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Kleefstra Syndrome Scientific Conference 2023

“Moving towards the uptake and use of Artificial Intelligence in research and clinical work”



Ljubljana, Slovenia, Hotel Slon, 1st – 2nd June 2023

The conference brings together Kleefstra syndrome patient organizations, clinicians and researchers from different domains having a common interest: share new research findings related to Kleefstra syndrome that will lead the Kleefstra syndrome community to optimize current care and to reach the final goal, the discovering of a life-changing treatment and cure for Kleefstra syndrome.

A special focus is given to artificial intelligence (AI) and data science, which is generally still something new for the rare disease communities, but it can play a crucial role, especially in shortening the time needed for new research insights.



Radboudumc



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TANJA ZDOLŠEK DRAKŠLER, PhD
Jožef Stefan Institute, SI, NGO IDefine Europe, SI and ERN ITHACA ePAG



Professor TJITSKE KLEEFSTRA, PhD, MD
Erasmus UMC and Radboud UMC, Center of Excellence for Neuropsychiatry at Vincent van Gogh, NL and ERN ITHACA board member

DAY 1: THURSDAY, JUNE 1ST, 2023

9:30 – 10:00 Registration and gathering with networking

10:00 – 10:05 Welcome

Tanja Zdolšek Draksler, PhD, Jožef Stefan Institute and IDefine Europe, Slovenia & prof. Tjitske Kleefstra, MD, PhD, Erasmus Rotterdam UMC, The Netherlands

10:05 – 10:15 An introduction to the Kleefstra syndrome community

Tanja Zdolšek Draksler, PhD, Jožef Stefan Institute and IDefine Europe, Slovenia

10:15 – 10:40 The Kleefstra syndrome journey

prof. Tjitske Kleefstra, MD, PhD, Erasmus Rotterdam UMC, The Netherlands

10:40 – 11:00 Kleefstra syndrome data collection and the Rare Diseases Observatory

Tanja Zdolšek Draksler, PhD, Jožef Stefan Institute and IDefine Europe, Slovenia

11:00 - 11:25 Addressing global challenges related to SDGs with AI

Mitja Jermol, Jožef Stefan Institute, IRCAI, UNESCO Chair on Open Technologies for OER and Open Learning, Slovenia

11:25 - 11:30 Discussion

11:30 - 11:45 Networking and coffee break

11:45 – 12:05 iPSC-derived neural cells and strategies for upregulating the wt allele in patient cells

prof. Hans van Bokhoven, PhD, Radboud University, The Netherlands

12:05 – 12:25 Inverse Molecular Docking as a New Powerful Tool for Drug-Repurposing

prof. Urban Bren, PhD, FKKT, University of Maribor, Slovenia

12:25 – 12:45 Growth and metabolism in Kleefstra syndrome

Arianne Bouman, MD, Radboud UMC, The Netherlands

12:45 – 13:05 EEG data analysis and AI/ML methods for brain conditions

Katerina Tzimourta, PhD, University of Western Macedonia, Greece

13:05 – 13:25 Characterizing sleep in Kleefstra syndrome individuals and fly models

Lara van Renssen, Radboud UMC, The Netherlands

13:25 – 13:30 Discussion

13:30 - 14:30 Lunch break (buffet) with networking discussions

14:30 – 14:40 Patient/caregiver reported data: Genida registry

Pauline Burger, PhD, IGBMC, France

14:40 – 15:00 Data visualisations for clinical and patients use

Alenka Guček, PhD, Jožef Stefan Institute, Slovenia and University of Uppsala, Sweden

15:00 – 15:20 How to tackle unstructured data with data science? Showcase of Genida example (HumaneAI micro project)

Erik Novak, Jožef Stefan Institute, Slovenia

15:20 – 15:40 Patients-centered care plan based on Big Data and AI technologies

Izidor Mlakar, PhD, FERi, University of Maribor, Slovenia

15:40 – 16:00 Cell consequences of loss of function of the epigenetic factor EHMT1

prof. Sabrina Rivero, PhD, University of Sevilla, Spain

16:00 – 16:20 Chromatinopathies study

Laura Pezzoli, MD, ASST Papa Giovanni XXIII, Bergamo & Federica Gaudioso, MD, Policlinico di Milano, Italy

16:20 – 16:30 Wrap up of the day

16:30 – 17:00 Networking discussions with coffee

**DAY 2: FRIDAY, JUNE 2ND, 2023**

9.30 – 10:00 Gathering with coffee

10:00 – 10:20 Dissecting the cell-type specific contribution of EHMT1 to neuronal network function using iPSC-derived brain models

prof. Nael Nadif Kasri, PhD, Radboud University, The Netherlands

10:20 – 10:40 Comprehensive EHMT1 variant interpretation provides insight into EHMT1 functions

Dmitrijs Rots, PhD, Erasmus Rotterdam UMC, The Netherlands and Children's Clinical University Hospital, Latvia

10:40 – 11:00 The establishment and characterization of iPSC derived 3D neurosphere cultures from a Kleefstra patient

Andrea Balogh, PhD, BioTalentum, Hungary

11:00 – 11:20 Advanced treatments and its transfer to patients

prof. Roman Jerala, PhD and Mojca Benčina, PhD, National Institute of Chemistry, Slovenia

11:20 – 11:40 AI/ML in healthcare

assist. prof. Aleksander Sadikov, PhD, Faculty of Computer and Information Science, University of Ljubljana

11:40 – 11:50 Discussion

11:50 – 12:10 Networking and coffee break

12:10 – 12:30 Kleefstra syndrome speech and language study

Lottie Morrison, Murdoch Children's Research Institute, Australia

12:30 – 12:50 Kleefstra and Kleefstra-like syndromes in the Spanish Undiagnosed Rare Diseases Program

Maria José Barrero, PhD, National Institute of Health Carlos III, Spain

12:50 – 13:10 Innovative approach towards the national organization of rare disease management – Slovenian National plan

assist. prof. Luca Lovrečić, MD, PhD, Ljubljana UMC, Slovenia

13:10 – 13:30 GestaltMatcher: rare disease matching using facial phenotypic descriptors

prof. Peter Krawitz, PhD, Universitätsklinikum Bonn, Germany

13:30-13:40 Discussion

13:40-14:40 Lunch break (buffet) with networking discussions

14:40 – 15:00 Unlocking New Possibilities for Rare Disease Healthcare Professionals and Patient Communities Through AI

Joao Pita Costa, PhD, Jožef Stefan Institute, Slovenia

15:00 – 15:20 The QualiSID system and related video analysis technologies for supporting individuals with intellectual disabilities

Vasileios Mezaris, PhD, CERTH, Greece

15:20 – 15:40 Kleefstra syndrome, a hidden metabolic disorder?

prof. Annette Schenck, PhD, Radboud UMC, The Netherlands (online lecture)

15:40 – 16:00 Arrhythmias including Atrial Fibrillation in Kleefstra Syndrome: a possible epigenetic link

Sunil K. Vasireddi, MD, Stanford University, Palo Alto, California, United States (online lecture)

16:00 – 16:30 Wrap up of the day followed with networking coffee