THE HUMAN VARIOME PROJECT
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WORKSHOP

4.00 pm - 6.00 pm
6 June 2017

Genetic Variant Interpretation

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While performance and output speed of sequencing platforms keep growing, so does the challenge of interpreting the significance of the list of genetic variants in a given individual. Integrative bioinformatic pipelines, accessible databases and *in silico* prediction tools facilitate this task. However, deciding about the potential pathogenicity of every variant is still an arduous work that requires a high level of expertise. In this workshop, examples of patients studied by NGS will be presented so that the attendees will have the opportunity to apply genetic variant filtering and interpretation protocols. The examples will include familial and sporadic cases of neurodegenerative disorders, intellectual disability and cancer. Proposed criteria, scoring systems and guidelines for variant interpretation will be discussed in order to address the main caveats of variant filtering and pathogenicity assessment.