

Example 1

A woman has a history of epistaxis and has been found to have an arterio-venous malformation in her lung. She has four children and she reports that they have all had epistaxis off and on, but two seem to be worse. Her eldest daughter is considering starting a family soon. Does the mother have Hereditary Hemorrhagic Telangiectasia?

Therapeutic Impact for Patient	Low Impact	It is likely that the management for the mother will be the same independent of genetic testing.
Therapeutic Impact for At Risk Relatives	Moderate Impact	There are four individuals at risk. If a pathogenic mutation is identified, and predictive testing is done, then specific screening recommendations and risk reduction strategies can be applied to those with the mutation. Those who do not have the mutation do not require special management.
Genetic Counselling Impact	Moderate Impact	Unless a child or their partner was pregnant and interested in prenatal testing in which case it would be high impact.

Example 2

A woman has a mutation in the ALK-1 gene. You now put in a request for testing of her eldest daughter who has an occasional nosebleed.

Therapeutic Impact for Patient	Moderate Impact	Management would be adjusted based on the result.
Therapeutic Impact for At Risk Relatives	No Impact	The daughter's results do not impact further on sibling risk since her mother's results are known. However, if the daughter has children, then there is moderate impact.
Genetic Counselling Impact	Moderate Impact	Pre-conception counselling is available. If the daughter were pregnant and interested in prenatal diagnosis, it would be high impact.

Example 3

A couple has a child who has a serious autosomal recessive disorder. You know the diagnosis and there is no specific treatment. However, the parents wish to consider another pregnancy. You request funding to perform genetic testing on the child.

Therapeutic Impact for Patient	No Impact	There is no change in management for the child.
Therapeutic Impact for At Risk Relatives	No Impact	There is currently no other individual at risk in the family. If there is a sibling at risk, then it would be low impact as there is no available treatment.
Genetic Counselling Impact	High Impact	If the couple was interested in prenatal diagnosis.

Example 4

A woman has a sister who carries a mutation for an X-linked disorder which has beneficial pre-symptomatic treatment. Female carriers of this condition have no or minimal symptoms and no specific management is indicated. The woman has two male children and is currently 6 weeks pregnant. She is not interested in prenatal diagnosis.

Therapeutic Impact for Patient	No Impact	There are no management issues for this woman.
Therapeutic Impact for At Risk Relatives	High Impact	Testing and pre-symptomatic management is potentially available for both her sons.
Genetic Counselling Impact	No Impact	No genetic counselling concerns.

Example 5

A man is suspected of having a dominant condition for which there is no treatment. There is a family history of the same condition in his deceased father. He is not planning to have a family but he is concerned that having this condition may preclude certain career choices.

Therapeutic Impact for Patient	No Impact	There is no management for this condition.
Therapeutic Impact for At Risk Relatives	No Impact	If he has no siblings. If he has siblings, it would be low impact.
Genetic Counselling Impact	Low Impact	Lifestyle planning (it is important to fully explain this point in the narrative section of the request).

Example 6

A child has a diagnosed metabolic disorder. Defining the specific genetic abnormality might mean increasing doses of medications that the child already takes. There are no other individuals in the family at risk and the parents have completed their family.

Therapeutic Impact for Patient	Moderate to No Impact	The impact depends in whether having a confirmed genetic diagnosis defines treatment.
Therapeutic Impact for At Risk Relatives	No Impact	There are no at-risk relatives.
Genetic Counselling Impact	No Impact	No genetic counselling concerns have been identified.