HUMAN GENOME ORGANISATION
HUGO

veptc2023

2-4 Oct.
Barcelona, Spain

VARIANT EFFECT PREDICTION TRAINING COURSE

will be of interest to:

- molecular geneticists
- clinical geneticists
- genetics researchers
- molecular diagnostics labs
- data analysts
- medical specialists with interests in genomics
- anyone interpreting DNA variants on a regular basis

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Don't miss this opportunity to enhance your skills and advance your career

VARIANT EFFECT PREDICTION TRAINING COURSE

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Analysis and interpretation of variations and their effects have been aided by various computational tools however end users often do not know which method or tools to choose, and how to use these tools to their fullest potential.

This course aims to assist you to:

- find and use the best tools for variant interpretation using various genome browsers,
- walk you through HGVS nomenclature with opportunities to discuss difficult to name variants,
- identify what method to apply and determine where things can go wrong in NGS,
- classify variants using ACMG classification system and learn recent recommendations,
- introduce you to Human Phenotype Ontology, (HPO) and learn how HPO´s can help you in variant prioritization,
- as well as explore topics such as RNA analysis and potential consequences, the mitochondrial genome and more.

If you are interpreting DNA variants on a regular basis we will cover all the basics you need to know and more in mixture of lectures & practical workshops.

Topics:

- Variants in the genome, position & possible consequences
- NGS: what method to apply (gene panel, WES or WGS) and where technology fails (inc. CNV & SNV calling)
- Variant Calling: short read vs long read
- How to use The Integrative Genomics Viewer (IGV) for different applications
- HGVS Nomenclature: describing variants workshop
- Human Phenotype Ontology (HPO)
- Integration of phenotypic and genomic data to diagnose patients with rare diseases workshop
- Annotating variants: Variant Effect Predictor workshop
- Annotating variants: Variant Annotation Integrator workshop
- Ensembl Genome Browser workshops: basic & advanced
- UCSC Genome Browser workshop
- General variant databases: OMIM, dbSNP, ExAC etc.
- Locus Specific Databases
- DNA diagnostics = sharing data
- Potential Consequences on the RNA Level
- RNA analysis: expanding the Dx yield (practice)
- Variants in the Mitochondrial Genome
- Long Read Sequencing Approaches
- Variant Classification: ACMG recommendations
- ACMG Classification workshops: basic & advanced
- MobiDetails - DNA Variant Interpretation
- NGS in Diagnostics: where things can go wrong workshop
- UCSC recommended track sets
- Functional Testing of Variants
- Future Developments
- & more...

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