Kleefstra Syndrome Scientific Conference 2023

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

1st – 2nd June 2023

MAIN ORGANIZERS

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

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An introduction to the Kleefstra Syndrome community

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Outline

My story

Non-profit NGO IDefine Europe

Kleefstra syndrome global community and the informal Kleefstra Syndrome Europe Alliance

Kleefstra Syndrome worldwide map
“The passion of parents can move mountains.”
FOXG1 research foundation
IDeﬁne Europe, Foundation for the Advanced Treatment of Rare Genetic Diseases

- Personal involvement in rare diseases
- Professional background in applied research
- Advocating activities

Resulted in starting a non-proﬁt NGO in 2022
- research on rare diseases, highlighting data and new technologies (data science, AI),
- raising awareness of rare diseases and patient advocacy,
- building partnerships as only together we are strong,
- knowledge transfer, learning & training activities.

ideﬁne-europe.org
I Define Europe, Foundation for the Advanced Treatment of Rare Genetic Diseases

- member of Eurordis, umbrella organization for RD in Europe
- European Reference Network for Rare Malformation Syndromes, Intellectual and Other Neurodevelopmental Disorders - ERN ITHACA ePAG (European Patient Advocacy Group)
- Genida - Genetic of Intellectual Disability and Autism Spectrum Disorders (patient reported data) [SAB member]

- Healthcare and health research should be patient-centered
- Patient organizations have a very important role
Kleefstra Syndrome global community

- The Kleefstra Syndrome Europe Alliance, coordination hub for Kleefstra syndrome in Europe, bringing together national Kleefstra syndrome patient organisations/groups from Europe
  - Kleefstra syndrome France
  - Kleefstra syndrome Italy
  - Kleefstra syndrome Spain
  - Kleefstra syndrome.org from UK
  - German speaking group, a community of parents from Germany, Austria and Switzerland
  - Kleefstra syndrome point in Greece
  - Kleefstra community in Netherlands, managed from Zeldsamen
  - Kleefstra syndrome community in Hungary
  - Kleefstra syndrome community in Poland
  - IDefine Europe is the Kleefstra syndrome contact point for Slovenia, Croatia and Serbia

- IDefine (USA)
- Kleefstra community in Australia
- Kleefstra community in Canada
- Kleefstra community in South America