Parallel session PS 3: Epidemiological and Clinical Research and the data challenge

21 November 15:20 – 17:05

Session leaders: ✓ Hélène Dollfus, Coordinator of ERN-EYE, Strasbourg University Hospital, FR ✓ Iiro Eerola, Unit E2, DG RTD	**Chairs:** ✓ Hélène Dollfus, Coordinator of ERN-EYE ✓ Iiro Eerola, Unit E2, DG RTD
Speakers: **Topic 1.** ✓ Gema Ariceta, Hospital Vall d' Hebron, ES ✓ Simona Martin, Unit F1, DG JRC **Topic 2.** ✓ Ana Rath, Orphanet US14, INSERM, FR ✓ Sergi Beltran Agulló, Centre de Regulació Genòmica, ES **Topic 3.** ✓ Olaf Horst Riess, University of Tübingen, DE ✓ Anthony J Brookes, University of Leicester, UK	**EC support official:** ✓ Hélène Le Borgne, Unit B3, DG SANTE

Aim of the parallel session:

ERNs represent a major task force to boost research and the links between ERNs and research fields have to be tightened. This session will consist in discussing and possibly start reaching a common understanding across ERNs and with external experts on how, in the ERN context, to best:
1) "Collect" data on Rare Diseases (RD) patients across the EU,
2) "Use" this EU data as big data in an ad hoc eco system (including Genome-phenome analysis), and thus:
3) "Solve" with the example of RD project: how genomics solve undiagnosed patients.

Main issues to be addressed:

Topic 1. "Collect" data on Rare Diseases patients across the EU
Data collection challenge: focusing on the purposes of data collection, the use of common solutions and tools (JRC standards, platform and tools) to gather ERN data in an optimised way, building upon the example of ERN ERKNet and of its registries' project ERKReg.

Topic 2. "Use" this EU data as big data in an *ad hoc* eco system
This will include the strategic tools to use the data at the best and show how ERNs can impulse research in the field of rare diseases by better characterising the conditions (natural history of diseases) and by linking with more fundamental research with preclinical aspects.

Topic 3. "Solve" RD project : how genomics solve undiagnosed patients
Many rare diseases patients remain unsolved on genetic grounds, the exemplarity of Solve-RD leans in the challenge of mutualising resources of ERNs as well as outreaching genomic technologies to resolve patients' diagnosis in a research context.

Overall, the session will outline the added value of ERNs as an invaluable task force to collect and use rare diseases data in order solve clinical challenges.
Despite former heterogeneous situations, a common "ERN research strategy" in the research ecosystem is advocated to better address these Research endeavours. The session should underline the future paths to lead to successful research ecosystem where ERNs are key players.

Format of the session:

Dynamic session (very short presentations setting the scene and dynamic exchange of views with very active and organised participation of the chairs)