Agenda – Workshop

From high throughput sequencing to diagnosis in immune mediated disorders
ESID/ERN-RITA Workshop

Imagine Institute, 24 Boulevard du Montparnasse, Paris, France
May 30-31, 2022

8:00-8:30 Participant welcoming: Marielle van Gijn (Chair of the ERN RITA Molecular Testing Working Group) and Anne Puel (Chair of the ESID Genetics Working Party)

Day 1: Focus on 'genome' analysis including innovative molecular and informatics tool research and diagnostic methodologies

MORNING

8.30-10.15

NGS in IEI identification: **Pr Isabelle Meyts** (Laboratory of Pediatric Immunology · Department of Microbiology and Immunology · UZ Leuven - Belgium)

From WES to WGS: **Dr Aurélie Cobat** (Laboratory of Human Genetics of Infectious Diseases, Imagine Institute, Paris, France)

Long-read sequencing and human diseases: **Dr Alexander Hoischen** (Lab of Genomic technologies & immuno-genomics, Radboud University Medical Centre, Nijmegen, the Netherlands)

Break 10.15-10.45

10.45-12.45

Mosaicism: **Dr Anna Mensa** (Lab of Immunogenetics of the autoinflammatory response, Hospital Clínic de Barcelona, Barcelona, Spain)

Clinical Genome resources, ClinGen, variant interpretation guidelines: **Pr Janna Saarela** (Institute for Molecular Medicine, Helsinki Finland)

In silico programs as tools for identifying diseases causing variants: **Dr Yuval Itan** (Lab of Human disease genomics and computational biology, Mount Sinai, New York, US)

Epigenetics and immune disease: DNA methylation: **Dr Esteban Ballestar** (Lab of Epigenetics and Immune Disease, Josep Carreras Research Institute (IJC), Barcelona, Spain).

Lunch 13.00-14.00

« This project has received funding from the European Union's Horizon 2020 research and innovation programme under the EJP RD COFUND-EJP N° 825575"

AFTERNOON

14.00-15.30

Human Phenotype Ontology – HPO and IEI: **Dr Marielle van Gijn** (Lab of Genome diagnostic, Department of Genetics, University Medical Centre, Groningen, the Netherlands)

Identification and study of Copy number variation (CNV): **Dr Jérémie Rosain** (Laboratory of Human Genetics of Infectious Diseases, Imagine Institute, Paris, France)

Somatic revertants: Dr Roger Colobran (Lab of Translational Immunology Research Group, Immunology Division / Genetics Department, Universitat Autònoma de Barcelona, Barcelona, Spain)

Gemma database: a new database for curation and search for variants, and high dimensional data in immune relevant genes: Dr Michele Proietti (Center for Chronic Immunodeficiency, University of Freiburg Medical Center, Freiburg, Germany).

Break 15.30-16.00

16.00-18.00

Case presentations by young fellows

Day 2: Functional validation strategies and evaluation of VUS – Patients' perspective

MORNING

8.30-10.30

Monogenic Inflammatory Bowel Disease - genomics and variant validation in clinical practice: Pr Holm Uhlig (Translational Gastroenterology Unit, University of Oxford, Oxford, UK)

Challenging cases from Pr Sophie Hambleton (Translational and Clinical Research Institute, Newcastle University, Newcastle upon Tyne, UK); **Dr Gigliola Di Matteo** (Bambino Gesù Hospital, Rome, Italy); **Dr Kimberly Gilmour** (*Great Ormond Street Hospital*, London, UK); **Dr Vanessa Sancho-Shimizu** (Imperial College, London, UK)

Challenging cases from New Born Screening: Dr Mirjam van der Burg (Laboratory for Immunology, Leiden University Medical Center, Dept. of Pediatrics, the Netherlands)

Break 10.30-11.00

11.00-13.00

Case presentations by young fellows

Lunch 13.00-14.00

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AFTERNOON

14.00-16.00

Patients' perspectives

Patients'perspective: Julie Power (Patient Contact and Policy Officer at Vasculitis Ireland Awareness)

Genetic diseases & Genomics, Genetic Counselling: Dr Marco Crimi (Executive director Kaleidos, Bergamo, Italy)

CLOSURE OF THE WORKSHOP

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