

 EUROPEAN JOINT PROGRAMME  
**RARE DISEASES**

**4<sup>th</sup> Training Course**

***“Quality assurance, variant interpretation and data management in the NGS diagnostics era”***

**October 19-21, 2022  
ONLINE**



## Program of the Training Course

19-21 October 2022 | online

### Wednesday, 19 October 2022

Berlin time (CEST)

**10:00 Welcome & Introduction**

*Holm Graessner, University of Tübingen, Tübingen, Germany & Gert Matthijs, KU Leuven, Leuven, Belgium*

**10:10 Basics of NGS diagnostics**

NGS-based diagnostic approaches for rare diseases  
*Tobias Haack, University of Tübingen, Tübingen, Germany*

Variant classification and re-classification  
*Peter Bauer, Centogene GmbH, Rostock, Germany*

**11:40 Break**

**12:10 Basics of NGS diagnostics – continued –**

Value of re-analysis  
*Katja Lohmann, University of Lübeck, Lübeck, Germany*

**12:55 Hands-on: variant filtering and prioritisation**

Short introduction on data filtering, prioritisation and classification + GPAP  
*nn, CNAG-CRG, Barcelona, Spain*

Case demo introducing key concepts  
*nn, CNAG-CRG, Barcelona, Spain*

- a) Enter structured phenotypic information (PhenoStore)
- b) Explanation and use of phenotypic data to filter and prioritise genomic data
- c) Explanation and use of in silico panels (PanelApp Genomics England)

Explain cases for homework

Questions

**14:00 End of day 1**



Thursday, 20 October 2022

Berlin time (CEST)

**10:00 Hands-on: variant filtering and prioritisation**

Review homework & questions from day 1  
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**10:20 Quality in NGS diagnostics**

[Recommendations for whole genome sequencing in diagnostics for rare diseases](#)

General

*Janneke (Marjan) Weiss, Radboud UMC, Nijmegen, the Netherlands*

Diagnostic strategies

*Janneke (Marjan) Weiss, Radboud UMC, Nijmegen, the Netherlands*

Bioinformatics

*Erika Souche, KU Leuven, Leuven, Belgium*

**12:00 Break**

**12:30 Quality in NGS diagnostics – continued –**

Quality Assessment

*Erika Souche, KU Leuven, Leuven, Belgium*

Ethics

*Helger Yntema, Radboud UMC, Nijmegen, the Netherlands*

**13:10 Communicating genomic results**

[Co-designing models for the communication of genomic results for rare diseases: a comparative study in the Czech Republic and the United Kingdom](#)

*Alessia Costa, Kings College London, London, UK*

**13:40 Hands-on: variant classification**

Explanation and use of variant classification following ACMG guidelines  
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- a) Use of webtools such as VarSome and Franklin – including CNV classification
- b) How to improve computational classification (add criterias manually: e.g. *de novo* parentally confirmed, etc.)
- c) TAG a variant accordingly in the system

Explain cases for homework

**14:00 End of day 2**



Friday, 21 October 2022

Berlin time (CEST)

**10:00 Hands-on: variant classification**

Review homework & questions from day 2  
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**10:20 Innovation & research**

Genome data: How could (should) we use it in the clinic?  
*Olaf Riess, University of Tübingen, Tübingen, Germany*

Economic evaluation of diagnostic innovations in medical genetics  
*Lisenka Vissers, Radboud UMC, Nijmegen, the Netherlands*

**11:50 Break**

**12:20 Innovation & research – continued –**

Long read sequencing for rare diseases  
*Alexander Hoischen, Radboud UMC, Nijmegen, the Netherlands*

Treatabome  
*Antonio Atalaia, Sorbonne University, Paris, France*

**13:50 Wrap-up**

*Gert Matthijs, KU Leuven, Leuven, Belgium*

**14:00 End of workshop**

Agenda may be subject to change.

**SPEAKERS**

**Tobias Haack**, University of Tübingen, Tübingen, Germany

**Peter Bauer**, Centogene GmbH, Rostock, Germany

**Katja Lohmann**, University of Lübeck, Lübeck, Germany

**Janneke (Marjan) Weiss**, Radboud UMC, Nijmegen, the Netherlands

**Erika Souche**, KU Leuven, Leuven, Belgium

**Helger Yntema**, Radboud UMC, Nijmegen, the Netherlands

**Alessia Costa**, Kings College London, London, UK

**Olaf Riess**, University of Tübingen, Tübingen, Germany

**Lisenka Vissers**, Radboud UMC, Nijmegen, the Netherlands

**Alexander Hoischen**, Radboud UMC, Nijmegen, the Netherlands

**Antonio Atalaia**, Sorbonne University, Paris, France



### **COURSE DIRECTOR**

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### **SCIENTIFIC SECRETARIAT**

**Gert Matthijs**, KU Leuven, Leuven, Belgium

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