The multi-omic approach to diagnosing rare disease

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The multi-omic approach to diagnosing rare disease
Katie Kerr, Dr Helen McAneney & Dr Amy Jayne McKnight

The paradox of rare disease is that whilst they are individually rare, approximately 350 million people worldwide 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!

So what are researchers doing to help?
Watch me!
Here's lots of patient experiences which give insights into living with a rare disease, including difficulties in getting a diagnosis.

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 affect gene activity.
- **Methylation**: The act of adding a chemical methyl group which can affect 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.

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