Interpretation of Genomic Test Results

A two day multi-disciplinary workshop with a focus on the interpretation of genomic test results for paediatric and childhood onset neurological disorders

The Rougemont Hotel, Exeter

21st - 22nd September

Day 1

Introduction to tools for variant interpretation

- Key concepts for variant interpretation for monogenic disorders
- Workshop - use of resources including; population frequency databases, *in silico* tools, allele frequency calculator
- Decipher workshop
- Use of genome sequencing for Short Tandem Repeat (STR) analysis
- Introduction to the ACGS version of the American College of Medical Genetics variant interpretation guidelines, variants of uncertain significance and the role of the MDT in interpreting results
- Networking

Day 2

Case-based variant interpretation

- Genome/Exome sequencing strategies for clinical diagnostic testing
- Workshop - Deciding which test to do and principles of variant interpretation for rare monogenic paediatric disorders
- The importance of the phenotype in variant interpretation
- Interpretation of copy number variants
- Workshop - Interpreting genomic test results for rare monogenic paediatric disorders
- Communicating genomic results and discussing uncertainty

Key speakers and facilitators

Karen Stals
Prof Caroline Wright
Dr Emma Baple
Dr Arianna Tucci
Dr Matthew Wakeling
Miranda Durkie
Dr Andrew Parish
Dr Ellen Thomas
Martina Owens
Dominic McMullan
Amanda Pichini
Ian Berry

Limited HEE funded places (including accommodation) are available for clinical geneticists, genetic counsellors, paediatricians and paediatric neurologists

Please book before 31/07/2022

To apply for a fully funded place on this course, please complete this form - Interpretation of Genomics Test Results Questionnaire.

If you have any queries, please email UEMS_CPD@exeter.ac.uk or call +44 (0) 1392 722964

NHS South West Genomic Laboratory Hub
NHS South West Genomic Medicine Service Alliance
University of Exeter
NHS Health Education England