ERN-Skin PEP & Scientific Days / 20-21 April 2023

Day 1 - Thursday 20.04.2023

9h30	Welcome of participants
10h00-13h35	**Patient Education program - Patient formation**
10h-10h0	Welcome and Introduction. *Christine Bodemer (Necker hospital, Paris)*
10h10-10h40	Patient Education Program (PEP): what does that mean and how to build a PEP. *Dr Cecile Godot (APHP, Necker hospital)*
10h40-11h10	An experience in chronic disease: "Set-up of a patient therapy education program on lipodystrophy syndromes". *Dr Camille Vatier (Inserm, Paris Sorbonne University, APHP)*
11h10-11h20	ERN-Skin survey: where we are ? *Catherine Champseix (ERN-Skin scientific project manager)*
11h20-12h25	**Examples of PEP in skin rare diseases, from different HCPs:**
11h20-11h45	Experience 1: Necker Hospital, Paris : "PEP in genodermatoses: experience of MAGEC-Necker reference center", *Hélène Le Goff*
11h45-12h10	Experience 2: Rotterdam: "PEP on CMN (congenital melanocytic naevus)", *Suzanne Pasmans*
12h10-12h25	Experience 3: -"French Albinism PEP", *Antoine Gliksohn (e-PAG, Association Genespoir)*
12h25-12h55	Open discussion about PEP with patient representatives and scientists - Patients/ Families' expectations *Chaired by Antoine Gliksohn (e-PAG, Association Genespoir)*
12h55-13h10	**Conclusion of the morning:** ERN-Skin PEP: How to progress with all ? *Maya El Hachem (Ospedale Pediatrico Bambino Gesù, IRCCS, Rome)*

13h10-14h10	**Lunch**
14h10-16h40	**Drug repositioning in rare diseases: Interest and difficulties**
14h10-15h10	European rules for clinical trials in rare diseases, *Noémie Manent (EMA)*
15h10-16h40	**Politics and experiences of Companies/start up:**
15h10-15h40	APTEEUS : "From the individual to the cohort, a personalized model for drug repurposing", *Terence Beghyn (CEO, Cofounder of APTEEUS)*
15h40-16h10	ALEXION-ASTRAZENECA RARE DISEASE: "Selumetinib, from clinical research in oncology to rare disease patient benefits", *Manuela Matasconi (Medical Director, Europe and Canada Medical NF1, ALEXION)*
16h10-16h40	CTRS: "Development in rare diseases: academic partnership and repurposing", *Christophe Pompon (Head of the development sCTRS Society, Theravia Group)*
16h40-17h10	**Conclusion**
19h30	*We will meet at O'Bistrot d'Or restaurant, 8 Av. du Maine, 75015 Paris.*

Day 2 - Friday 21.04.2023

8h00	Welcome of participants
08h30-9h30	**Fast talks on scientific <u>update</u>:** (13mn + 2 mn discussion /thematic group) to share research programs that could be valued through the network
	Thematic groups:
8h30–8h45	AIBD: Branka Marinovic: “Autoimmune blistering diseases group (activities)”
8h45-9h00	ED: Holm Schneider: “A causal treatment for hypohidrotic ectodermal dysplasia: Long-term results of short-term prenatal protein replacement”
9h00-9h15	DNA: Fanny Morice-Picard: “Advances in DNA repair disorders”
9h15-9h30	EB: Beyza Sayar: “Readthrough drugs for JEB” Teresa Odorisio: "Histone deacetylase inhibitors as repurposing drugs for recessive dystrophic epidermolysis bullosa".
9h30-10h	**Keynote lecture. Euro-NMD (neuromuscular disorders)**: *Teresinha Evangelista.* “The experience of another ERN: advances and challenges in the last 5 years”.
10h-10h30	**Break**
10h30-11h30	**Interesting and/or puzzling clinical cases (10 min max each case including question)**
10h30-10h40	Diego Martinelli (Rome): "A perplexing case of metabolic neuroichtyosis"
10h40-10h50	Joséphine Hofmann (Munich): “4-year old boy with more than scaling skin”
10h50-11h00	Valentina Ruffo di Calabria (Florence): "A case of bullous pemphigoid with linear deposition of IgM at the demo-epidermal junction and atypical clinical features"
11h00-11h10	Suzanne Pasmans (Rotterdam): “A child with a rare vascular malformation; what would be your advice ?”

11h10-11h20	Marta Ivars (Barcelone): “A case of phakomatosis pigmentokeratotica”
11h20-11h30	Alessandra Gelmetti (Bologna): “About a plate-like osteoma cutis of an adult female: a hard case”
11h30-12h00	**Keynote lecture. “Auto-inflammatory diseases: Lung inflammation and type I interferonopathies: at the single cell level”.** *Marie Louise Fremond (Reference center for rare auto-immmune and inflammatory diseases, Imagine Institute, Paris), Mickael Ménager, (Imagine Institute, Paris)*
12h00-13h00	**Fast talks on scientific update:** (13mn + 2 mn discussion /thematic group) to share research programs that could be valued through the network
	Thematic groups:
12h00-12h15	ALLOCATE: Christos Zouboulis: “Atrophic Papulosis Köhlmeier-Degos: New data on epidemiology and pathogenesis of a rare vasculopathy"
12h15-12h30	IPPK: “Process of Quality of Life Questionnaire”
12h30-12h45	MCTD: Bert Callewaert: “Opposite phenotypes in elastic fiber pathologies: the balance of the fibrotic niche”
12h45-13h00	MOSAIC: Teresa Oranges - *Title tbc* -
13h00-14h00	**Lunch**
14h-15h	**Interesting and/or puzzling clinical cases (10 min max each case including question)**
14h00-14h10	Nathalia Bellon (Paris): “Early and particular cutaneous features in Kindler syndrome”
14h10-14h20	Julie Bonigen (Paris): “Anal sphincter botulinum toxin injection for constipation in RDEB: one case”
14h20-14h30	Yao Wang (Freiburg) : “Late onset EB”
14h30-14h40	Josephine Hofman (Munich): “Update on Morbus Hailey-Hailey and Morbus Darier“

14h40-14h50	Anne-Sophie Sarkis (Bruxelles): "A case of syndromic HS"
14h50-16h00	Smail Hadj Rabia (Paris) : “Congenital heart defects and Ectodermal Dysplasia”
15h-15h30	**Keynote lecture. "Development of a non-viral gene therapy platform for genodermatoses: RDEB treatment as a proof of concept"**. Irene Lara-Sáez (University College Dublin, UCD · Charles Institute of Dermatology)
15h30-16h15	**Summary and further progress**