EUROPEAN JOINT PROGRAMME RARE DISEASES

ESHG EUROPEAN SOCIETY OF HUMAN GENETICS EuroGentest

4th Training Course

"Quality assurance, variant interpretation and data management in the NGS diagnostics era"

October 19-21, 2022

ONLINE

This Training has received funding from the European Union's Horizon 2020 Research and Innovation Programme under Grant Agreement No 825575 - European Joint Programme on Rare Diseases.

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
Jorge Pinto-Basto, 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**

Introduction
Sergi Beltran, CNAG-CRG, Barcelona, Spain

Case demo introducing key concepts
Steve Laurie, 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**

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Thursday, 20 October 2022 — Berlin time (CEST)

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

Review homework & questions from day 1
Steve Laurie, CNAG-CRG, Barcelona, Spain

10:20 **Quality in NGS diagnostics**

Recommendations for whole genome sequencing in diagnostics for rare diseases

Introduction and diagnostic strategies
Janneke (Marjan) Weiss, Radboud UMC, Nijmegen, the Netherlands

Bioinformatics and quality assessment
Erika Souche, KU Leuven, Leuven, Belgium

12:00 **Break**

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

Ethical considerations and reporting
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
Leslie Matalonga, CNAG-CRG, Barcelona, Spain

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**

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Friday, 21 October 2022 — Berlin time (CEST)

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

Review homework & questions from day 2
Leslie Matalonga, CNAG-CRG, Barcelona, Spain

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

Treatabolome
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.

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SPEAKERS

Antonio Atalaia, Sorbonne University, Paris, France

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Alessia Costa, Kings College London, London, UK

Tobias Haack, University of Tübingen, Tübingen, Germany

Alexander Hoischen, Radboud UMC, Nijmegen, the Netherlands

Steve Laurie, CNAG-CRG, Barcelona, Spain

Katja Lohmann, University of Lübeck, Lübeck, Germany

Leslie Matalonga, CNAG-CRG, Barcelona, Spain

Jorge Pinto-Basto, Centogene GmbH, Rostock, Germany

Olaf Riess, University of Tübingen, Tübingen, Germany

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