



POTENTIAL CONSEQUENCES OF GENETIC TESTING

Page 1 of 2

- The tables on page 2 bring together the types of consequences which might follow the different possible genetic test outcomes for an individual patient.
- Different elements will be more or less prominent for different people, and different disorders.
- You can use this in any way you feel appropriate to help you assess whether a genetic test is likely to be beneficial for your patient.

Types of Results:

Mutation Present	→	1. Disease Causing Mutation Present Presence of a gene mutation that is predicted to cause or increase risk for disease.
	→	2. Variant of Unknown Significance A variation in the relevant gene is identified, but it is not yet established whether it is disease-associated or a harmless variant (polymorphism).
Mutation Absent	→	3. Clear Negative Test Result Absence of a known gene mutation previously identified in the family.
	→	4. Uninformative Test Result No variation in the relevant gene(s) has been identified, but the family history suggests a genetic basis to the disease. This could mean one of the following: <ul style="list-style-type: none"> • A mutation exists that current technology can not yet identify • Another, as yet undiscovered, gene exists which causes the disease in this individual • A mutation exists in the family (not yet identified), but this individual did not inherit it • Despite the family history, this occurrence of the disease is a random event

POTENTIAL CONSEQUENCES OF GENETIC TESTING

BENEFITS

Test Result (as described on page 1) → → →	MUTATION PRESENT		MUTATION ABSENT	
	1. Disease Causing Mutation Present	2. Variant of Unknown Significance	3. Clear Negative Test Result	4. Uninformative Test Result
Clinical	Clear basis for existing clinical interventions that improve outcome		Avoidance of unnecessary interventions	
Health Behaviour	Importance of positive health behaviour can be reinforced	Importance of positive health behaviour can be reinforced	Importance of positive health behaviour can be reinforced	Importance of positive health behaviour can be reinforced
Family	Others at risk can be identified		Children can be reassured about their risk status	
Emotional	Relief of uncertainty, permission to take charge of affairs		Relief from worry about disease risk	
Socio-legal			Higher insurance premiums should be avoided	Higher insurance premiums might be avoided

HARMS

Test Result (as described on page 1) → → →	MUTATION PRESENT		MUTATION ABSENT	
	1. Disease Causing Mutation Present	2. Variant of Unknown Significance	3. Clear Negative Test Result	4. Uninformative Test Result
Clinical	<ul style="list-style-type: none"> Clinical interventions may carry risks or may not improve outcome Disease may have incomplete penetrance - patient may never get disease 	Continue clinical interventions which may carry risks or may not improve outcome		Continue clinical interventions which may carry risks or may not improve outcome
Health Behaviour	Fatalistic attitude to health	Fatalistic attitude to health	Complacent attitude to health	Complacent or fatalistic attitude to health
Family	Other family members distressed on patient's or their own behalf	Confusion – some may perceive it as a negative result, some as a positive result	Dysfunctional dynamics arising from challenging family beliefs about disease	Uncertainty leads to interpersonal problems between family members
Emotional	Anxiety, depression	Anxiety, depression, uncertainty, i.e. "no-man's land"	Survivor guilt; cannot identify with disease group; no excuse for life problems	Uncertainty, confusion
Socio-legal	Insurance premiums may be increased	Will insurance premiums be increased?		Insurance premiums may not be reduced