

Explanation Sections:

1. *What should I include in the explanation sections?*

Use these sections to address the rationale of your choices. Specify what will change medically for the patient being tested and for at-risk relatives. Indicate why a genetic laboratory diagnosis is required versus a clinical diagnosis or a diagnosis based on other laboratory testing. Explain what the genetic counselling issues are in this family and what events are depending on the test results (ie. pregnancy planning, career planning). **Please note that the test request form is not considered complete if the explanations are left blank.**

2. *Should I outline previous targeted diagnostic tests?*

Genetic testing is not the gold standard for diagnosis in all cases. There may be more sensitive tests for diagnosis which are not genetically based and other forms of testing may be available locally. The clinician reviewers find relevant information regarding previous testing of value in their assessment of all test requests.

Therapeutic Impact for the Patient

1. *To what does this refer?*

This refers to the impact on the **patient** being tested. This applies regardless of whether the patient is affected or pre-symptomatic and regardless of the inheritance pattern (ie. dominant, recessive, x-linked or mitochondrial).

2. *What constitutes “new management”?*

Examples include: an implantable device, medications that would not otherwise be used, imaging that would not otherwise be considered.

3. *What constitutes “adjust to more specific management”?*

Examples include: the significant refining of medications or imaging protocols.

4. *Why is “cease or reduce investigation for diagnosis” considered low impact?*

This rationale is non-specific and can be a reason for any test. You can elaborate on this in the explanation section if there are specific issues you wish to have considered.

Therapeutic Impact for At Risk Relatives

1. *To what does this refer?*

This refers to any **at-risk relatives** of the individual being tested.

2. *What constitutes “preventive management”?*

Examples include: an implantable device, medication or surgery that would not otherwise be used.

3. *What constitutes “specific screening recommendations or risk reduction strategies”?*

Examples include: imaging protocols and non-specific medical management (lifestyle changes).

Genetic Counselling Impact

1. *To what and whom does this refer?*

This refers to any genetic counselling issue related to the **patient** and **any at-risk relatives**. This may include pre-conception/ prenatal issues or lifestyle planning.