Genomics Session

Session Three is repeated at the following times:

11:30 to 12:30 (1 hour)

13:30 to 14:30 (1 hour)

Overview

A session led by the East of England Genomic Medicine Centre who are working to transform genetic testing in the NHS to help patients with rare diseases access genomic testing.

Who will find this session relevant?

Individuals or family members of individuals with rare disease, especially those who don't yet know the genetic change that causes their disease, including Syndromes Without A Name (SWAN).

Those involved with rare disease support and advocacy groups so that information can be given to members who are struggling to get a genetic diagnosis.

Covering the following

- **Why is it useful to know the genetic change that causes your rare disease?**
- **The problems of traditional genetic testing**
- **What is genomic testing?**
- **The promise of genomics for individuals with rare disease**
- **Genomics research studies for rare diseases including the 100,000 Genomes Project**
- **Things to consider if taking part in a genomic study**
- **The future of genomic testing within the NHS**

Activities

To include:

- Presentation and Q&A session

Dr Gemma Chandratillake

Gemma is trained as a molecular geneticist and genetic counsellor. She is the Education & Training Lead for the East of England Genomic Medicine Centre (NHS). Gemma is a Trustee of the Cambridge Rare Disease Network.

Karola Rehnstrom

Karola has been working on several large-scale genomics projects, including the 100,000 Genomes project and the UK10K. She is currently the Programme Lead for the East of England Genomic Medicine Centre, delivering the 100,000 Genomes Project.

Full biographies can be found in Welcome Packs handed out on the day