Genomic Medicine and the dawn of treating the individual

Summary

The role of Clinical Genetics is changing due to the increasing ability to offer next generation sequencing, clinical exomes to assess the coding regions of all of the genes and whole genome sequencing. NIHR supported projects such as Deciphering Developing Disorders and the 100,000 Genome Project have acted as a catalyst to finding new genes and increasing our diagnostic power. This is raising debates about the ethical implications of testing in the NHS and commercial sectors, the impact on screening based on risk, the cost of personalised medicine, bioinformatics capacity and education for clinicians on variant interpretation. More broadly this challenges the way we teach and practice medicine in the hospital and community setting in the 21st century as we re-consider the interplay between humans and machine learning.

Professors' Julian Barwell and Pradeep Vasudevan explained this with examples and suggested how we may approach the challenges of mainstreaming of testing, the rise of big data sets and how these can be used by politicians, academics and the population to drive change and develop equations for well-being, improved diagnostics and prognostics.