The multi-omic approach to diagnosing rare disease

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The multi-omic approach to diagnosing rare disease
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INTRODUCING MULTI-OMICS

- Multi-omics is an approach to biological research which looks at multiple ‘omes’ including the genomics, epigenomics and transcriptomics. (Check out the glossary below if you’re not sure what any terms mean!)
- The 100,000 genomes project\(^2\) is a UK based project which is attempting whole genome sequencing of patients with rare disease and their families to aid in diagnosis.
- From this data we can then carry out multi-omic research, increasing the likelihood of obtaining a diagnosis, which leads us to this project’s research...

Glossary of genetics terms

- **Genomics**: The study of the structure, function and evolution of a person’s genetic material.
- **Epigenomics**: The study of non-sequence level DNA modifications which effects gene activity.
- **Methylation**: The act of adding a chemical methyl group which can effect gene activity levels.
- **RNA**: Ribonucleic acid which does many things, including acting as a messenger (mRNA) that carries instructions from the DNA to the cells to carry out cell duties, like synthesise proteins!
- **Gene expression**: The process of acting on the instructions contained in the active gene.
- **Transcriptomics**: The study of the total sum of mRNA which indicates gene expression levels.
- **Whole genome sequencing**: The impressive process of discovering the entire sequence of an organism’s DNA in a single attempt, also known as high throughput sequencing.

This research project involves...

- Conducting systematic reviews into the current literature surrounding multi-omics and rare disease.
- Epigenomic and transcriptomic analyses of patient samples from the 100,000 genomes project which will have been subjected to whole genome sequencing but it was insufficient to render a diagnosis.
- Specifically I will be studying differential methylation (epigenomics) and gene expression, or RNA levels (transcriptomics).

PATIENT CENTERED RESEARCH

- The paradox of rare disease is that whilst they are individually rare, approximately **350 million people worldwide**\(^1\) are affected...
- There are an estimated **8000 types of rare disorders**, often with variations in clinical presentation, making diagnosis challenging...
- Patients can wait several years for a diagnosis, which can negatively impact prognosis, quality of life and make access to effective treatment and support difficult!

Watch me!
Here’s lots of patient experiences which give insights into living with a rare disease, including difficulties in getting a diagnosis.

So what are researchers doing to help?

HOPE FOR THE FUTURE

By utilising an allied multi-omic approach to studying rare disease we will be able to...

- Improve diagnosis speed and accuracy.
- Improve our understanding of the biological mechanisms behind rare disease.
- Identify potentially novel therapeutics.

Therefore, multi-omic approaches to studying rare disease will positively impact the lives of people living and working with rare disease.

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2. The 100,000 Genomes Project. [Accessed 26/02/2018] Available from: [https://www.genomicsengland.co.uk/the-100000-genomes-project/](https://www.genomicsengland.co.uk/the-100000-genomes-project/)