kids.

**Department of Molecular Genetics**

# Consent Form for Massively Parallel Sequencing Testing

| Genetic File: | MRN: |
|---|---|

**Parent or Guardian**
**(Patient under age or unable to consent)**

| Surname: | Given names: |
|---|---|
| Address: | |
| | Postcode: |
| Date of birth: | Telephone: |

**Patient**

| Surname: | Given names: |
|---|---|
| Address: | |
| | Postcode: |
| Date of birth: | Telephone: |

## Part A: Provision of Information to Patient

**To be completed by Health Professional**

**Testing requested:**

☐ **Panel of genes known to cause** _____

☐ **Whole exome/whole genome testing**

**Possible outcomes of genetic testing:**

1. An **informative** result means that a variant has been found and that it explains the clinical findings.
2. If a variant is not found, the result is **uninformative**. This may be because a variant is present but could not be found using current technology. An uninformative result **does not** exclude the diagnosis.
3. Results of **uncertain significance**: sometimes a variation in a gene is found but its meaning is unclear. In this situation, further testing of other family members may be required. The interpretation of a result may also alter as knowledge of genetics improves. I may be contacted if this occurs but the time frame for any additional results is variable.
4. There is a small chance that co-incidental findings about my/my child's health not related to the diagnosis in me/my child may be identified when genomic testing is carried out. I will be informed of these co-incidental findings only if an expert committee, in consultation with my doctor or genetic counsellor, assess that these findings could have a significant impact on my/my child's/my relatives' health care.
5. Testing may reveal non-paternity or non-maternity of a presumed natural parent
6. Testing may possibly affect my/my child's ability to obtain some types of insurance.
7. Relevant clinical testing results will be given in person.
8. The information from genetic testing will be stored by the laboratory according to government regulations.
9. Sample collected (blood/muscle/skin/_____) may be stored for an indefinite time and can be retested if future testing may be more informative.

I, _____ have informed this patient/parent/guardian
Insert name of Health Professional and designation

as detailed above, of the nature, limitations, likely results and risks associated with the testing of genes. We

have discussed the procedures and consequences of testing and storage of the patient's sample.

| _____ | _____ | _____ |
|---|---|---|
| Signature of Health Professional | Signature of Interpreter (if present) | Date |

## Part B: Patient Consent

**To be completed by Patient/Guardian**

1. I consent to testing a panel of genes known to cause_____ and/or whole exome/genome sequencing as requested by my healthcare provider.

   ☐ **YES, I consent to genetic testing.** ☐ **NO, I do not consent to genetic testing.**

2. I agree that the result may also be used, if necessary, to help other family members, for their counseling and diagnosis, without disclosing specific details about the person tested.

   ☐ **YES, I consent to results being made known if reasonably indicated to other family members**

   ☐ **NO, I request that results only be made known to the following people** _____

   In the event of my death, the test results should be released to:

   Name: _____ Relationship: _____

   Contact details:_____

3. I agree that my de-identified genetic data may be submitted to genome-wide depositories as part of a global effort to better understand the role of gene variations in disease and health.

   ☐ **YES, my de-identified may be submitted to genome-wide depositories.**

   ☐ **NO, my de-identified may not be submitted to genome-wide depositories.**

4. After testing is completed, there may be research studies that I/my child may be eligible for and may be of interest. The laboratory may share my contact information with researchers who have a research study with appropriate ethics approval and for which I/my child may be eligible for participation.

   ☐ **YES, the laboratory may share my contact information if deemed appropriate.**

   ☐ **NO, the laboratory may not share my contact information for any research projects.**

### I request and consent to the test described above.

I have read the consent information and understand the potential benefits, limitations and consequences involved in the testing. I have had the opportunity to ask additional questions and I am satisfied with the explanations and the answers provided. I understand that genetic counselling will be available for myself and my family.

| _____ | _____ | _____ |
|---|---|---|
| Signature of Patient/Guardian | Print name | Date |