Workshop: Implementation of clinical genetic databases

Tuesday 17th December 2013
The Nowgen Centre, 29 Grafton Street, Manchester, M13 9WU, UK

This workshop will provide training for current and future implementers of accessible or shared databases for collection of actionable clinical genotype and phenotype data.

Outline programme

09:00 – 09:30 Welcome/Registration
09:30 – 11:00 Session 1 – tools and resources for genetic database implementation
11:00 – 11:20 Break
11:20 – 13:00 Session 1 continued
13:00 – 13:45 Lunch
13:45 – 15:45 Session 2 – elements of a successful databasing project
15:45 – 16:00 Break
16:00 – 17:00 Session 2 continued
17:00 End of workshop

Session 1

This session will provide information and practical training on tools and resources that are important for the implementation of a clinical genetic database. Topics that will be covered include: database frameworks such as LOVD, DMuDB, DECIPHER; data collection tools and resources such as PhenoTips and Alamut; standards and tools that use standards, such as HGVS, Mutalyzer, HPO and Phenomizer; data formats and data handling.

Session 2

Working in groups delegates will complete exercises that explore the areas and issues that must be addressed by a successful database project, including: project structure, policies, user engagement, data collection strategies and processes, curation activities, standards and interconnectivity.