

# **Variant effect prediction**

Course 2542

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April 10 to 13 2018

Avans Hogeschool  
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**International Post-graduate Course**  
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**Tuesday April 10, 2018**

**The basics**

- 13.00 Arrival and registration  
*(Dr. Martie Verschuren, Avans Hogeschool)*
  
- 13.15 General course introduction [2]  
*(Dr. Martie Verschuren, Avans Hogeschool)*
  
- 13.30 Lecture: Variants in the genome, position and possible consequences  
*(Prof. dr. Johan den Dunnen, Clinical Genetics & Human Genetics, LUMC, Leiden)/ Ruben Martherus Avans*
  
- 14.15 Lecture: Calling DNA variants (SNV, SV and CNV) and limitations from sequencing technologies  
Dr. Victor Guryev, UMCG/ERIBA, Groningen
  
- 15.00 Break
  
- 15.45 Lecture: HGVS nomenclature - describing variants  
*(Prof. dr. Johan den Dunnen, Clinical Genetics & Human Genetics, LUMC, Leiden)*
  
- 16.30 Lecture: DNA variants - the big databases  
*(Dr. Peter Taschner HSLeiden)*
  
- 17.15 General discussion with participants
  
- 18.00 End of the course day one
  
- 18.30 Welcome Diner
  
- 20.30 End of day 1

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**Wednesday April 11, 2018**

**Gathering information**

- 09.00 Welcome coffee and introduction of day 2
- 09.30 Lecture: Gene variant databases and the importance of sharing information  
*Dr. Ivo Fokkema, LUMC, Leiden*
- 10.15 Lecture: Human Phenotype Ontology (HPO)  
*Dr. Jeroen van Reeuwijk, RadboudUMC, Nijmegen*
- 11.00 Coffee/Thee
- 11.15 Lecture: Potential consequences on RNA level  
*Dr. Carli Tops, Clinical Genetics, LUMC, Leiden/ Ruben Martherus Avans*
- 12.00 Lecture: Viewing the data  
Ensembl Outreach team, EMBL-EBI Dr. Emily Perry
- 12.45 Lunch
- 13.30 Introduction to the computer practical's
- 13.45 Computer practical, parallel sessions:  
Session 1: genome browser (Ensembl, UCSC)  
Ensembl Outreach team, EMBL-EBI Dr. Emily Perry  
Session 2: Variant nomenclature (Mutalyzer) (30 min)  
Submitting data to a database (LOVD, ClinVar) (bring your data/from an article)  
Prof.dr. Johan den Dunnen
- 17.00 End of the course day two

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**Thursday April 12, 2018**

**Predicting consequences and variant classification**

- 09.00 Welcome coffee and introduction day 3
- 09.30 Lecture: Potential consequences on protein level  
*(Prof. dr. Johan den Dunnen, Clinical Genetics & Human Genetics, LUMC, Leiden)*
- 10.15 Lecture: Protein prediction tools  
*(Dr. Hanka Venselaar, RadboudUMC, Nijmegen)*
- 11.00 Coffee/Thee
- 11.15 Lecture: Variant annotation using VEP  
*(Ensembl Outreach team, EMBL-EBI Dr. Emily Perry)*
- 12.00 Lecture: Variant classification using the ACMG recommendations  
*(Dr. Rolph Pfundt, RadboudUMC, Nijmegen)(to be confirmed)*
- 13.00 Lunch
- 13.30 Introduction to the computer practical's
- 13.45 Computer practical, parallel sessions on:  
Session 1: ACMG *Dr. Rolph Pfundt*  
Session 2: VEP Ensembl Outreach team, EMBL-EBI Dr. Emily Perry
- 17.15 End of day 3

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**Friday April 13, 2018**

**Functional testing and reporting**

- 09.00 Welcome coffee and introduction day 4
- 09.30 Lecture: Functional testing; animal models and lab testing  
*(Dr Frans Verheijen; Erasmus MC, Rotterdam/ Ruben Martherus Avans)*
- 10.30 Coffee/Thee
- 11.00 NIPT, liquid biopsies, quality standards and reporting results  
*(Dr Erik Sijm; VUMC, Amsterdam) to be confirmed*
- 11.45 Future developments  
*(Prof. dr. Johan den Dunnen, Clinical Genetics & Human Genetics, LUMC, Leiden?)*
- 12.30 Evaluation and official closing of the course
- 13.00 End of the course

Avans Hogeschool reserves the right to modify course materials or substitute speakers without notice.