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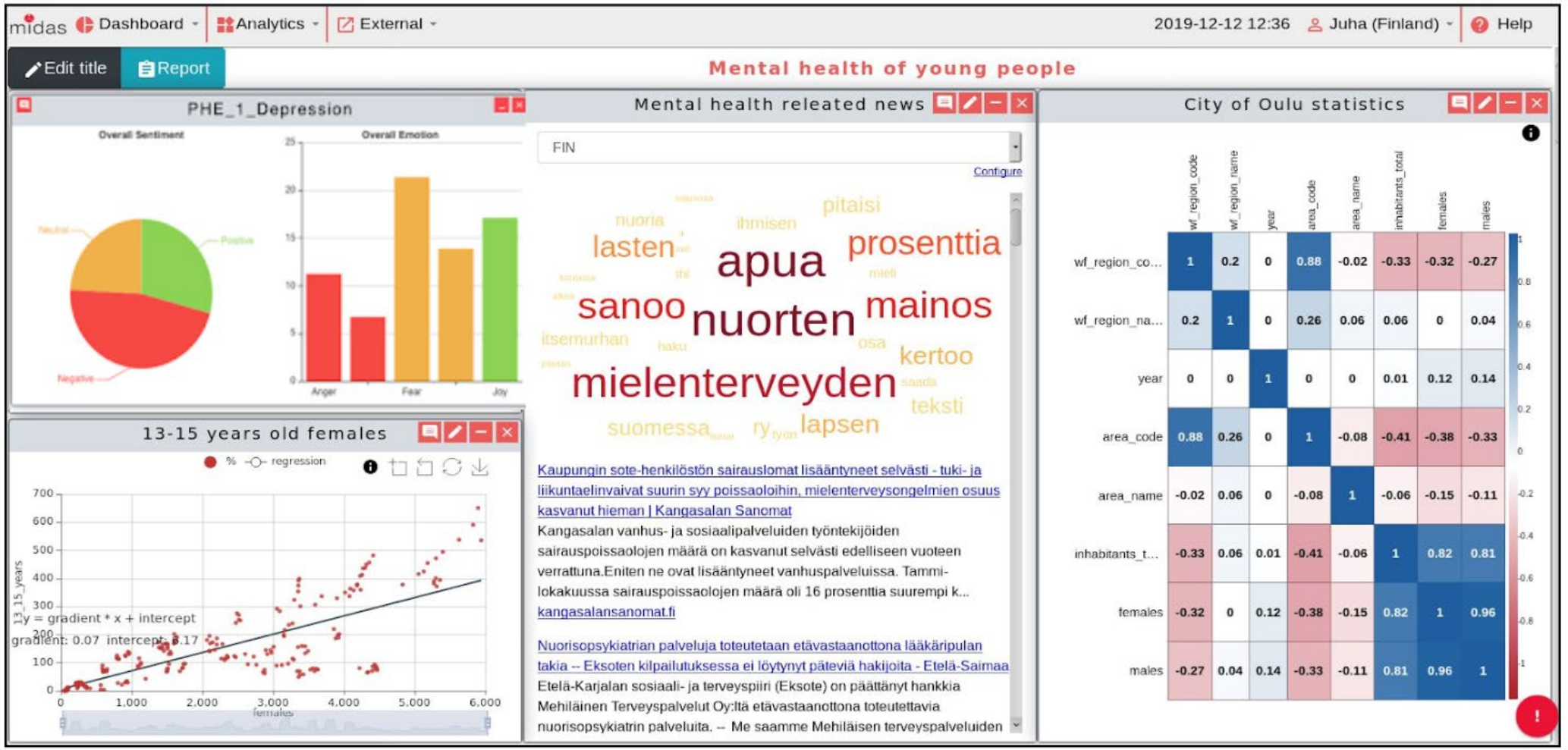
IRCAI SDG Observatory

Unlocking New Possibilities for Rare Disease Healthcare Professionals and Patient Communities Through AI

Kleefstra Syndrome Scientific Conference 2023

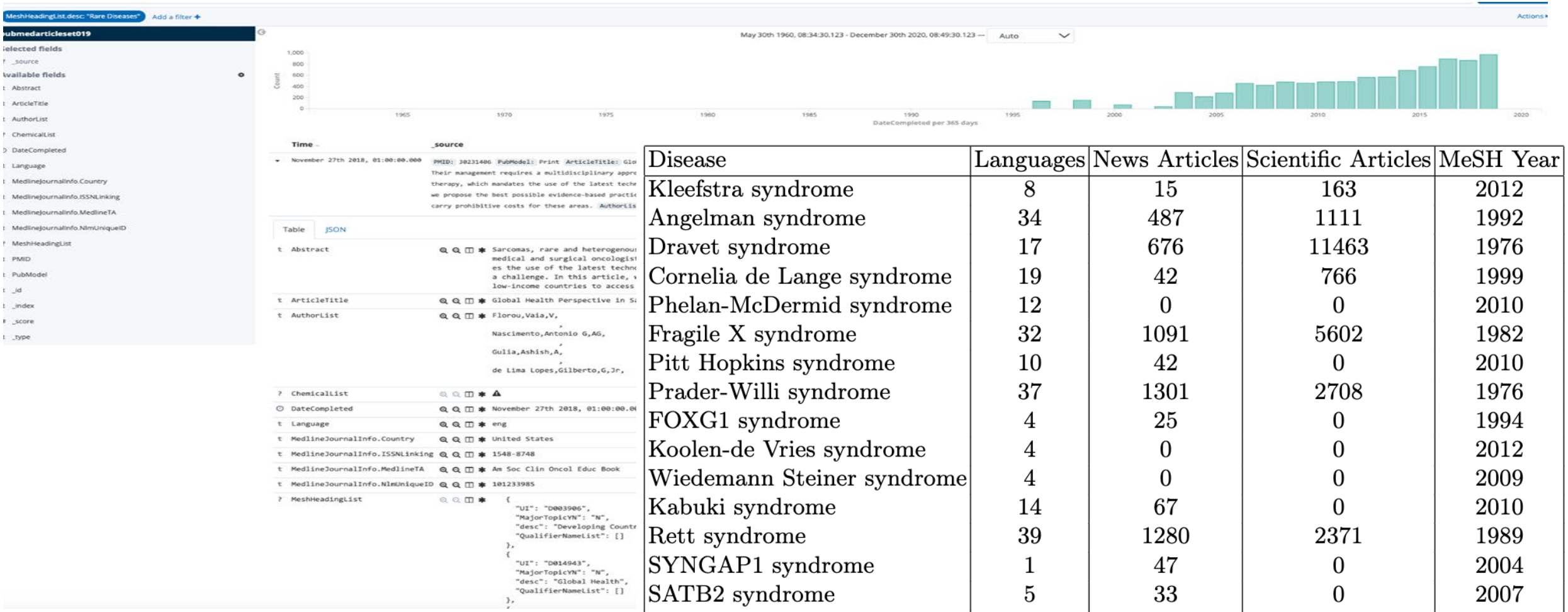
Joao Pita Costa, IRCAI

LESSONS LEARNT FROM DATA - DRIVEN PUBLIC HEALTH



Pita Costa, Joao, Marko Grobelnik, Flavio Fuart, Luka Stopar, Gorka Epelde, Scott Fischaber, Piotr Poliwoda et al. "Meaningful big data integration for a global COVID-19 strategy." IEEE Computational Intelligence Magazine 15, no. 4 (2020): 51-61.

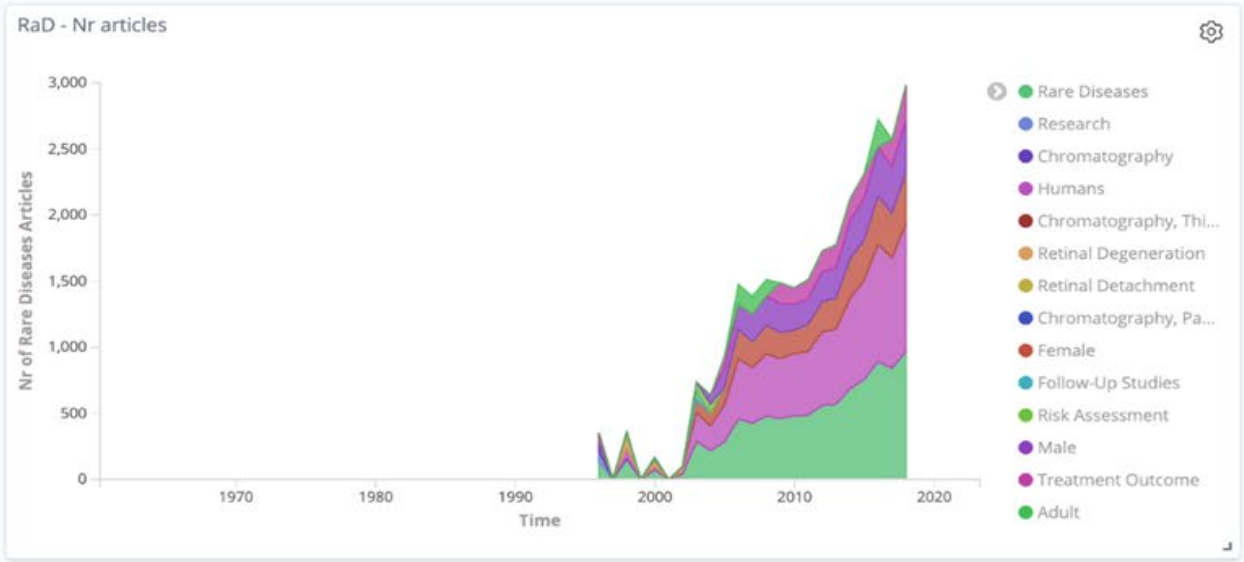
DATA COVERAGE FOR RARE DISEASES

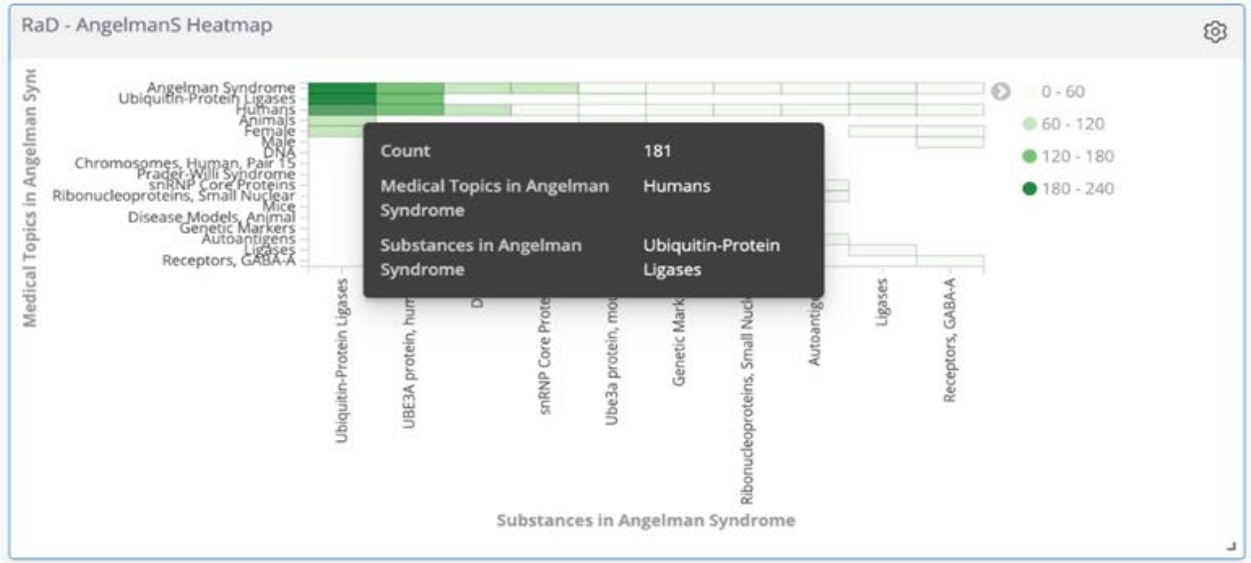


Disease	Languages	News Articles	Scientific Articles	MeSH Year
Kleefstra syndrome	8	15	163	2012
Angelman syndrome	34	487	1111	1992
Dravet syndrome	17	676	11463	1976
Cornelia de Lange syndrome	19	42	766	1999
Phelan-McDermid syndrome	12	0	0	2010
Fragile X syndrome	32	1091	5602	1982
Pitt Hopkins syndrome	10	42	0	2010
Prader-Willi syndrome	37	1301	2708	1976
FOXG1 syndrome	4	25	0	1994
Koolen-de Vries syndrome	4	0	0	2012
Wiedemann Steiner syndrome	4	0	0	2009
Kabuki syndrome	14	67	0	2010
Rett syndrome	39	1280	2371	1989
SYNGAP1 syndrome	1	47	0	2004
SATB2 syndrome	5	33	0	2007
CTNNB1 syndrome	12	142	0	2005

Pita Costa, J., Zdolšek Draksler T. (2023) Exploring rare diseases with automatic text classification and interactive data visualisation on published science and news. Proceedings of the International Conference on Artificial Intelligence in Medicine (AIME)

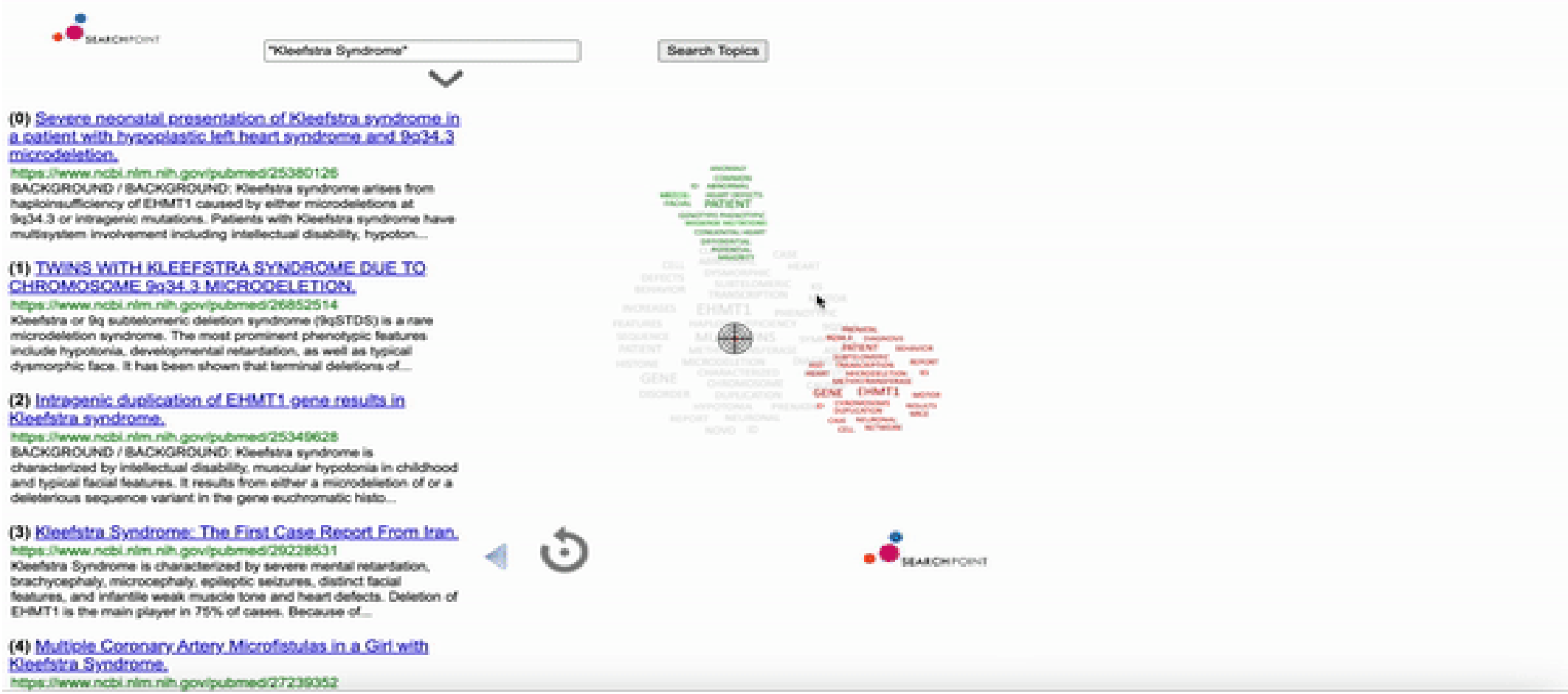
SUPPORTING CITIZEN SCIENCE WITH INTERACTIVE DATA VIZ





Pita Costa, J., Fuart, F., Stopar, L., Paolloti, D., Hirsch, M., Mexia, R., Carlin, P. and Wallace, J., (2019) Local-to-global analysis of influenza-like-illness data. Proceedings of the Slovenian Conference on Data Mining and Data Warehouses (SIKDD)

LITERATURE REVIEW ACROSS 28+ MILLION RESEARCH RESULTS



https://qmidas.quintelligence.com/searchpoint/result.html?q=%22Kleefstra%20Syndrome%22&c=kmeans

Pita Costa, J., Fuart, F., Stopar, L., Grobelnik, M., Mladenic, D. and Kosmerlj, A. (2019) Health news bias and epidemic intelligence for public health. Proceedings of the Slovenian Conference on Data Mining and Data Warehouses (SIKDD)

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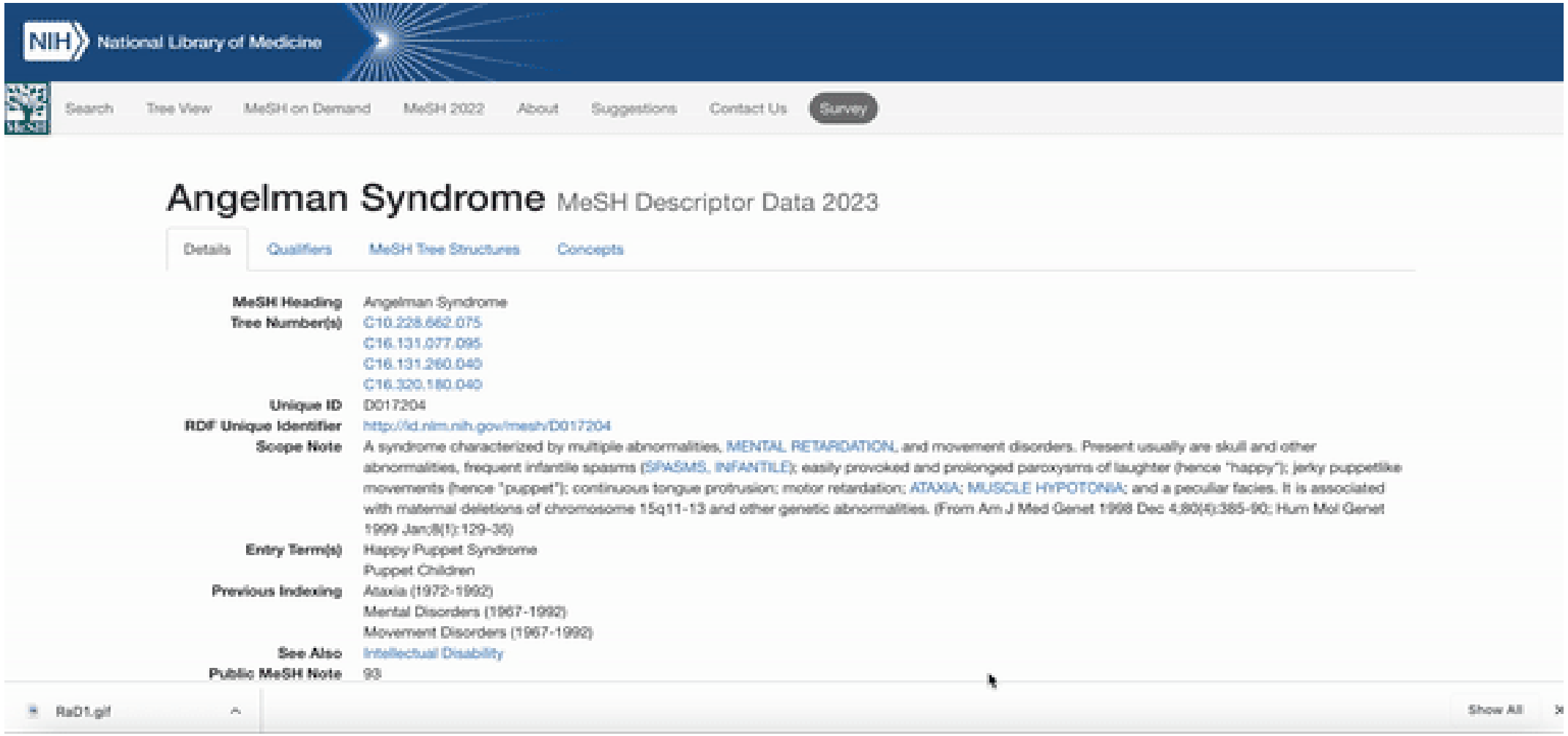
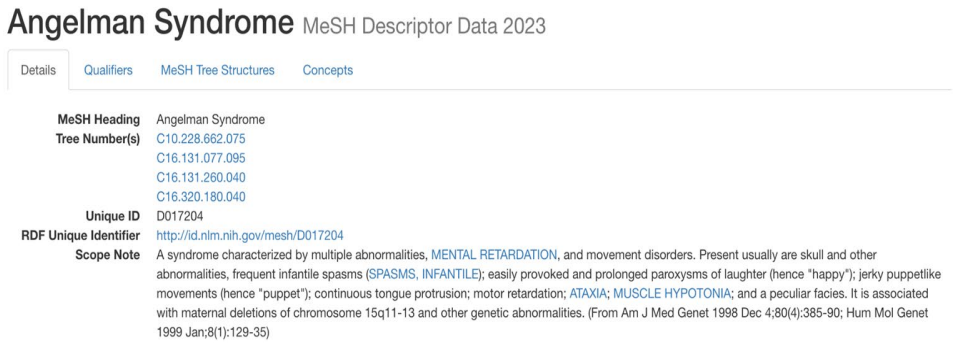
AUTOMATED CLASSIFICATION BASED ON PUBMED CATEGORIES

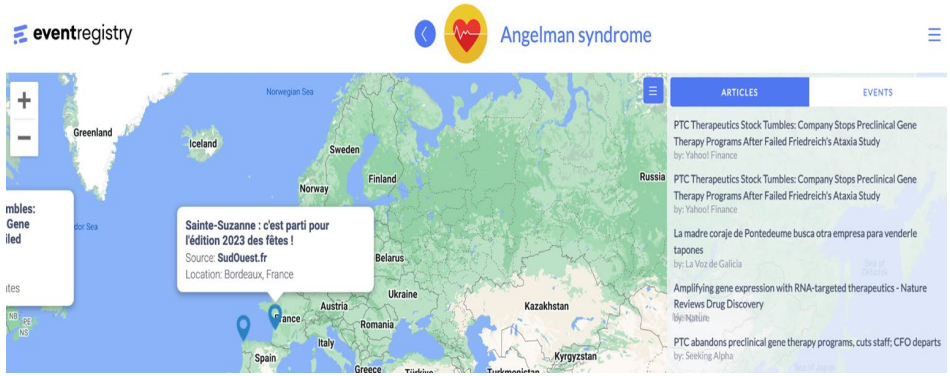


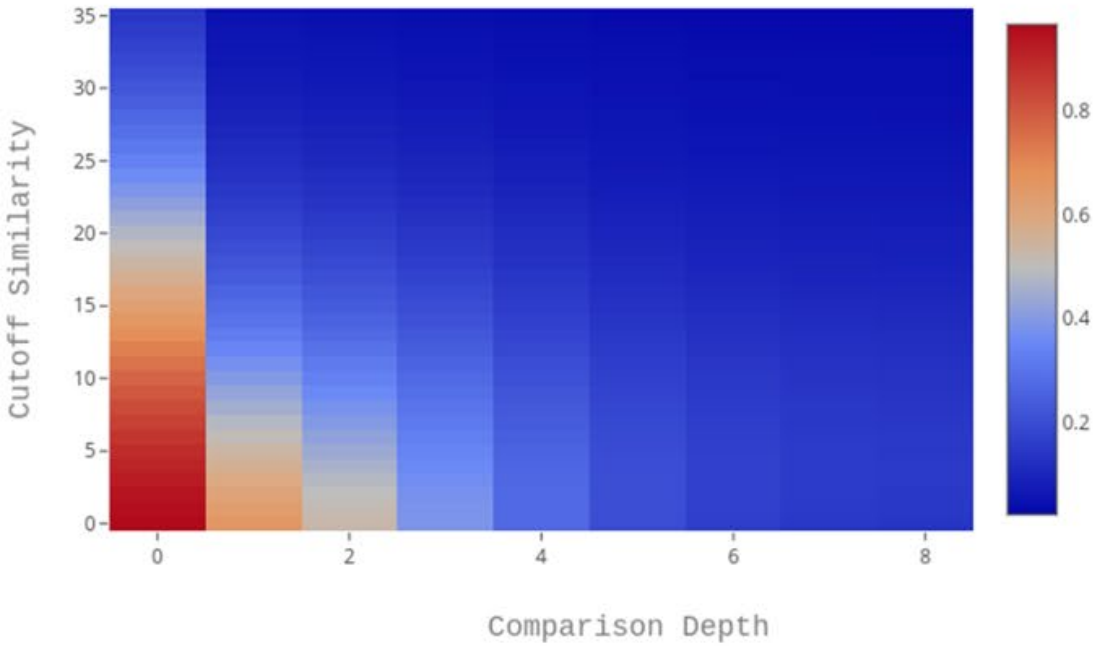
https://qmidas.quintelligence.com/classify-mesh-major/

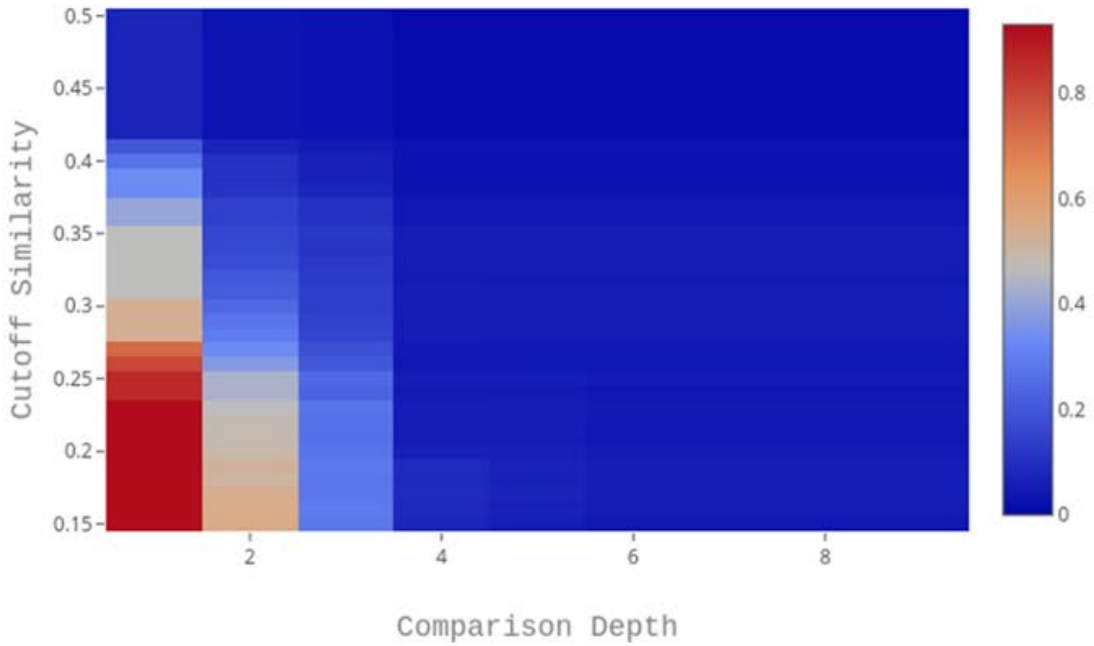
Costa, J.P., Rei, L., Stopar, L., Fuart, F., Grobelnik, M., Mladenić, D., Novalija, I., Staines, A., Pääkkönen, J., Konttila, J. and Bidaurrazaga, J., 2021. NewsMeSH: a new classifier designed to annotate health news with MeSH headings. Artificial Intelligence in Medicine, 114, p.102053.

EVALUATING THE QUALITY OF THE AUTOMATED CLASSIFICATION

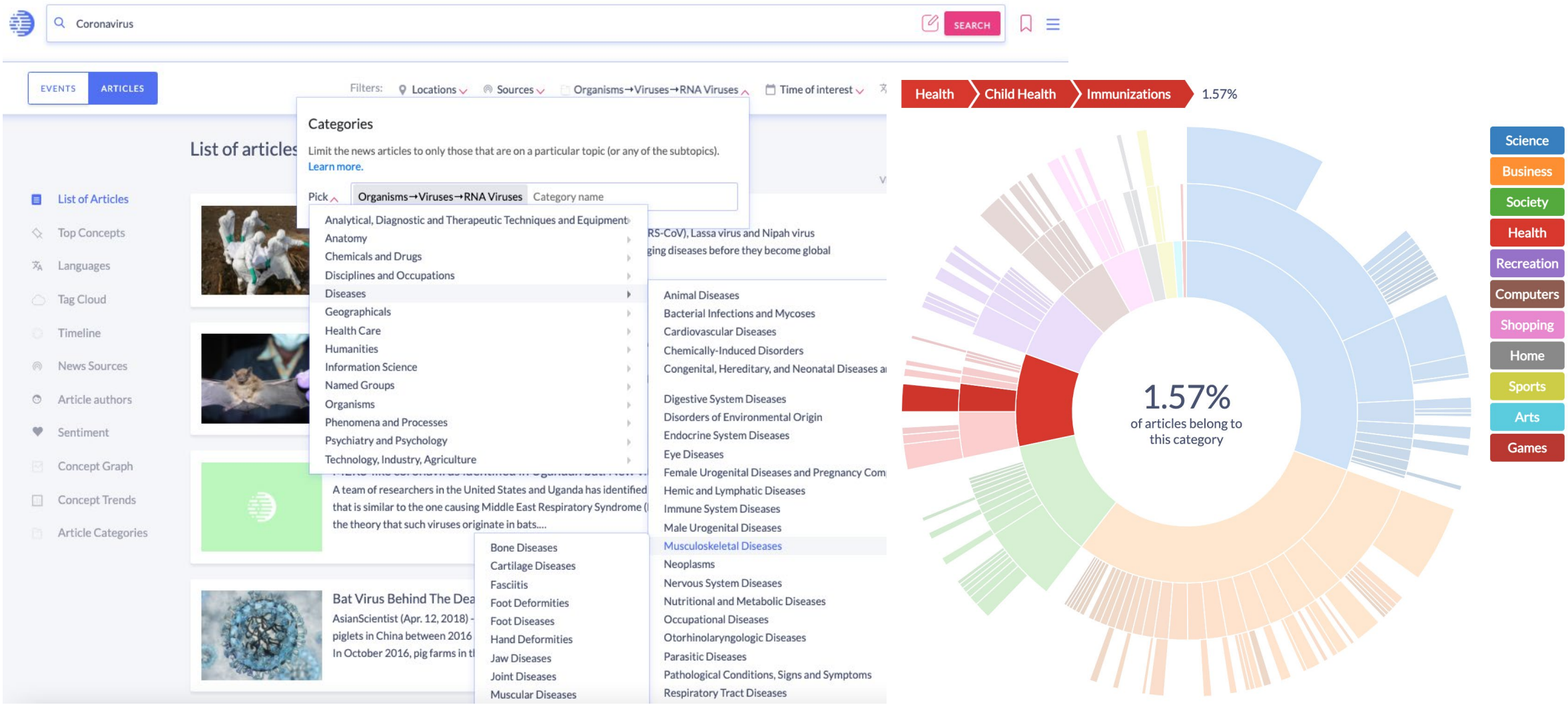


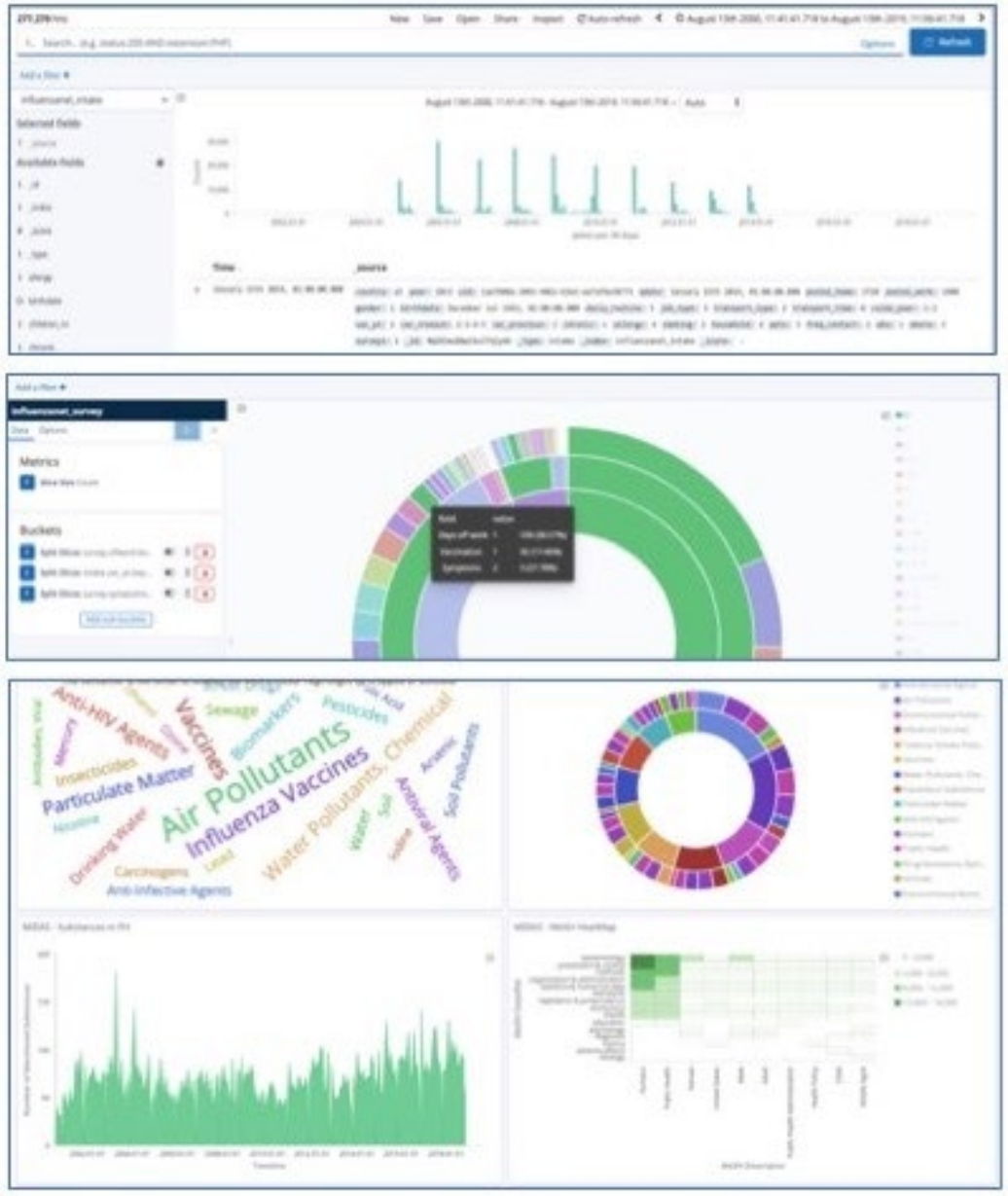


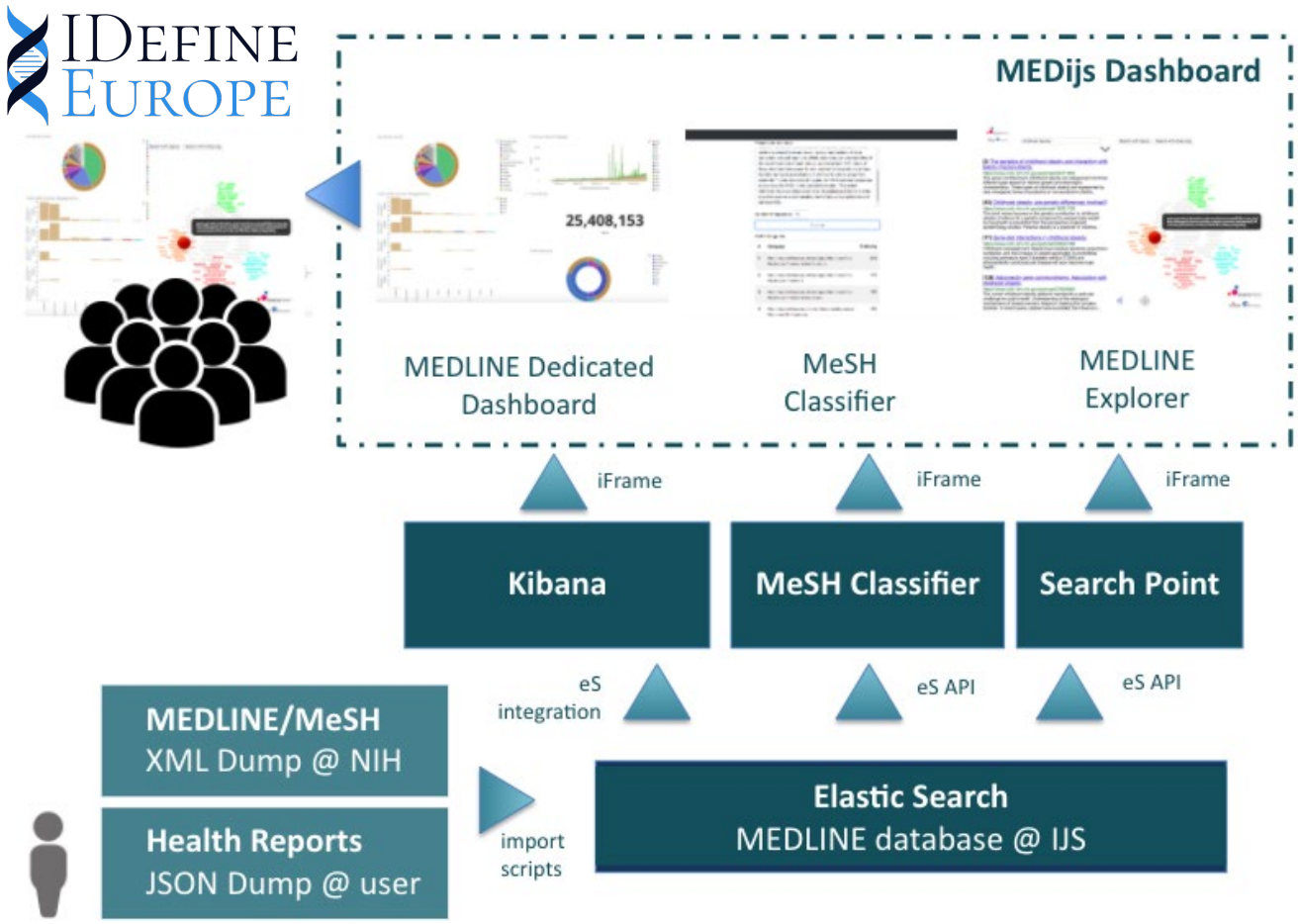




QUERYING THE WORLDWIDE NEWS BASED ON MED CATEGORIES







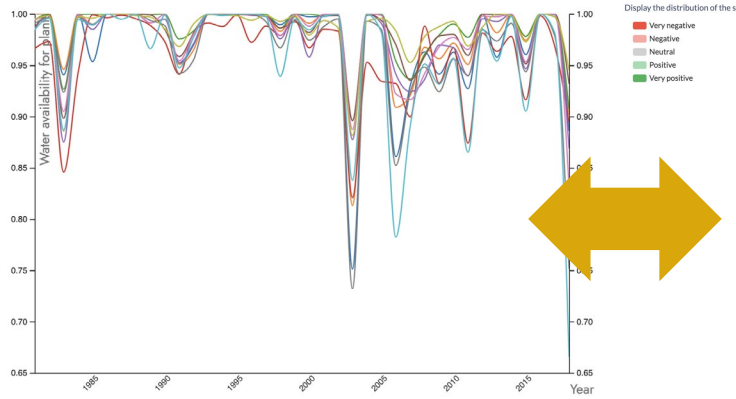
Pita Costa, J., Stopar, L., Rei, L., Massri, B. and Grobelnik, M., 2021. Exploring biomedical records through text mining-driven complex data visualisation. *medRxiv*, pp.2021-03.

EXPLORING RARE DISEASES ACROSS DIFFERENT PERSPECTIVES

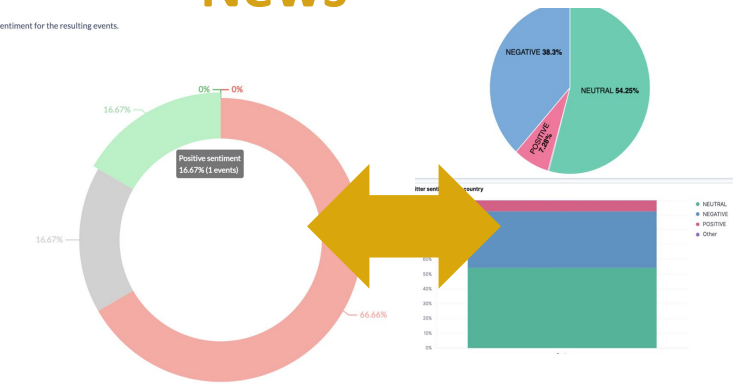
EXPLORING RARE DISEASES ACROSS DIFFERENT PERSPECTIVES

Support Community Priorities

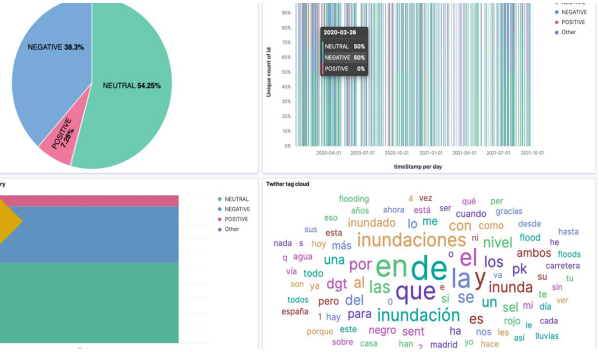
Indicators



News

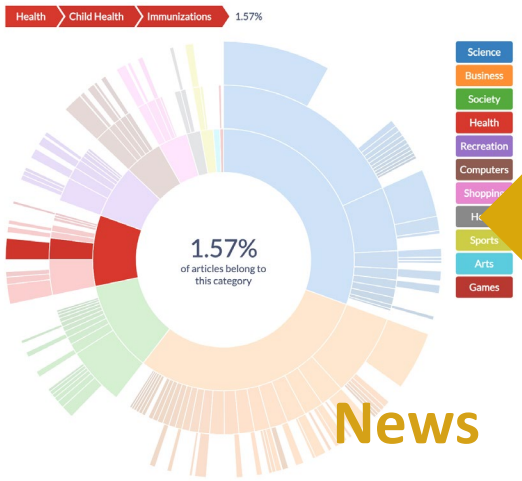


Social Media

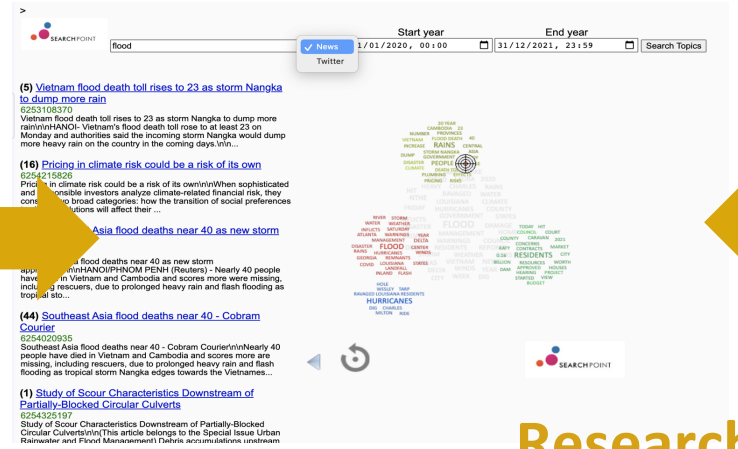


Explore Worldwide Practices

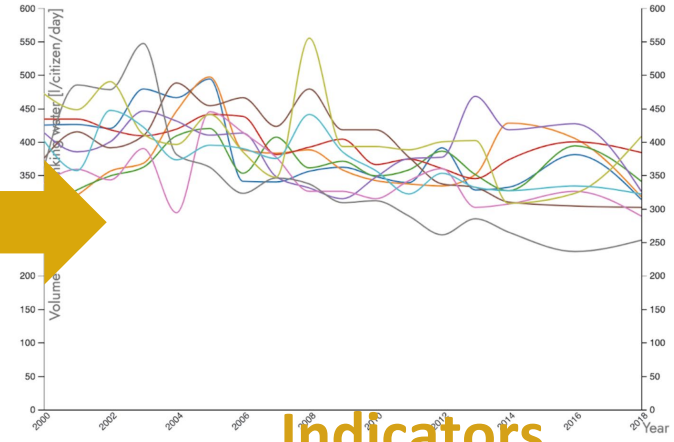
News



Research



Indicators



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Hope in the Dark: How AI is Unlocking New Possibilities for Rare Disease Patients

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learning in aiding in the analysis and understanding of rare diseases and their associated biomedical data.

Machine learning-based text mining technologies present a unique opportunity to overcome these obstacles by providing more comprehensive and accessible

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