ERN | EuroBloodNet

Meeting for unsolved cases of Rare Anemias Disorders based on NGS and / or other laboratory diagnostic tests

Wednesday 28th June 2023 – 14:00-17:00 (CEST)

ONLINE

Agenda:

Meeting chaired by Pr. Béatrice Gulbis *– ERN-EuroBloodNet Co-coordinator & Chair of non-oncological hub, ERASME Hospital*

14:00 14:20 **"The Use of Next-Generation Sequencing in the Diagnosis of Rare Inherited Anaemias: A Joint BSH/EHA Good Practice Paper"**
Noémi Roy - *Oxford University Hospitals NHS Foundation Trust*

14:20 14:40 **"Addressing the Diagnostic Gaps in Pyruvate Kinase Deficiency: Consensus Recommendations on the Diagnosis of PKD"**
Paola Bianchi - *Fondazione IRCCS Ca' Granda Ospedale Maggiore Policlinico Milano*

14:40 17:00 **Cases Discussion:**

- **Natalia Scaramellini** *– Fondazione IRCCS Ca' Granda Ospedale Maggiore Policlinico, Milano*
- **Boglarka Brugos** *– University of Debrecen*
- **Cornelis van der Torren** *– UMC Amsterdam*
- **Panayiota Papasavva** *– Cyprus Institute of Neurology and Genetics*
- **Gonzalo De Luna** *– AP-HP, Henri Mondor Hospital*
- **Beatrice Gulbis** *– ERASME Hospital*

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European Reference Network for rare or low prevalence complex diseases
Network Hematological Diseases (ERN EuroBloodNet)