**What does ‘no primary findings’ mean?**

**Genomics 101 explainer episode transcript**

**Florence: What does ‘no primary findings’ mean? I'm joined by Adrianto Wirawan, Director of Bioinformatics Engineering for Genomics England, to find out more. So firstly, Adrianto, when we speak about findings from genomic tests, what does this mean? What are we looking for when we do a genomic test?**

**Adrianto:** Our DNA is made up of a long sequence of letters that act like instructions for your body.

Genomic testing analyses these letters to see if there are any unusual patterns or changes that might change your health. You can imagine your DNA as a book full of recipes for your body. Every recipe tells your body how to make proteins that keep you healthy, and sometimes there might be a typo in the recipe, like missing an ingredient or mixing up the steps. This could result in a health problem, just like how a changed recipe can lead to a bad dish.

On average, we would expect about 5 million out of our 3 billion DNA letters to be different. And each of these, we call them a genetic variant. Genomic testing is designed to examine some of these variants to help inform our healthcare. So, for example, in understanding why certain health problems happen and in choosing the best treatment based on our unique genetic makeup.

**Florence: And what do we mean by primary findings?**

**Adrianto:** Primary findings mean that in a patient's genomic testing, we identified a set of variants that is linked to the patient's condition. The variance that we have makes us who we are. However, not all of them cause a disease or contribute to a health problem. our bioinformatics pipelines will automatically prioritise variants of potential relevance to the patient's conditions. Using this data, the NHS clinical scientists will then determine whether any of these prioritised variants are linked to the patient's condition and whether a genetic diagnosis has been identified, which would explain why certain health problems happen.

**Florence: So, then what happens when there are no primary findings?**

**Adrianto:** When no primary findings are found, that means that no genetic diagnosis has been identified. As developments are made and our knowledge of the variance improves over time, additional findings might be identified in the future.

The clinical team responsible for a patient's care may request reanalysis of data according to the national guidance, following a change in the patient's clinical status to inform reproductive decisions, or after significant new disease gene associations have emerged.

In addition, Genomics England also provides the diagnostic discovery pathway where we focus on uncovering new diagnosis, where the participants of the 100,000 Genomes Project, as well as the patient's sequenced through the NHS Genomic Medicine Service

This is meant to be more equitable as we don't rely on the clinical teams to raise individual separate requests.

**Florence: And finally, what do we mean by secondary findings?**

**Adrianto:** Secondary findings are additional findings not related to the conditions in which the patient was recruited for. For example, if a patient was recruited for one type of cancer, but perhaps we found variants linked to a different condition. We explored secondary findings for the 100,000 Genomes Project but we do not do secondary findings for the Genomic Medicine Service.

**Florence: That was Adrianto Wirawan explaining what we mean by ‘no primary findings’. If you'd like to hear more explainer episodes like this, you can find them on our website at** [**www.genomicsengland.co.uk.**](https://www.genomicsengland.co.uk)

**Thank you for listening.**