ESID/ERN-RITA Workshop

May 30-31 2022

FROM HIGH THROUGHPUT SEQUENCING TO DIAGNOSIS IN IMMUNE MEDIATED DISORDERS

Imagine Institute, 24 Boulevard du Montparnasse, Paris, France

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

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 WGE: **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)

10:15-10:45 BREAK

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, online)

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

13:00-14:00 LUNCH

14:00-15:30

Human Phenotype Ontology - HPO: **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)

15:30-16:00 BREAK

16:00-18:00

Case presentations: young fellows.

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DAY 2

FUNCTIONAL VALIDATION STRATEGIES AND EVALUATION OF VUS – PATIENTS' PERSPECTIVE

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: 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)

New Born Screening: DR MIRJAM VAN DER BURG (Laboratory for Immunology, Leiden University Medical Center, Dept. of Pediatrics, the Netherlands)

10:30-11:00 BREAK

11:00-13:00

Case presentations by young fellows.

13:00-14:00 LUNCH

14:00-15:00

Patients' perspectives: JULIE POWER (Patient Contact and Policy Officer at Vasculitis Ireland Awareness)

Genetic diseases & Genomics, Genetic Counselling: DR MARCO CRIMI (Executive director Kaleidos, Bergamo, Italy)