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IDEFINE EUROPE

Kleefstra syndrome data collection and the Rare Diseases Observatory

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HOW IS THE KS COMMUNITY COLLECTING DATA?

- **Kleefstra Syndrome worldwide map: www.kleefstraworldmap.org**
- **GENIDA, collecting caregiver-reported data: https://genida.unistra.fr/**
- **RARE-X, collecting caregiver reported data - https://kleefstra.rare-x.org/**
- **AllStripes, collecting de-identified eHRs (only USA, Canada, UK) – IDefine USA**

LESSONS LEARNED

- **Raising awareness about the importance of data collection is crucial and still needed**
- **Need for learning and training for health literacy and digital literacy**
- **Data collection programs need to adapt to different users – mobile versions crucial, personalized for different users, better UX/UI, enabling different languages, data visualizations…**

RARE 2030 RECOMMENDATIONS (FORESIGHT)

Rare2030
Foresight in Rare Disease Policy

- **No.7: OPTIMISING DATA FOR PATIENT AND SOCIETAL BENEFIT**
- **Data used to its maximum to improve the health and well-being of people living with a rare disease.**
- **Lead by Eurordis**

www.rare2030.eu/recommendations/

RARE DISEASES OBSERVATORY

- **Enabling easy monitoring of rare diseases, providing information and knowledge to empower all: parents, clinicians, researchers, policy makers…**
- **By combining different global data sources**
  - **For monitoring global media news (2014-present): Event Registry system (news intelligence platform),**
  - **For social media monitoring (2008-present): Twitter data (Twitter API for Academic Research)**
  - **For scientific publications monitoring (2001-present): Medline, Microsoft Academic Graph and OpenAlex.**

RARE DISEASES OBSERVATORY - Methodology

- **Data extracted based on 20 specific keywords (16 rare diseases and 4 general terms)**
- **Pilot version RDO presents 16 neurodevelopmental disorders:**

Kleefstra syndrome, Kabuki syndrome, Koolen-de Vries syndrome, Phelan-McDermid syndrome, Pitt Hopkins syndrome, Cornelia de Lange syndrome, Angelman syndrome, Fragile X syndrome, Dravet syndrome, Rett syndrome, SATB2 syndrome, Prader-Willi syndrome, SYNGAP1 syndrome, CTNNB1 syndrome, Wiedemann-Steiner syndrome, FOXG1 syndrome.

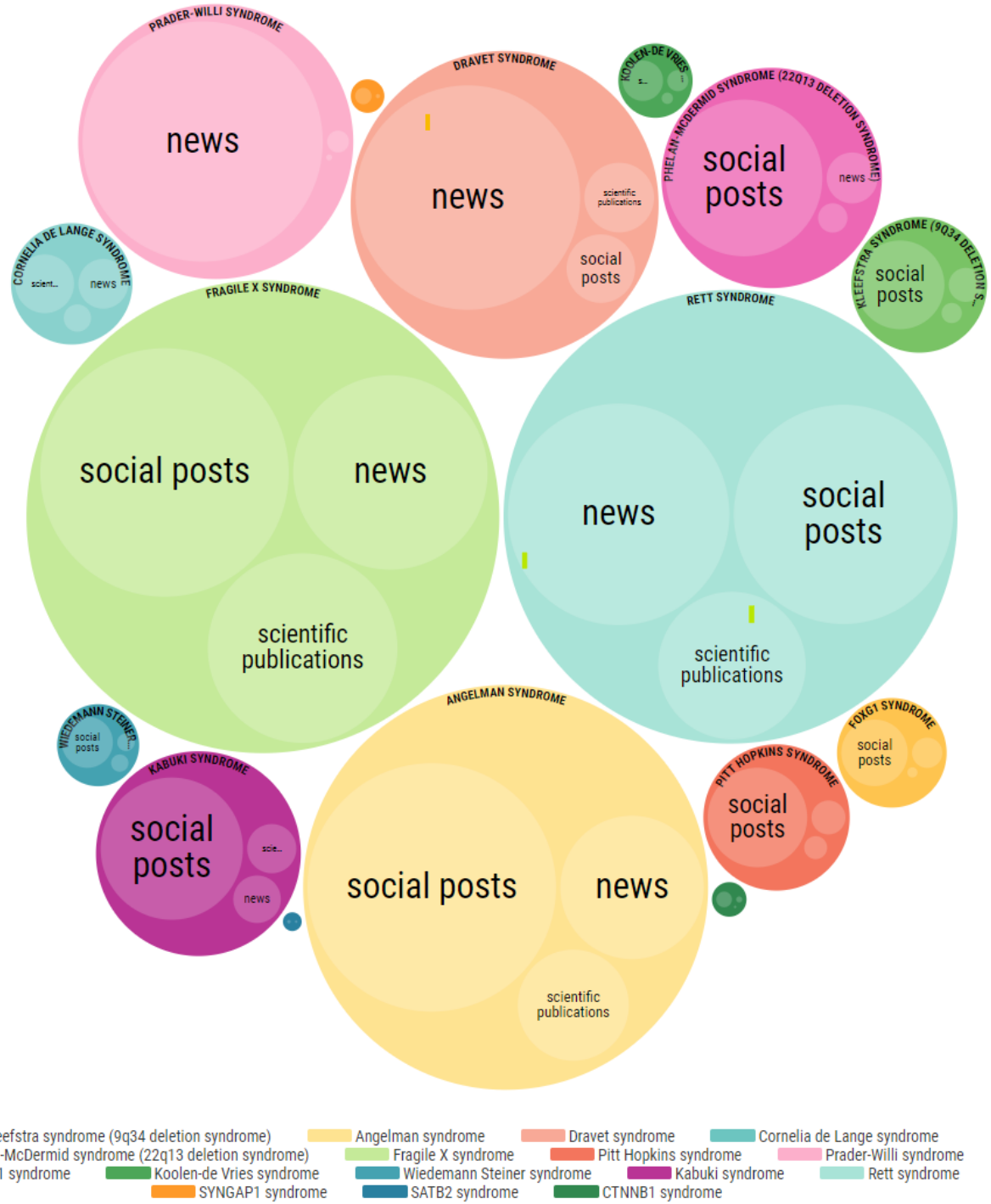
- **ELK Stack (Elastic Search, visualizations: Kibana)**
- **Sentiment analysis: open-source library NLTK - vader_lexicon for Twitter data and performed from EventRegistry for media news**

Future plans

- Increase the number of monitored rare diseases (focus on NDDs stays)
- Custom made visualizations
- Real-time data feed
- New data sources?
- Improved UI/UX
- Other based on focus group and testing users feedback

16 MONITORED NDDs AT A GLANCE

Shows NDDs and their occurrence in global media news, social media, and scientific publications. The area of the circle represents the number of news, social posts, and scientific publications.



MONITORING GLOBAL MEDIA NEWS

news_basic_search_no_filter

34868 documents

Columns | 1 field sorted

	title	authors.na...	source.title
@ 13:13:00	Tonix Pharma Expedites Fibromyalgia and Chronic Migraine Programs By Investing.com	-	Investing.com South ...
@ 13:09:00	Tonix Pharma (TNXP) Expedites Fibromyalgia and Chronic Migraine Programs	-	StreetInsider.com
@ 13:05:00	Tonix Pharmaceuticals Expedites Fibromyalgia and Chronic Migraine Programs	-	Green Stock News
@ 12:54:00	Met Police officers guilty of gross misconduct over racist WhatsApp messages	-	The Independent
@ 11:38:00	Texas A&M Experts Achieve Groundbreaking Phase Angelman Syndrome Development	-	India Education,E...
@ 07:32:00	Colchester police officer to run marathon for friend's child fighting rare illness	-	The Gazette
@ 05:30:00	Trente-sept équipes de futsal au 4e challenge Gard-Ardèche Manon Valla	-	midilibre.fr

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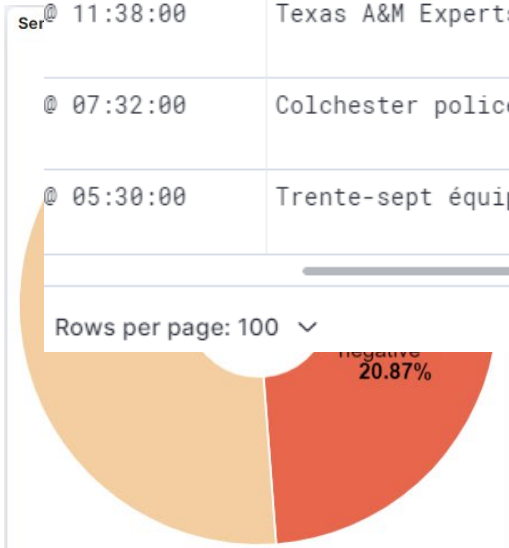
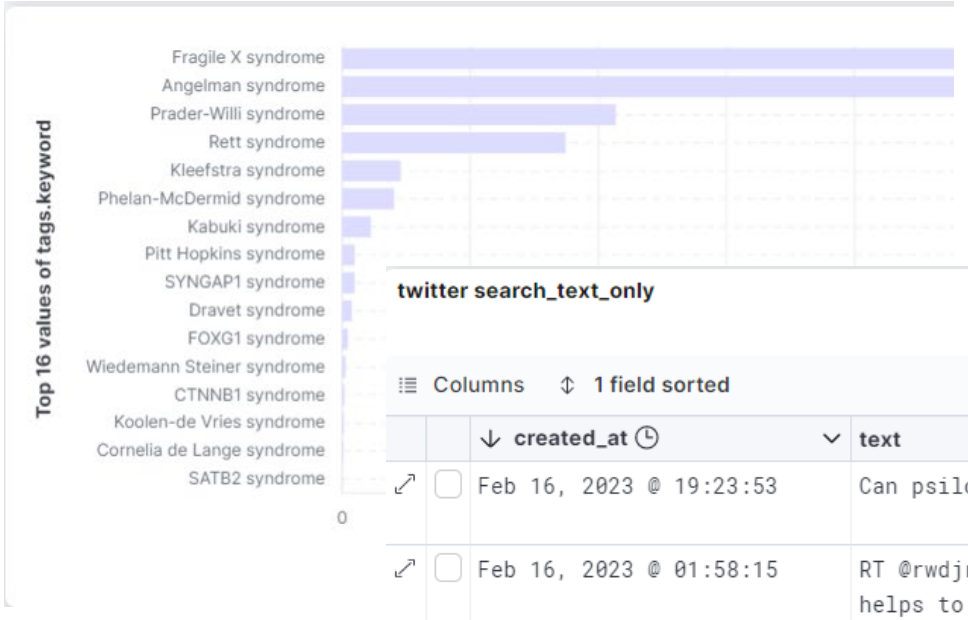
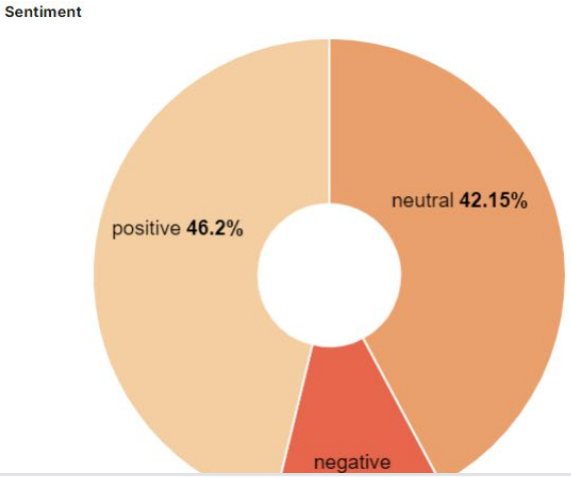
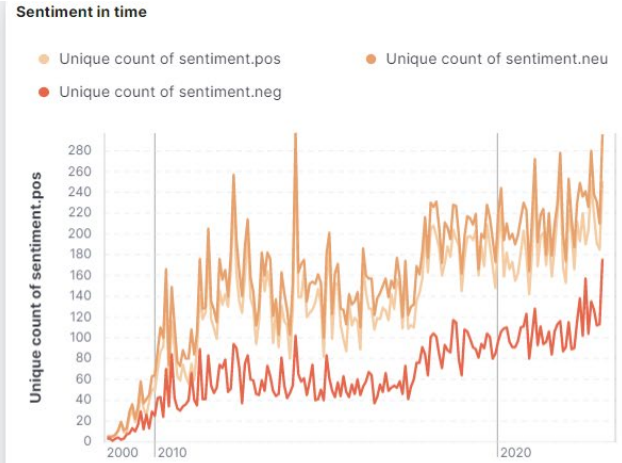


MONITORING SOCIAL M







twitter search_text_only

400702 documents

Columns | 1 field sorted

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Feb 16, 2023 @ 19:23:53	Can psilocybin be used to treat Fragile X Syndrome: https://t.co/qwLcTnEqN2 #psilocybin #fragilex #research
Feb 16, 2023 @ 01:58:15	RT @rwdjr37: I share all this because today is International Angelman Syndrome Awareness Day. I hope this helps to spread awareness about w…
Feb 16, 2023 @ 00:02:32	RT @cureSYNGAP1: New SYNGAP1 blog post from SRF: "Support Simons Searchlight 2023 Campaign - Sign up!" …
Feb 16, 2023 @ 00:01:34	RT @cureSYNGAP1: New SYNGAP1 blog post from SRF: "Support Simons Searchlight 2023 Campaign - Sign up!" …
Feb 15, 2023 @ 23:32:18	RT @cureSYNGAP1: New SYNGAP1 blog post from SRF: "Why Getting a Genetic Diagnosis Matters, Especially for SYNGAP1"…
Feb 15, 2023 @ 23:27:38	RT @cureSYNGAP1: New SYNGAP1 blog post from SRF: "How to Get Free Genetic Testing for People with Autism, Special Needs & Epilepsy"…

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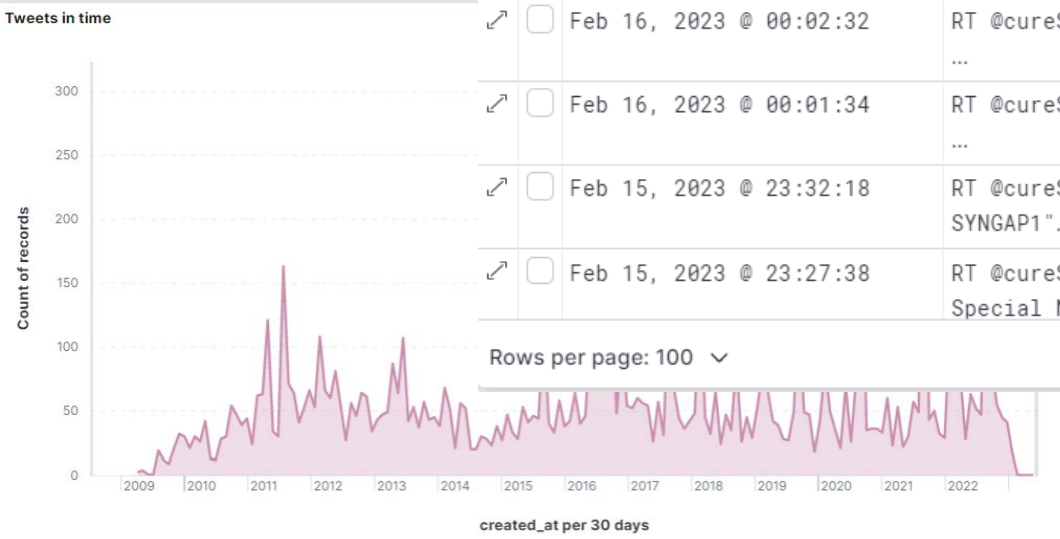

MONITORING SCIENTIFIC PUBLICATIONS

scientific_search_withDOI | Panel filters

15115 documents

Columns | 1 field sorted

public	title	authorships.institutio...	c
Mar 2023	A review on Angelman syndrome	-	h
Mar 2023	Fragile X mental retardation protein coordinates neuron-to-glia communication for clearance of devel…	[Vanderbilt Univers…	h
Mar 2023	Correction: Prader-Willi syndrome patient with atypical phenotypes caused by mosaic deletion in the …	[Tianjin Children's…	h
Mar 2023	Differential Cognitive and Behavioral Development from 6 to 24 Months in Autism and Fragile X Syndro…	[University of Nort…	h
Mar 2023	Negative affect and respiratory sinus arrhythmia are differentially related to social anxiety and au…	-	h
Mar 2023	Investigational new drugs for the treatment of Dravet Syndrome: an update	[University of Krag…	h
Mar 2023	Executive Function and Working Memory Deficits in Females with Fragile X Premutation	[Tel Aviv Universit…	h
Mar 2023	Social Behavioral Impairments in SYNGAP1-Related Intellectual Disability	[Baylor College of …	h
Mar 2023	CTNNB1 in neurodevelopmental disorders	Wenzhou Medical Uni…	h
Mar 2023	Methylmalonic Acidemia Masquerading as Rett Syndrome: An Atypical “Neurodegenerative” Variant?	[Central Institute …	h
Mar 2023	Burden of Illness in Rett Syndrome: Initial Evaluation of a Disorder-Specific Caregiver Survey	[Emory University, …	h
Mar 2023	Mecp2 deletion results in profound alterations of developmental and adult functional connecti…	[Washington Univers…	h
Mar 2023	From circuits to behavior: Amygdala dysfunction in fragile X syndrome	University of Color…	h

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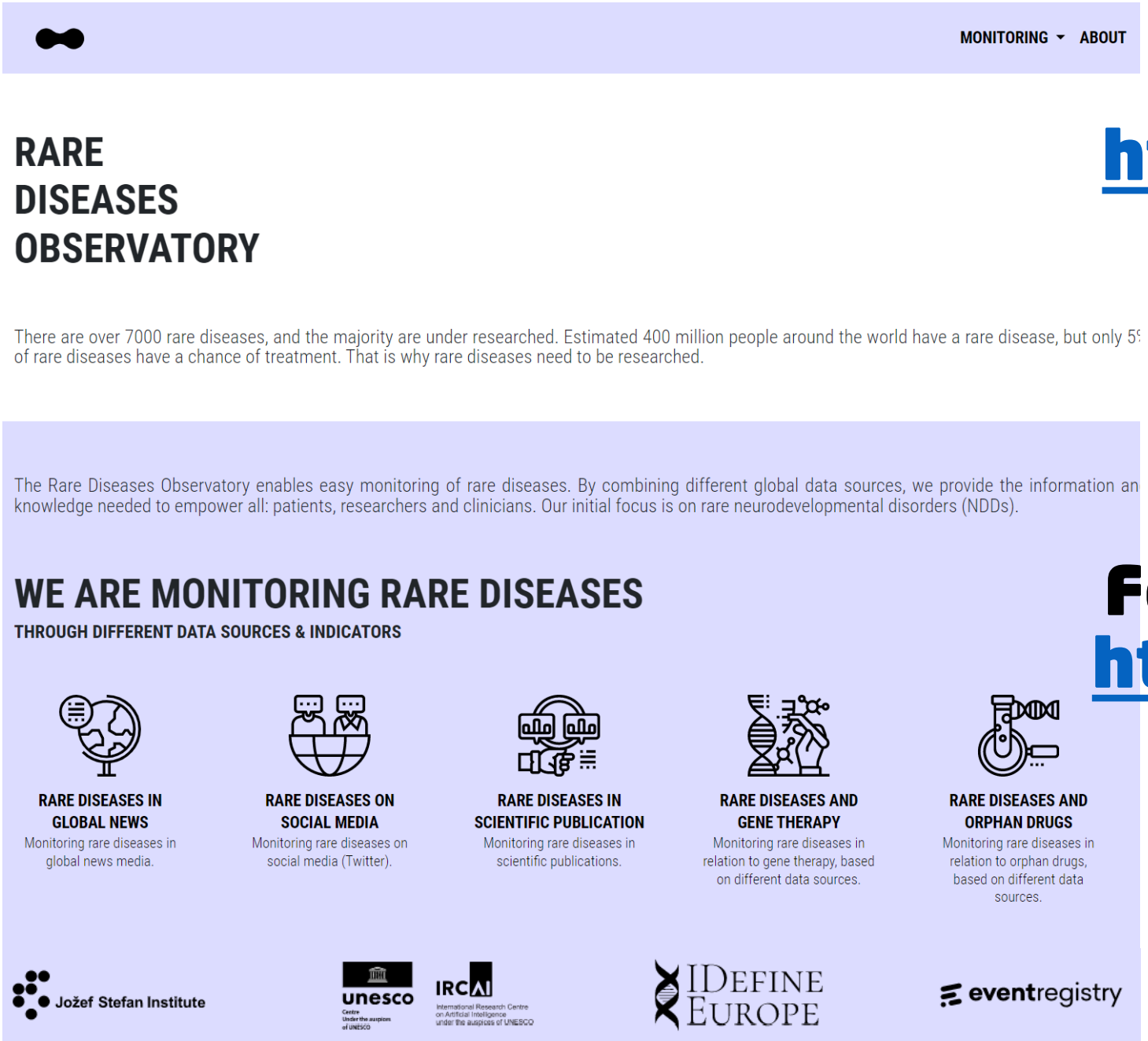

https://rarediseases.ijs.si/

Feedback via GoogleForm
https://forms.gle/cHtJbinsaiTt8FaR8