ELA families-scientists meeting 2022
Scientific Program

Day 1 - Saturday March 26th

Introduction Guy Alba, *president of ELA International* 2 PM CET

Adrenoleukodystrophy ALD – Adrenomyeloneuropathy AMN

Stephan KEMP (The Netherlands): Dutch newborn screening pilot for X-linked adrenoleukodystrophy: The X-Factor 2:15 PM

Florian EICHLER (USA): Upcoming trials for women with ALD and men with AMN 2:35 PM

Wolfgang KOEHLER (Germany): Improving quality of life in women with X-ALD 2:55 PM

Caroline SEVIN (France): ALD in children: screening and treatment in France 3:15 PM

Jean-Hugues DALLE (France): Main results of cell and gene therapy in the treatment of X-linked cerebral adrenoleukodystrophy 3:35 PM

Questions & Answers **3:55 PM**

BREAK 4:25 PM

Genetic and undetermined leukodystrophies - Pelizaeus-Merzbacher Disease PMD - POLR3-HLD 4H

Isabelle THIFFAULT (USA): On the verge of diagnosis: Applying new technologies, dynamic analysis and investigation for new Leukodystrophy genes 4:40 PM

David ROWITCH (UK): Advanced diagnosis, functional genomics and new treatments for Pelizaeus-Merzbacher Disease (PMD) 5:00 PM

Nicole WOLF (The Netherlands): Pelizaeus-Merzbacher disease, POLR3-related leukodystrophies and the rare hypomyelinating leukodystrophies – what can we learn from each disease about hypomyelination 5:20 PM

Vivi HEINE (The Netherlands): A patient stem cell-based research platform for leukodystrophies 6:00 PM

Questions & Answers **6:20 PM**

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Day 2 - Sunday March 27th

Introduction Guy Alba, *president of ELA International* 2 PM CET

Megalencephalic leukoencephalopathy with subcortical cysts MLC, CACH syndrome VWM, Canavan and Alexander diseases

Assumpció BOSCH (Spain): Preclinical gene therapy for megalencephalic leukoencephalopathy with subcortical cysts by specific glial transduction 2:15 PM

Marjo van der KNAAP (The Netherlands): Update on vanishing white matter VWM - CACH syndrome 2:35 PM

Annette BLEY (Germany) : Canavan disease - care and study 2:55 PM

Elly HOL (The Netherlands): Human mini-brains in a dish: a novel technology and its use in developing novel therapies for rare brain diseases 3:15 PM

Angela GRITTI & Vasco MENEGHINI (Italy): Exploring novel gene editing technologies as potential treatments for Alexander's disease 3:35 PM

Questions & Answers **3:55 PM**

BREAK 4:25 PM

Aicardi-Goutières Syndrome AGS, Zellweger spectrum disorders, Krabbe disease and Metachromatic leukodystrophy MLD

Yannick CROW (UK): Aicardi-Goutières Syndrome - an update 4:40 PM

Marc ENGELEN (The Netherlands): Zellweger spectrum disorders: standards of care 5:00 PM

Maria ESCOLAR (USA): Krabbe disease 5:20 PM

Caroline SEVIN (France): Metachromatic leukodystrophy in 2022: how to improve early diagnosis and management? 5:40 PM

Francesca FUMAGALLI (Italy): Gene Therapy: updates and future perspectives on the first approved treatment for early onset metachromatic leukodystrophy 6:00 PM

Questions & Answers **6:20 PM**

END **6:50 PM**