# The different types of genomic test results

## Introduction

Patient undergoes diagnostic germline testing, with four possible outcomes:

1. Confirmed genetic diagnosis
2. Incidental findings
3. No significant findings
4. Variant(s) of uncertain significance

## What does this mean?

1. **Confirmed genetic diagnosis:** A pathogenic or likely pathogenic variant(s) consistent with the patient’s condition is identified. This should be reviewed by the clinical team who can correlate this result to the patient’s existing condition.
2. **Incidental findings:** A result was found, unrelated to the reason for requesting the genomic test. Examples: a deletion that encompasses a cancer predisposition gene when seeking a cause of a child’s developmental delay’ or misattributed parentage.
3. **No significant findings:** A genetic diagnosis is not confirmed. Based on current knowledge, no significant variants associated with the patient’s existing condition have been found.
4. **Variant(s) of uncertain significance:** A genetic diagnosis is not confirmed. There is currently insufficient information to confirm whether this is part of natural genomic variation or is relevant to the patient’s condition.

## Implications for the patient and family

1. **Confirmed genetic diagnosis;** and
2. **Incidental findings**
   * The genomic result may or may not affect the patient’s current or future management, provide insight about prognosis or susceptibility to other health conditions. Onward referrals may need to be made.
   * Cascade testing (such as predictive or carrier testing), preconception or prenatal testing may be offered to the patient and certain family members.
   * Further support and genetic counselling may be offered to adapt to the diagnosis and/or to share information with family members.
   * Incidental findings such as those about parentage often raise ethical issues about how to disclose these results to the patient and family.
3. **No significant** **findings**
   * This does *not* necessarily mean that the patient does not have an underlying genetic basis for their condition.
   * This can be a relief for some patients concerned about a possible outcome, or devastating if they have been seeking a diagnosis.
   * Clinical management would be based on the patient’s clinical and/or family history rather than genetic result.
   * It may be that a conclusion cannot be reached at this time. The patient’s data may be reviewed in the future (e.g. based on changes to the patient’s clinical or family history, or in light of new information).
4. **Variant(s) of uncertain significance**
   * Information may be outlined in the lab report if further actions can be taken (such as further samples or tests, testing family members).
   * Predictive, preconception or prenatal testing cannot be offered to the patient or family members for this variant.
   * Clinical management would be based on the patient’s clinical and/or family history rather than genetic result.
   * It may be that a conclusion cannot be reached at this time. The patient’s data may be reviewed in the future (for example, based on changes to the patient’s clinical or family history, or in light of new information).

## Key points to consider

* Ensure you have read the laboratory report carefully, including any recommended actions.
* Consider how this result will be communicated (such as letter, phone, face to face, location, who should be present), and provide an opportunity for further contact.
* Discuss with your patient if there are any subsequent genomic or other medical tests that may be considered, or any onward referrals for their management.
* Discuss the importance of sharing information with family members, and which relatives may be offered testing depending on the inheritance pattern.
* Review and provide details/links to support groups and information that may be available.
* Offer to provide the patient with a copy of their report and discuss sharing this with other healthcare professionals involved in their care.
* Discuss any relevant research initiatives that may be offered based on the test or result.
* If you have any queries, consider discussing with your Genomic Laboratory Hub, Clinical Genetics Service, clinical colleagues and/or at a multidisciplinary team meeting.