The Ehlers Danlos Society

GENETICALLY DEFINED EDS:

Strategies & Solutions for Unmet Needs

AUGUST 30-31, 2023

BELGIUM

AGENDA

WEDNESDAY AUGUST 30, 2023

TIME (CEST) :	PRESENTATION	SPEAKER(S)	AFFILIATIONS
08:30 - 08:50	Webinar opens for virtual attendees to join		
08:50 - 09:00	Welcome to the event		
SESSION 1: Pathways to Diagnosis			
09:00 - 09:03	Introduction to session theme and speakers		
09:03 - 09:10	Community Voice: The Diagnostic Odyssey	Edward Fraser	
09:10 - 09:20	The Diagnostic Pathway: How To Improve Early Diagnosis of Genetically Defined Types of EDS?	Fransiska Malfait	Ghent University Hospital, Belgium
09:20 - 09:45	Dominant or Recessive? Functional Variant Interpretation in Genetic Diagnostics	Johannes Zschocke	Institute of Human Genetics, Medical University Innsbruck, Austria
09:45 - 10:00	Variants of Unknown Significance	Marco Castori	Division of Medical Genetics Fondazione IRCCS Casa Sollievo della Sofferenza, Italy
10:00 - 10:30	Panel Discussion		
BREAK: 10:30 - 10:45			
SESSION 2: After the diagnosis: Aspects of Genetic Counselling			
10:45 - 11:00	Preimplantation Genetic Testing in Genetically Defined Types of EDS	Chloé Angwin	Imperial College London, UK
11:00 - 11:15	A Clinical Diagnosis of Rare EDS Without Genetic Cause: What Next?	Fleur van Dijk	National EDS Service, North West Thames Regional Genetics Service, London North West University Healthcare NHS Trust Department of Metabolism, Digestion and Reproduction, Section of Genomics and Genetics, Imperial College London, London, UK
11:15 - 11:30	Panel Discussion		
SESSION 3: Classification and Terminology			
11:30 - 11:40	Introduction on Current Classification, Its Limitations and Needs for Change	Fransiska Malfait	Ghent University Hospital, Belgium
11:40 - 12:30	Panel Discussion	Fransiska Malfait, Peter Byers, Johannes Zschocke, Marco Castori, Glenda Sobey	Fransiska Malfait: Ghent University Hospital, Belgium Peter Byers: Pathology at University of Washington Medical Center, Seattle, USA Johannes Zschocke: Institute of Human Genetics, Medical University Innsbruck, Austria Marco Castori: Division of Medical Genetics Fondazione IRCCS Casa Sollievo della Sofferenza, Italy Glenda Sobey: UK Ehlers-Danlos Syndrome National Diagnostic Service, Sheffield Children's NHS, UK
LUNCH: 12:30 - 13:30			
SESSION 4: Natural History and Clinical Variation			
13:30 - 13:40	Community Voice	Amanda Archer	
13:40 - 14:00	Vascular Manifestations in non-vascular EDS: An Underrecognized Problem?	Michael Frank, Karelle Bénistan	Michael Frank: AP-HP, Hôpital Européen Georges Pompidou, Paris, France Karelle Bénistan: Hôpital Raymond-Poincaré – Hôpitaux universitaires Paris Ile-de-France Ouest, France
14:00 - 14:10	Community Voice	Elaine Fox	
14:10 - 14:20	Community Voice	Jose Kusters	
14:20 - 15:15	Natural History Studies	Sherene Shalhub, Jonneke van Gorp, Marlies Colman, Nimah Wilkinson	Sherene Shalhub: Division of Vascular Surgery, School of Medicine, Oregon Health and Science University, USA Jonneke van Gorp: Department of Clinical Genetics, Erasmus MC, University Medical Center Rotterdam, Rotterdam, The Netherlands
BREAK: 15:15 - 15:30			
SESSION 5: Management of a Genetically Defined Type of EDS			
15:30 - 15:40	Community Voice	Christel Mols	
15:40 - 16:30	Pregnancy in Vascular EDS	Glenda Sobey, Michael Frank, Lisa van Berkelaar	Glenda Sobey: UK Ehlers-Danlos Syndrome National Diagnostic Service, Sheffield Children's NHS, UK Michael Frank: AP-HP, Hôpital Européen Georges Pompidou, Paris, France Lisa van Berkelaar: Department of Clinical Genetics, Erasmus MC, University Medical Center Rotterdam, Rotterdam, The Netherlands
16:30 - 16:40	Community Voice	Kevin Aertgeerts	
16:40 - 17:25	Pain in Genetically Defined Types of EDS: Also an Underrecognized Problem?	Delfien Syx, Marlies Colman, Robin Vroman, Margaret Warren-Perry, Ruth Nicholson	Delfien Syx: Ghent University Hospital, Belgium Marlies Colman: Ghent University Hospital, Belgium Robin Vroman: Ghent University Hospital, Belgium Margaret Warren-Perry: Cardiff University, School of Medicine, Cardiff, Wales Ruth Nicholson: Cardiff University, School of Medicine, Cardiff, Wales
17:25 - 17:35	Panel Discussion		
17:35 - 17:55	Update From The Patient Survey of Patient Needs and Challenges	Eva Collado, Charissa Frank	Eva Collado: ePAG ReCONNET Charissa Frank: ePAG ReCONNET
18:00	END		

THURSDAY AUGUST 31, 2023

TIME (CEST) :	PRESENTATION	SPEAKER(S)	AFFILIATIONS
08:30 - 08:50	Webinar opens for virtual attendees to join		
08:50 - 09:00	Welcome to the event		
SESSION 6: Strategies to Study Mechanisms and Implications for Treatment			
09:00 - 09:25	The Use of Induced Pluripotent Stem Cells (iPSC) to Study Heritable Connective Tissue Disorders	Shireen Lamandé	Murdoch Children's Research Institute, Melbourne, Australia
09:25 - 09:35	iPSC in musculocontractural EDS	Tomoki Kosho	Shinshu University, Japan
09:35 - 09:55	The GAGopathies: How to Study Pathomechanisms and Develop Treatment Strategies: The Strength of Collaboration	Göran Larson, Delfien Syx	Göran Larson: University of Gothenburg, Sweden Delfien Syx: Ghent University Hospital, Belgium
09:55 - 10:15	Panel Discussion		
BREAK: 10:15 - 10:30			
10:30 - 10:50	Curative and symptom relief-therapy approaches for collagenopathies - Insights from dystrophic epidermolysis bullosa	Alexander Nystrom	Albert-Ludwigs University Freiburg, Germany
10:50 - 11:10	The MUSC EDS biorepository: A gateway to understanding the rarer forms of Ehlers Danlos Syndromes	Chip Norris	Medical University of South Carolina, USA
11:10 - 11:20	Panel Discussion		
SESSION 7: Organization of Care for Genetically Defined Types of EDS: In Europe and Beyond			
11:20 - 11:35	Organization of the Rare Disease Program for Musculoskeletal Diseases in Hungary	Zoltan Szekanecz	Rheumatology Clinic, University of Debrecen, Hungary
11:35 - 11:45	Organization of Care for EDS in the Netherlands	Serwet Demirdas, Marjin Vis	Serwet Demirdas: Department of Clinical Genetics, Erasmus MC, University Medical Center Rotterdam, Rotterdam, The Netherlands Marijn Vis: Department of Rheumatology, Erasmus Medical Center, Rotterdam, The Netherlands
11:45 - 11:55	Organization of Care for EDS in the UK	Glenda Sobey, Fleur van Dijk	Glenda Sobey: UK Ehlers-Danlos Syndrome National Diagnostic Service, Sheffield Children's NHS, UK Fleur van Dijk: Northwick Park Hospital, London, UK
11:55 - 12:05	Organization of Care in the Rare Disease Center in France	Baptiste Vierne	Hôpital Raymond-Poincaré – Hôpitaux universitaires Paris Ile-de-France Ouest, France
12:05 - 12:15	The Ehlers-Danlos Society Centers and Networks of Excellence	Clair Francomano	
12:15 - 12:30	Panel Discussion		
LUNCH: 12:30 - 13:30			