

# Kleefstra Syndrome Scientific Conference 2023

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



1<sup>st</sup> – 2<sup>nd</sup> June 2023



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

## MAIN ORGANIZERS



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



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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.



[define-europe.org](https://define-europe.org)

# IDefine 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

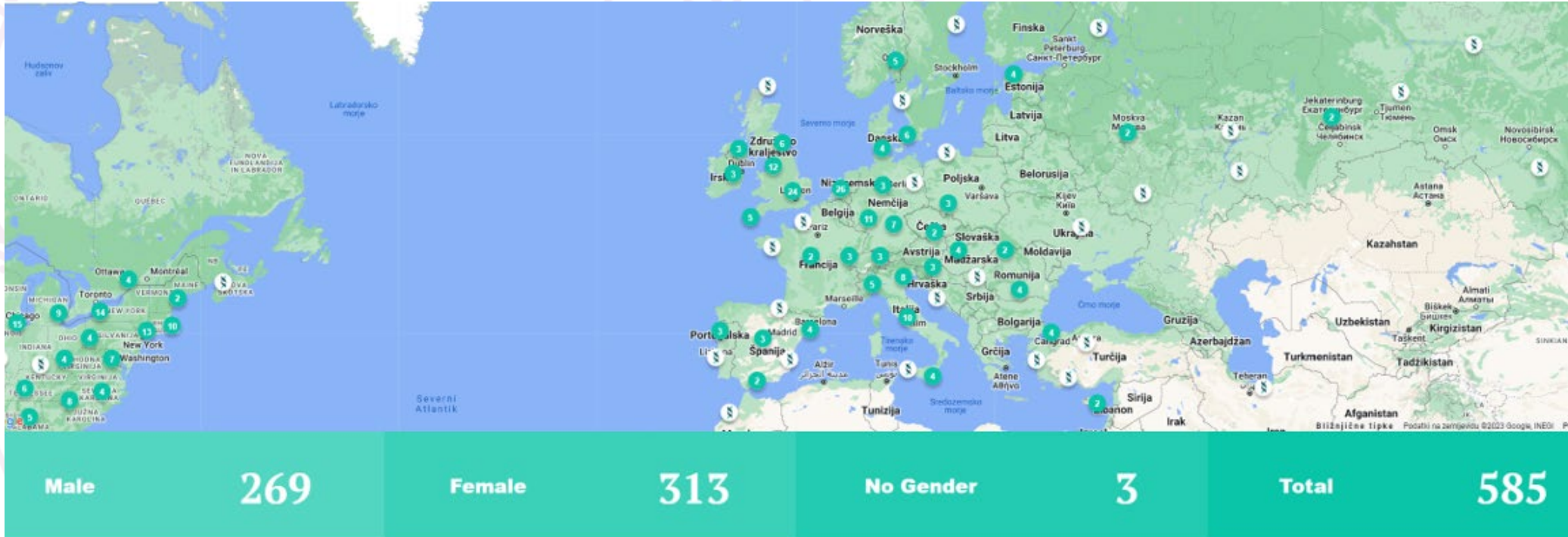
- The Kleefstra Syndrome Europe Alliance, [coordination hub for Kleefstra syndrome in Europe, bringing together national Kleefstra syndrome patient organisations/groups from Europe](#)
  - Kleefstra syndrome France
  - Kleefstra syndrome Italy
  - Kleefstra syndrome Spain
  - Kleefstra syndrome.org from 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



**KLEEFSTRA SYNDROME  
EUROPE ALLIANCE**



# Kleefstra Syndrome worldwide map



<https://www.kleefstraworldmap.org/>