WELLCOME GENOME CAMPUS
CONNECTING SCIENCE
ADVANCED COURSES+ SCIENTIFIC CONFERENCES

# Genomics of Rare Disease

## 22-24 March 2021

*Virtual Conference Agenda*

| Start (GMT) | Finish (GMT) | Presenter details |
|---|---|---|

## Monday, 22 March 2021

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|---|---|---|
| **13:00** | **13:10** | **Welcome** |
| | | ***Scientific Programme Committee:***<br>*Jennifer Posey, Baylor College of Medicine, USA*<br>*Kaitlin Samocha, Wellcome Sanger Institute, UK*<br>*Lisenka Vissers, Radboudumc University, The Netherlands*<br>*James Ware, Imperial College London, UK* |
| **13:10** | **14:00** | **Keynote** |
| | | Introduction to the session<br>*Chair: Lisenka Vissers, Radboudumc University, The Netherlands* |
| 13:10 | 13:40 | The rise of rare diseases: partnerships in practice<br>*Dian Donnai, University of Manchester, UK* |
| 13:40 | 14:00 | Q&A<br>*Chair: Lisenka Vissers, Radboudumc University, The Netherlands*<br>*Moderator: Jennifer Posey, Baylor College of Medicine, USA* |
| 14:00 | 14:10 | Break |

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| **14:10** | **16:00** | **Session 1: What's New in Rare Disease?** |
| | | Introduction to the session<br>*Chair: Jennifer Posey, Baylor College of Medicine, USA* |
| 14:10 | 14:30 | Rare causes of common conditions and building rare disease communities<br>*Wendy Chung, Columbia University, USA* |
| 14:30 | 14:50 | Next generation cytogenetics – optical genome mapping for comprehensive structural variant detection<br>*Alex Hoischen, Radboudumc University, The Netherlands* |
| 14:50 | 15:00 | Genotype-phenotype correlations related to Robinow syndrome<br>*Chaofan Zhang, Baylor College of Medicine, USA* |
| 15:00 | 15:10 | Accurate long-read whole-genome sequencing identifies pathogenic variants missed with short reads<br>*Aaron Wenger, PacBio, USA* |
| | | Highly commended poster lightning talk<br>*James Poulter, University of Leeds, UK* |
| 15:10 | 15:40 | Q&A<br>*Chair: Jennifer Posey, Baylor College of Medicine, USA*<br>*Moderator: James Ware, Imperial College London, UK* |
| 15:40 | 16:00 | Informal topic discussion |
| 16:00 | 16:20 | Break |

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| 16:20 | 18:25 | Session 2: Developmental and Functional Genomics |
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| | | Introduction to the session<br>*Chair: Lisenka Vissers, Radboudumc University, The Netherlands* |
| 16:20 | 16:40 | Multi-omics integration for molecular diagnostics of Mendelian disorders<br>*Holger Prokisch, Technical University Munich, Germany* |
| 16:40 | 17:00 | Autism genes – do they even exist?<br>*Christian Schaaf, Heidelberg University, Germany* |
| 17:00 | 17:10 | Examining human neurodevelopmental haploinsufficiency disorders in mouse models<br>*Sebastian Gerety, Wellcome Sanger Institute, UK* |
| 17:10 | 17:20 | Transcriptome-based variant calling and aberrant mRNA discovery enhance diagnostic efficiency of neuromuscular diseases<br>*Sung Eun Hong, Seoul National University, South Korea* |
| 17:20 | 17:30 | PRIM1 deficiency causes a distinctive primordial dwarfism syndrome<br>*David Parry, University of Edinburgh, UK* |
| | | Highly commended poster lightning talks<br>*Paul Marcogliese, Baylor College of Medicine, USA*<br>*Aren Marshall, CHEO Research Institute, Canada* |
| 17:35 | 18:05 | Q&A<br>*Chair: Lisenka Vissers, Radboudumc University, The Netherlands*<br>*Moderator: Kaitlin Samocha, Wellcome Sanger Institute, UK* |
| 18:05 | 18:25 | Informal topic discussion |

## Tuesday, 23 March 2021

| 13:00 | 14:50 | Session 3: Therapeutic Approaches to Rare Disease |
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| | | Introduction to the session<br>*Chair: James Ware, Imperial College London, UK* |
| 13:00 | 13:20 | Targeted therapy in patients with PIK3CA related overgrowth syndromes<br>*Guillaume Canaud, Hôpital Necker Enfants Malades, France* |
| 13:20 | 13:40 | A genetically informed paradigm for primary prevention of prion disease<br>*Eric Minikel, Broad Institute, USA* |
| 13:40 | 13:50 | Penetrance of aortic arch defects in 22q11DS can be modulated by dietary vitamin A and response to supplementation or deficiency depends on the mother's genotype<br>*Damian Suñer, Hospital Son Espases, Spain* |
| 13:50 | 14:00 | In search of genetic modifiers that explain the phenotypic variability in SMAD3-related aortopathy<br>*Joe Davis Velchev, University of Antwerp, Belgium* |
| | | Highly commended poster lightning talks<br>*Agnese Feresin, University of Trieste, Italy* |
| 14:00 | 14:30 | Q&A<br>*Chair: James Ware, Imperial College London, UK*<br>*Moderator: Lisenka Vissers, Radboudumc University, The Netherlands* |
| 14:30 | 14:50 | Informal topic discussion |
| 14:50 | 15:00 | Break |

# Genomics of Rare Disease

## 22-24 March 2021

| 15:00 | 15:50 | Poster Session I |
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| 15:00 | 15:10 | Highly commended poster lightning talks<br>*Haowei Du, Baylor College of Medicine, USA*<br>*Julia Dunn, Cincinnati Children's Hospital Medical Center, USA*<br>*Amy Hunter, Genetic Alliance UK, UK* |
| 15:10 | 15:20 | Poster session I lightning talks |
| 15:20 | 15:50 | Poster session I |
| **15:50** | **17:45** | **Session 4: Informatics and Big Data** |
| | | Introduction to the session<br>*Chair: Kaitlin Samocha, Wellcome Sanger Institute, UK* |
| 15:50 | 16:10 | Identification of low-level parental somatic mosaicism for clinically relevant SNVs and InDels in exomes from a large cohort of trios with Mendelian conditions<br>*Tomasz Gambin, Warsaw University of Technology, Poland* |
| 16:10 | 16:20 | Natural Human Knockouts and deep phenotyping in Italian genetic isolates: a different perspective on autosomal recessive Mendelian disorders<br>*Beatrice Spedicati, University of Trieste, Italy* |
| 16:20 | 16:30 | Investigating the contribution of common variants to neurodevelopmental disorders: expression-modifying variants and polygenic scores<br>*Emilie Wigdor, Wellcome Sanger Institute, UK* |
| 16:30 | 16:40 | Large-scale whole-genome sequencing reveals genetic architecture of posterior urethral valves and implicates two novel genetic loci<br>*Melanie Chan, University College London, UK* |
| 16:40 | 16:50 | Deciphering Developmental Disorders in Africa (DDD-Africa) - evaluating clinical exome sequencing in an African setting<br>*Zané Lombard, National Health Laboratory Service, South Africa* |
| | | Highly commended poster lightning talks<br>*Matt Danzi, University of Miami, USA*<br>*Sonia Garcia Ruiz, UCL Institute of Child Health, UK* |
| 16:55 | 17:25 | Q&A<br>*Chair: Kaitlin Samocha, Wellcome Sanger Institute, UK*<br>*Moderator: James Ware, Imperial College London, UK* |
| 17:25 | 17:45 | Informal topic discussion |

# Genomics of Rare Disease

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## Wednesday, 24 March 2021

| 13:00 | 15:00 | Session 5: Mainstreaming Genomics in Healthcare |
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| | | Introduction to the session<br>*Chair: Jennifer Posey, Baylor College of Medicine, USA* |
| 13:00 | 13:20 | Incidental findings in exome sequencing: the Nijmegen experience<br>*Helger Yntema, Radboudumc University, The Netherlands* |
| 13:20 | 13:40 | Cancer Susceptibility Genes: lessons from common and rare phenotypes<br>*Clare Turnbull, Institute of Cancer Research, UK* |
| 13:40 | 13:50 | The PREGCARE study: personalised evaluation of recurrence risk after the birth of a child with a de novo mutation<br>*Anne Goriely, University of Oxford, UK* |
| 13:50 | 14:00 | The singleton missense variant found in a rare adult-onset cancer- SDHB/SDHD as an exemplar for quantifying phenotypic specificity<br>*Chey Loveday, Institute of Cancer Research, UK* |
| | | Highly commended poster lightning talks<br>*James Buchanan, University of Oxford, UK*<br>*Guylaine DAmours, CHU Sainte-Justine, Canada*<br>*Margit Noukas, University of Tartu, Estonia* |
| 14:10 | 14:40 | Q&A<br>*Chair: Jennifer Posey, Baylor College of Medicine, USA*<br>*Moderator: Lisenka Vissers, Radboudumc University, The Netherlands* |
| 14:40 | 15:00 | Informal topic discussion |
| 15:00 | 15:10 | Break |

| 15:10 | 16:00 | Poster Session II |
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| 15:10 | 15:20 | Highly commended poster lightning talks<br>*Angad Jolly, Baylor College of Medicine, USA*<br>*Antony Kaspi, Walter and Eliza Hall Institute of Medical Research, Australia*<br>*Rowan Taylor, University of Leeds, UK* |
| 15:20 | 15:30 | Poster session II lightning talks |
| 15:30 | 16:00 | Poster session II |

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| **16:00** | **17:30** | **Session 6: Non-Coding Variants in Disease** |
| | | Introduction to the session<br>*Chair: James Ware, Imperial College London, UK* |
| 16:00 | 16:20 | Talk title TBC<br>*Connie Bezzina, Amsterdam University Medical Center, The Netherlands* |
| 16:20 | 16:40 | Talk title TBC<br>*Heather Mefford, St Jude Children's Research Hospital, USA* |
| 16:40 | 16:50 | Non-coding region variants cause severe developmental disorder through three distinct loss-of-function mechanisms: the case of MEF2C<br>*Nicola Whiffin, Imperial College London, UK* |
| 16:50 | 17:00 | GREEN-DB: a framework for the annotation and prioritization of non-coding regulatory variants in whole-genome sequencing<br>*Edoardo Giacopuzzi, University of Oxford, UK* |
| 17:00 | 17:30 | Q&A and discussion<br>*Chair: James Ware, Imperial College London, UK*<br>*Moderator: Kaitlin Samocha, Wellcome Sanger Institute, UK* |
| **17:30** | **17:40** | **Closing remarks** |
| | | ***Scientific Programme Committee:***<br>*Jennifer Posey, Baylor College of Medicine, USA*<br>*Kaitlin Samocha, Wellcome Sanger Institute, UK*<br>*Lisenka Vissers, Radboudumc University, The Netherlands*<br>*James Ware, Imperial College London, UK* |