Eosinophilic Gastrointestinal Disorders and genomics

About this document

This document is designed to give an overview of what is currently known about how genomics might affect Eosinophilic Gastrointestinal Disorders (EGID) including Eosinophilic Oesophagitis (EoE). It is written in **plain English** and is designed to be **read in less than 5 minutes.**

These links are a helpful starting point for understanding 'genomics':

- 'What is genomics' – [infographic]
- 'What is genomics' – [short video]
- Definitions and explanations

What do we know about genomics and EoE?

- Eosinophilic Oesophagitis (EoE) is a **long-term** (chronic) inflammatory condition that is **most likely caused by exposure to food antigens**.[1]
- **EoE affects people of all ages, gender and ethnic backgrounds**.[2]
- Many **people with EoE are also affected by other diseases associated with allergies** including asthma. [3]
- **Dietary elimination therapy is effective for many patients** with EoE, which is therefore the first-line management option. However, **how the immune system is activated by food antigens in EoE remains a mystery**.[1,4,5]

Why genomic research?

Genomic research may lead to better understanding and management of the disease.[3]

Recent research suggests that being affected by the disease might depend on the DNA you inherit. While people from around the world are affected, current research suggests that men with European ancestry are more likely to be affected.[3] Despite this prevalence, the mechanism of how the disease works is likely to be similar regardless of ancestry. [6]

People with EoE often have a specific variations in the parts of their DNA which code for certain proteins used in different parts of the body. For example, the DNA which codes for proteins used in the lining of the throat, stomach and gut (epithelium) would involve a number of 'genes' – and a specific variation (or 'mutation') in these genes may influence how people are affected by EoE.[3]

However, the disease is complex and current research suggests that there are multiple variations in multiple interacting genes. Some of these variations might have a beneficial (protective) effect and others may cause EoE. These different genes may interact, with each gene potentially also being influenced by the environment (for example, by diet).[3]

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Another factor which affects EoE might be which genes are 'switched on' or 'off' – that is, whether the body instructs these parts of the DNA code to build protein.[7] The study of these switches is called 'epigenetics' – which comes from the Greek 'epi' which means 'outside of'. Epigenetic changes are not alterations in the DNA but are molecular tags attached to the DNA molecule that let the body know if it is 'switched on' or 'off'. To add to the complexity, epigenetic switches can be affected by the environment. More research is required to understand these all of these interactions.

How is genomic research helping people already?

There are ways that genomic research can currently help people:

- It can **help us understand the basic underlying mechanisms of the disease**, and what role our genes might play.
- it can **lead to improvements in diagnosis and treatment**. For example, there is a molecular test that has the potential to improve understanding of oesophagitis and enable future design of effective therapies.[7]
- Genomic research also has the potential to **help discover and design specific drugs** (a substance which produces a biological effect) which might help treat the disease or the symptoms in a 'not so distant future'.[3]

The future

There is still much that is not understood. The more people that participate in this kind of research and the more data that is shared, the greater our understanding. Involving people affected by EoE in shaping future research could help ensure that the research is relevant and well-designed.

Who wrote this document?

This document was written by Jack Nunn (La Trobe University) as part of the project 'Genomics Research and Involving People', with support from Paul Lacaze (Monash University), Sarah Gray (ausEE), Hamish Philpott (Eastern Health Australia).

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