



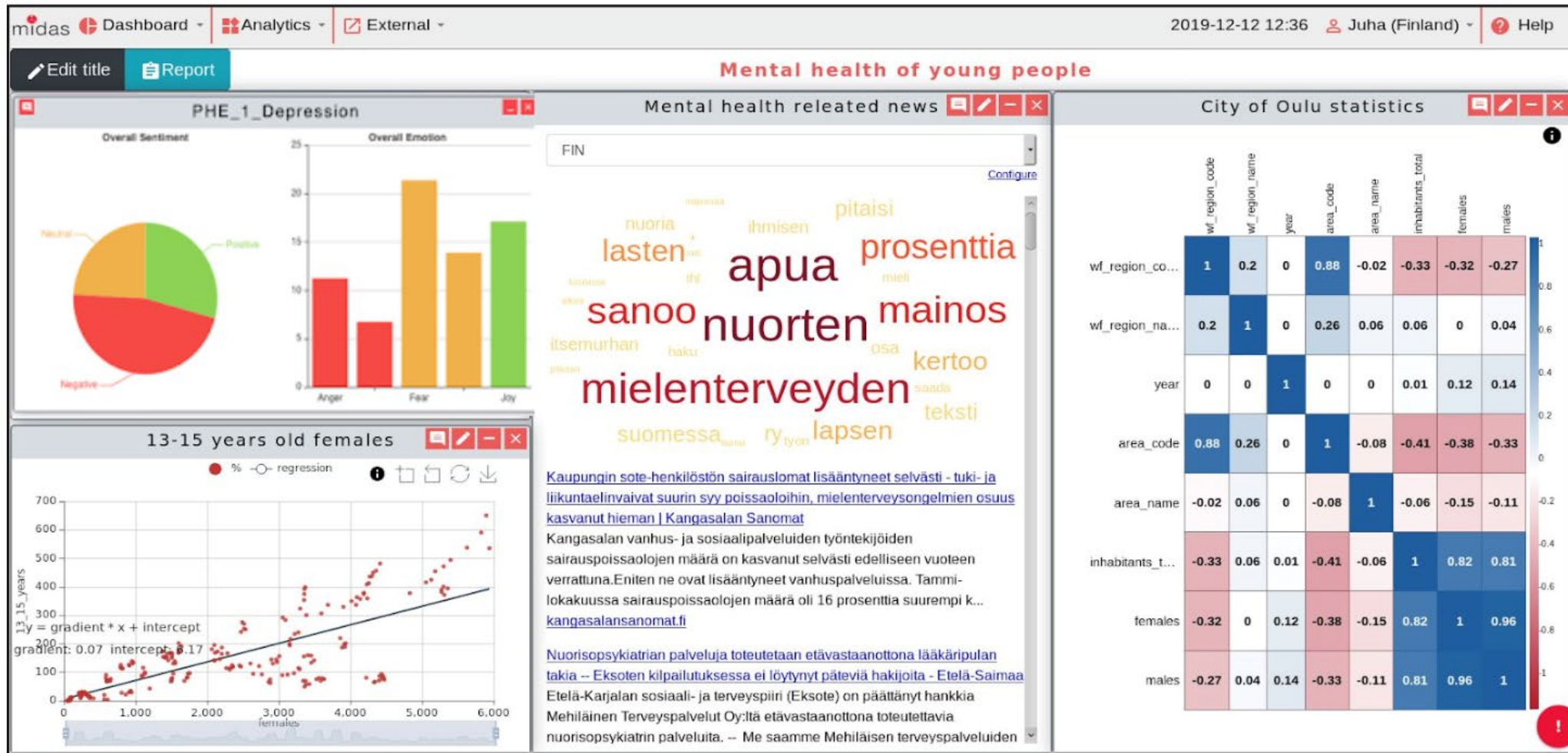
# IRCAI SDG Observatory

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

**Kleefstra Syndrome Scientific Conference 2023**

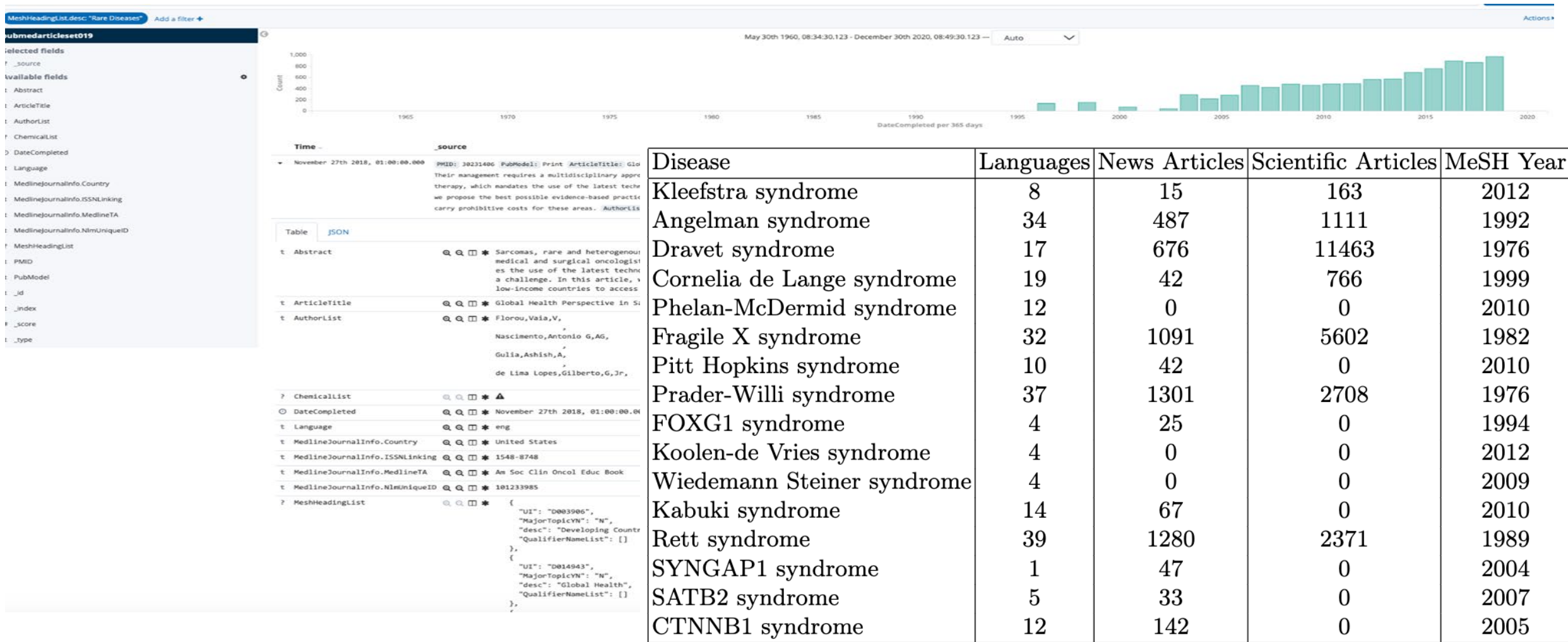
**Joao Pita Costa, IRCAI**

# LESSONS LEARNT FROM DATA - DRIVEN PUBLIC HEALTH



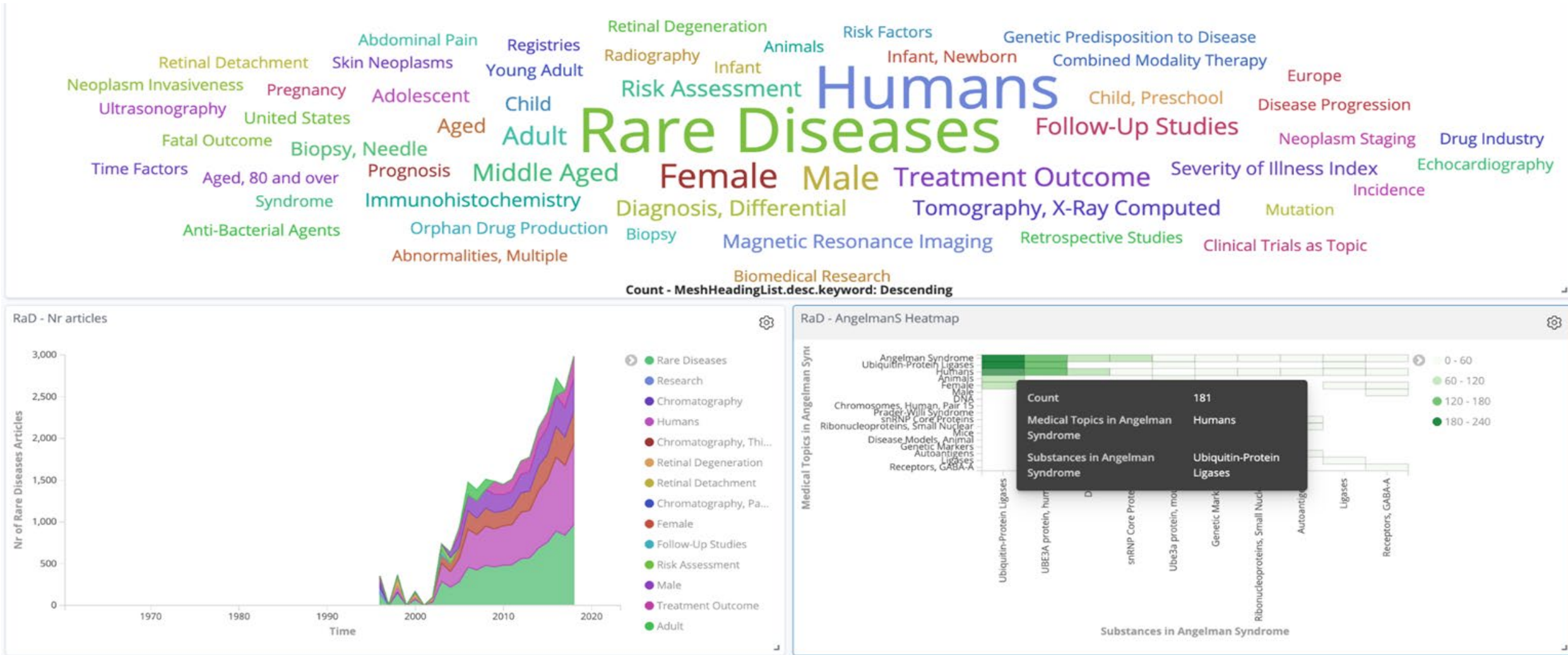
Pita Costa, Joao, Marko Grobelnik, Flavio Fuart, Luka Stopar, Gorka Epelde, Scott Fischaber, Piotr Poliwoda et al. "Meaningful big data integration for a global COVID-19 strategy." IEEE Computational Intelligence Magazine 15, no. 4 (2020): 51-61.

# DATA COVERAGE FOR RARE DISEASES



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., Paolotti, 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

The screenshot displays the Searchpoint interface for a literature review on 'Kleefstra Syndrome'. At the top, the search term 'Kleefstra Syndrome' is entered in a search bar, and a 'Search Topics' button is visible. Below the search bar, four search results are listed, each with a title, a URL, and a brief background description. A word cloud visualization is positioned in the center-right of the interface, showing terms related to the search results, with 'EHMT1' being the most prominent word. Navigation arrows are present at the bottom of the results list.

**(0) Severe neonatal presentation of Kleefstra syndrome in a patient with hypoplastic left heart syndrome and 9q34.3 microdeletion.**  
<https://www.ncbi.nlm.nih.gov/pubmed/25380126>  
BACKGROUND / BACKGROUND: Kleefstra syndrome arises from haploinsufficiency of EHMT1 caused by either microdeletions at 9q34.3 or intragenic mutations. Patients with Kleefstra syndrome have multisystem involvement including intellectual disability, hypoton...

**(1) TWINS WITH KLEEFSTRA SYNDROME DUE TO CHROMOSOME 9q34.3 MICRODELETION.**  
<https://www.ncbi.nlm.nih.gov/pubmed/26852514>  
Kleefstra or 9q subtelomeric deletion syndrome (9qSTS) is a rare microdeletion syndrome. The most prominent phenotypic features include hypotonia, developmental retardation, as well as typical dysmorphic face. It has been shown that terminal deletions of...

**(2) Intragenic duplication of EHMT1 gene results in Kleefstra syndrome.**  
<https://www.ncbi.nlm.nih.gov/pubmed/25348628>  
BACKGROUND / BACKGROUND: Kleefstra syndrome is characterized by intellectual disability, muscular hypotonia in childhood and typical facial features. It results from either a microdeletion of or a deleterious sequence variant in the gene euchromatic histo...

**(3) Kleefstra Syndrome: The First Case Report From Iran.**  
<https://www.ncbi.nlm.nih.gov/pubmed/29228531>  
Kleefstra Syndrome is characterized by severe mental retardation, brachycephaly, microcephaly, epileptic seizures, distinct facial features, and infantile weak muscle tone and heart defects. Deletion of EHMT1 is the main player in 75% of cases. Because of...

**(4) Multiple Coronary Artery Microfistulas in a Girl with Kleefstra Syndrome.**  
<https://www.ncbi.nlm.nih.gov/pubmed/27239352>

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

Pita Costa, J., Fuat, F., Stopar, L., Grobelnik, M., Mladenec, 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)

# AUTOMATED CLASSIFICATION BASED ON PUBMED CATEGORIES

The screenshot shows the MeSH website interface. At the top, there is a navigation bar with the NIH logo and the text "National Library of Medicine". Below this, there is a search bar and several menu items: "Search", "Tree View", "MeSH on Demand", "MeSH 2022", "About", "Suggestions", "Contact Us", and a "Survey" button. The main content area is titled "Angelman Syndrome MeSH Descriptor Data 2023". Below the title, there are four tabs: "Details", "Qualifiers", "MeSH Tree Structures", and "Concepts". The "Details" tab is selected, showing the following information:

<b>MeSH Heading Tree Number(s)</b>	Angelman Syndrome C10.228.662.075 C16.131.077.095 C16.131.260.040 C16.320.180.040
<b>Unique ID</b>	D017204
<b>RDF Unique Identifier</b>	<a href="http://id.nlm.nih.gov/mesh/D017204">http://id.nlm.nih.gov/mesh/D017204</a>
<b>Scope Note</b>	A syndrome characterized by multiple abnormalities, <b>MENTAL RETARDATION</b> , and movement disorders. Present usually are skull and other abnormalities, frequent infantile spasms ( <b>SPASMS, INFANTILE</b> ); easily provoked and prolonged paroxysms of laughter (hence "happy"); jerky puppetlike movements (hence "puppet"); continuous tongue protrusion; motor retardation; <b>ATAXIA</b> ; <b>MUSCLE HYPOTONIA</b> ; and a peculiar facies. It is associated with maternal deletions of chromosome 15q11-13 and other genetic abnormalities. (From Am J Med Genet 1998 Dec 4;80(4):385-90; Hum Mol Genet 1999 Jan;8(1):129-35)
<b>Entry Term(s)</b>	Happy Puppet Syndrome Puppet Children
<b>Previous Indexing</b>	Ataxia (1972-1992) Mental Disorders (1957-1992) Movement Disorders (1957-1992)
<b>See Also</b>	<a href="#">Intellectual Disability</a>
<b>Public MeSH Note</b>	93

At the bottom of the page, there is a "RaD1.gif" logo on the left and a "Show All" button on the right.

<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.

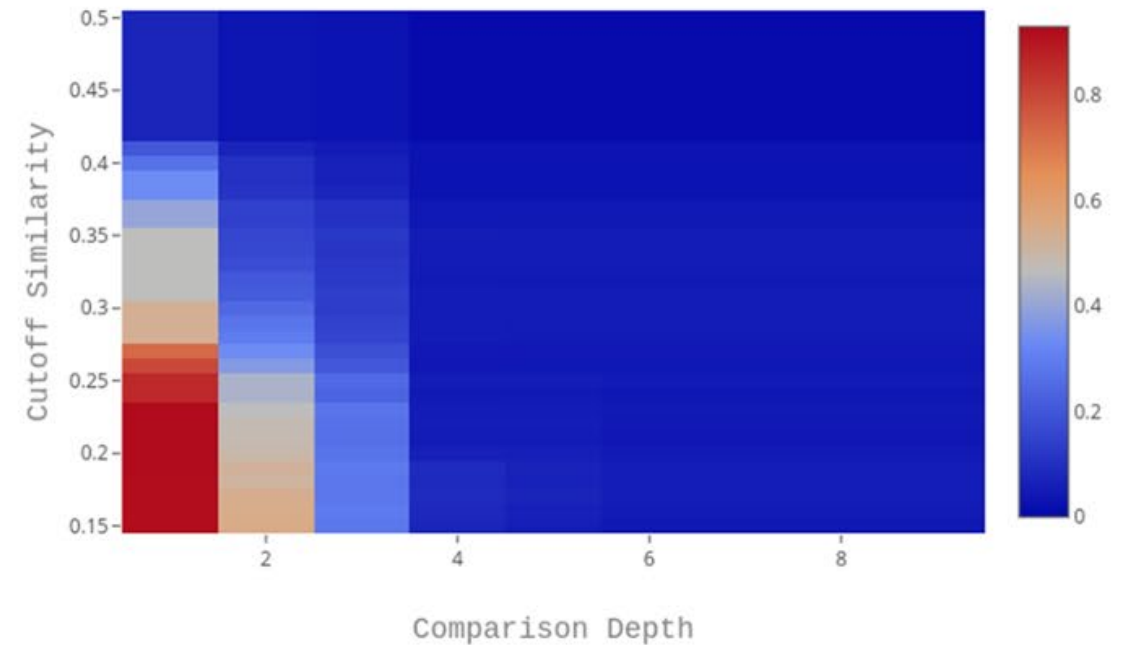
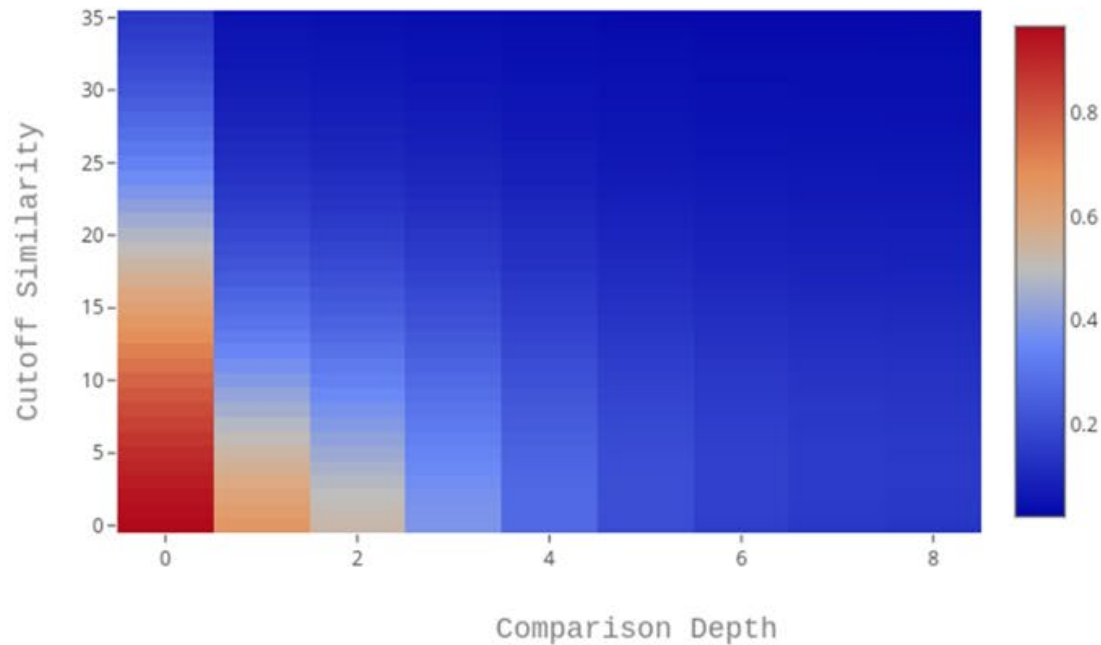
# EVALUATING THE QUALITY OF THE AUTOMATED CLASSIFICATION

## Angelman Syndrome MeSH Descriptor Data 2023

Details Qualifiers MeSH Tree Structures Concepts

**MeSH Heading** Angelman Syndrome  
**Tree Number(s)** C10.228.662.075  
 C16.131.077.095  
 C16.131.260.040  
 C16.320.180.040  
**Unique ID** D017204  
**RDF Unique Identifier** <http://id.nlm.nih.gov/mesh/D017204>  
**Scope Note** A syndrome characterized by multiple abnormalities, **MENTAL RETARDATION**, and movement disorders. Present usually are skull and other abnormalities, frequent infantile spasms (**SPASMS, INFANTILE**); easily provoked and prolonged paroxysms of laughter (hence "happy"); jerky puppetlike movements (hence "puppet"); continuous tongue protrusion; motor retardation; **ATAXIA**; **MUSCLE HYPOTONIA**; and a peculiar facies. It is associated with maternal deletions of chromosome 15q11-13 and other genetic abnormalities. (From Am J Med Genet 1998 Dec 4;80(4):385-90; Hum Mol Genet 1999 Jan;8(1):129-35)

The screenshot shows the 'eventregistry' website interface. At the top, there's a search bar with 'Angelman syndrome' entered. Below the search bar is a map of Europe. A popup window is open over France, displaying the text: 'Sainte-Suzanne : c'est parti pour l'édition 2023 des fêtes!' with source 'SudOuest.fr' and location 'Bordeaux, France'. To the right of the map, there are two tabs: 'ARTICLES' and 'EVENTS'. Under 'ARTICLES', several news snippets are visible, including 'PTC Therapeutics Stock Tumbles: Company Stops Preclinical Gene Therapy Programs After Failed Friedreich's Ataxia Study' and 'Amplifying gene expression with RNA-targeted therapeutics - Nature Reviews Drug Discovery'.



# QUERYING THE WORLDWIDE NEWS BASED ON MED CATEGORIES



SEARCH

FILTERS: Locations Sources Organisms → Viruses → RNA Viruses Time of interest

**Categories**  
 Limit the news articles to only those that are on a particular topic (or any of the subtopics). [Learn more.](#)

Pick **Organisms → Viruses → RNA Viruses** Category name

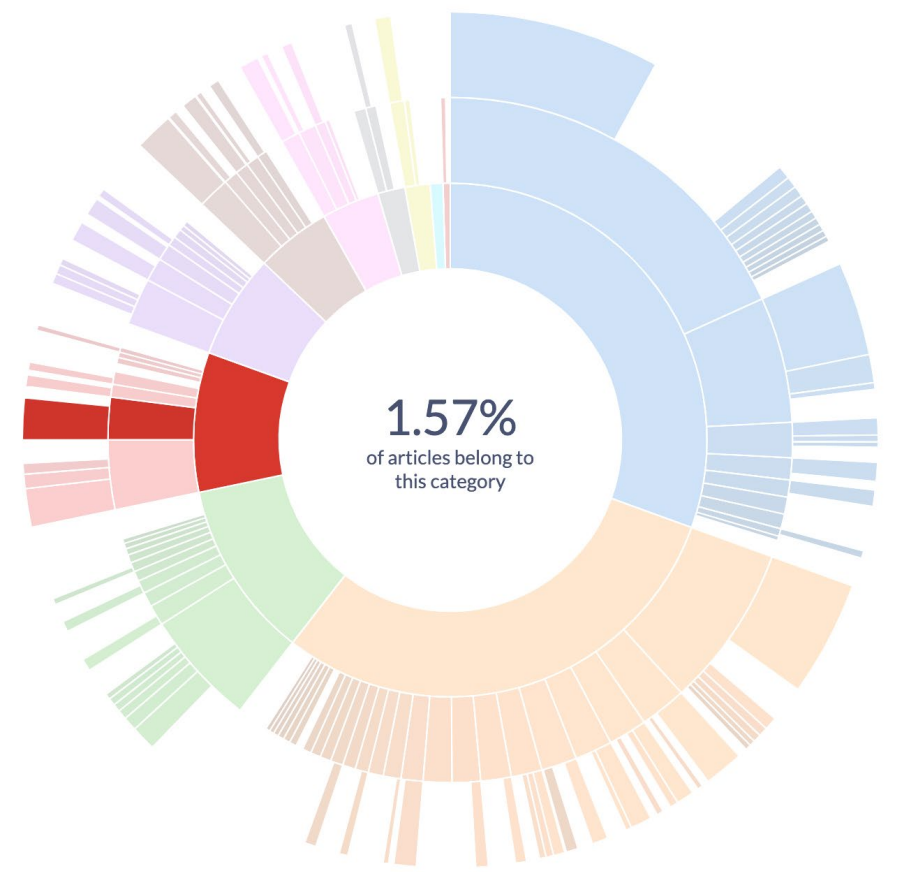
- Analytical, Diagnostic and Therapeutic Techniques and Equipment
- Anatomy
- Chemicals and Drugs
- Disciplines and Occupations
- Diseases
- Geographicals
- Health Care
- Humanities
- Information Science
- Named Groups
- Organisms
- Phenomena and Processes
- Psychiatry and Psychology
- Technology, Industry, Agriculture

Bat Virus Behind The Death of 100 Asian Scientist (Apr. 12, 2018) - piglets in China between 2016 In October 2016, pig farms in t

- Bone Diseases
- Cartilage Diseases
- Fasciitis
- Foot Deformities
- Foot Diseases
- Hand Deformities
- Jaw Diseases
- Joint Diseases
- Muscular Diseases

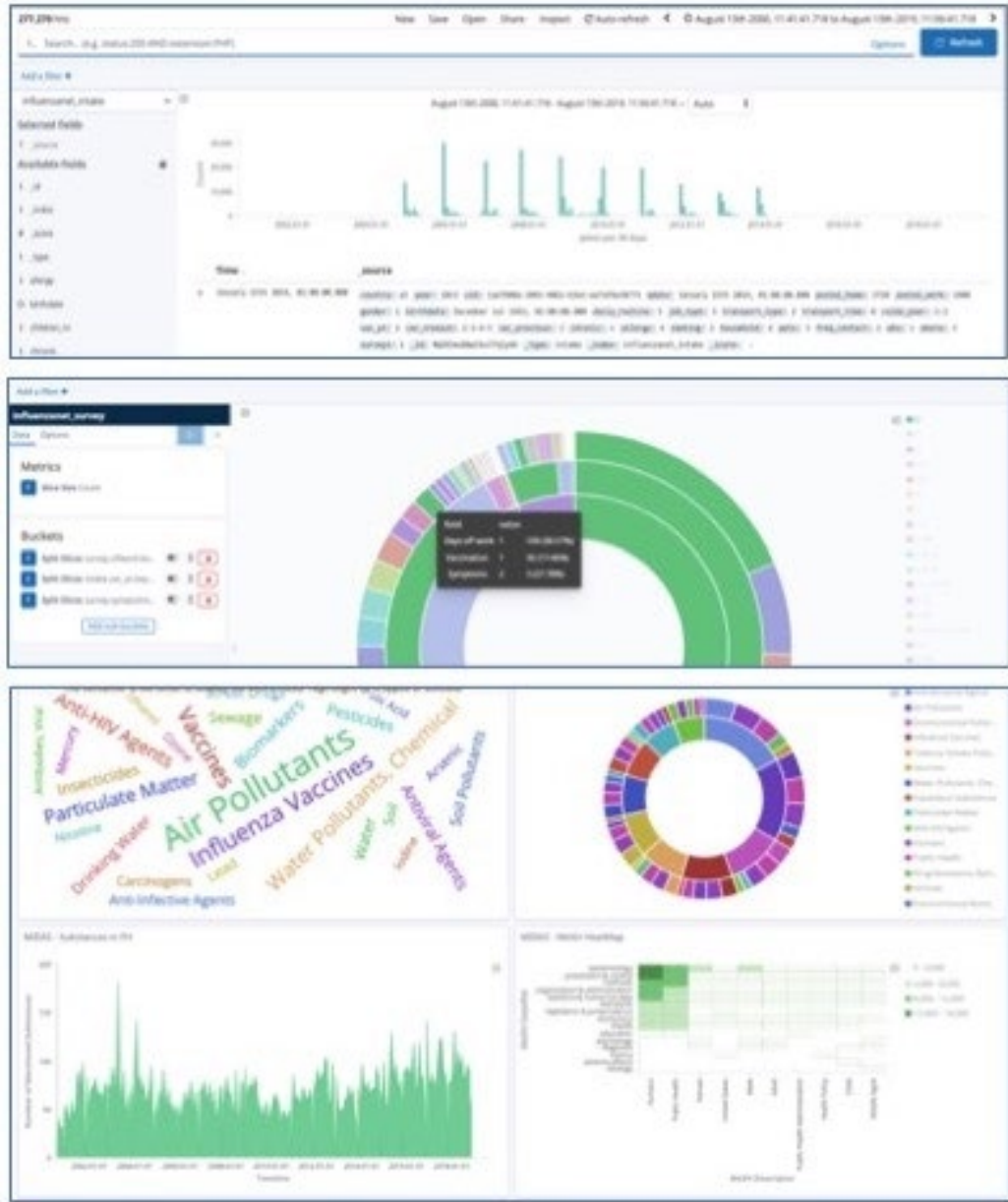
- RS-CoV), Lassa virus and Nipah virus
- ing diseases before they become global
- Animal Diseases
- Bacterial Infections and Mycoses
- Cardiovascular Diseases
- Chemically-Induced Disorders
- Congenital, Hereditary, and Neonatal Diseases a
- Digestive System Diseases
- Disorders of Environmental Origin
- Endocrine System Diseases
- Eye Diseases
- Female Urogenital Diseases and Pregnancy Com
- Hemic and Lymphatic Diseases
- Immune System Diseases
- Male Urogenital Diseases
- Musculoskeletal Diseases
- Neoplasms
- Nervous System Diseases
- Nutritional and Metabolic Diseases
- Occupational Diseases
- Otorhinolaryngologic Diseases
- Parasitic Diseases
- Pathological Conditions, Signs and Symptoms
- Respiratory Tract Diseases

Health > Child Health > Immunizations 1.57%



- Science
- Business
- Society
- Health
- Recreation
- Computers
- Shopping
- Home
- Sports
- Arts
- Games





MEDLINE/MeSH XML Dump @ NIH  
 Health Reports JSON Dump @ user



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



## Education

Architecture <sup>(4)</sup> Arts <sup>(42)</sup> Astronomy <sup>(4)</sup> Biology <sup>(12)</sup> Business <sup>(4)</sup> Chemistry <sup>(2)</sup>  
Computers <sup>(2)</sup> Computer Science <sup>(2)</sup> Data Science <sup>(1)</sup>  
Earth Sciences <sup>(4)</sup> Environment <sup>(2)</sup> Events <sup>(2)</sup> Health Sciences <sup>(7)</sup> Humanities <sup>(7)</sup> Life Sciences <sup>(5)</sup>  
Mathematics <sup>(2)</sup> Medicine <sup>(2)</sup> Military <sup>(2)</sup> Philosophy <sup>(1)</sup> Physics <sup>(4)</sup> Psychology <sup>(2)</sup>  
Regional <sup>(2)</sup> Science <sup>(2)</sup> Social Sciences <sup>(2)</sup> Sports <sup>(4)</sup> Technology <sup>(2)</sup>

20056 Items Sorted... Page 1 of 549 > Next < Prev

Preglovi kolokvij  
Drogovi kolokvij na Kemijskem inštitutu Preglovi kolokvij je potrditev po Frederiku Fribzu Preglovi kolokvij in znanosti slovenskega rodu, ki je...



## Legislation & Reg

Start year: 1/1/2019, 01/01 End year: 31/12/2019, 21/12

(3) Vietnam food death toll rises to 23 as storm Nangka looms closer

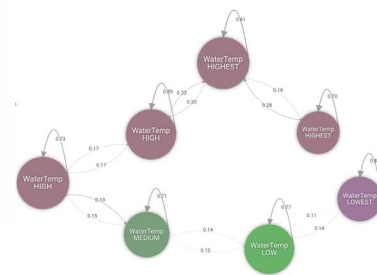
(4) Southeast Asia food deaths near 40 as new storm looms closer

(4) Southeast Asia food deaths near 40 - Cobram Courier

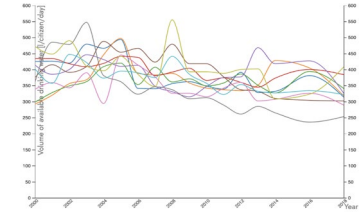
(5) Study of Blood Chimeras Reveals Downstream of Parvovirus Genetic Control



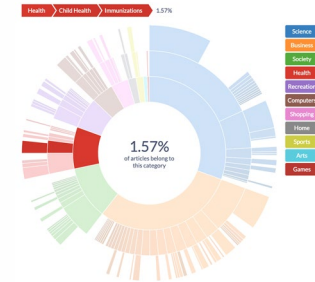
## Citizen Science



## Indicators



## News



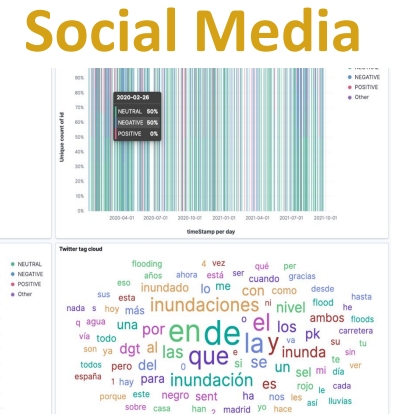
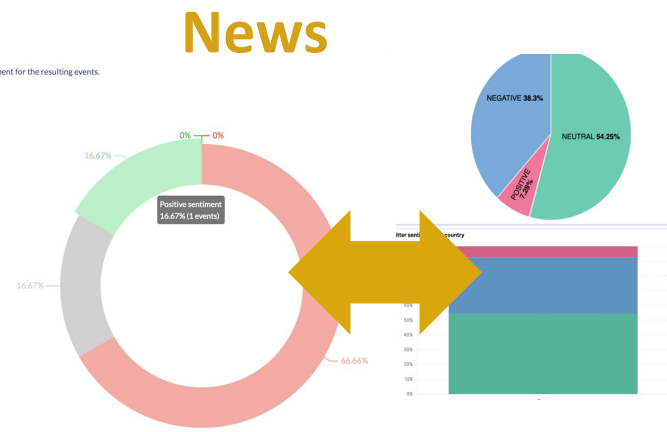
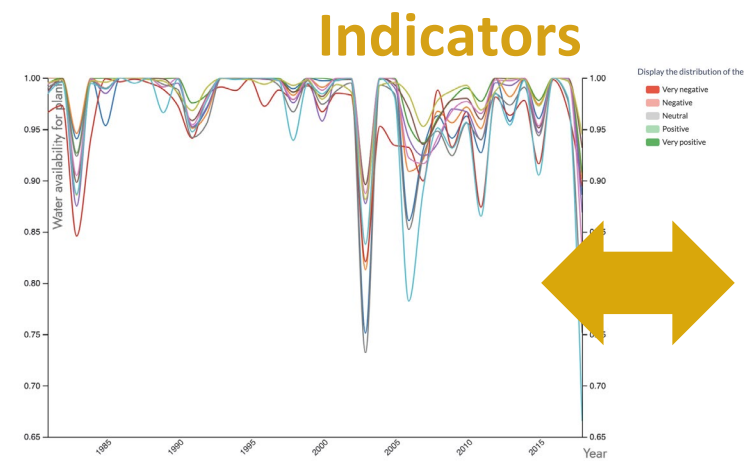
## Social Media

## Science & Patents

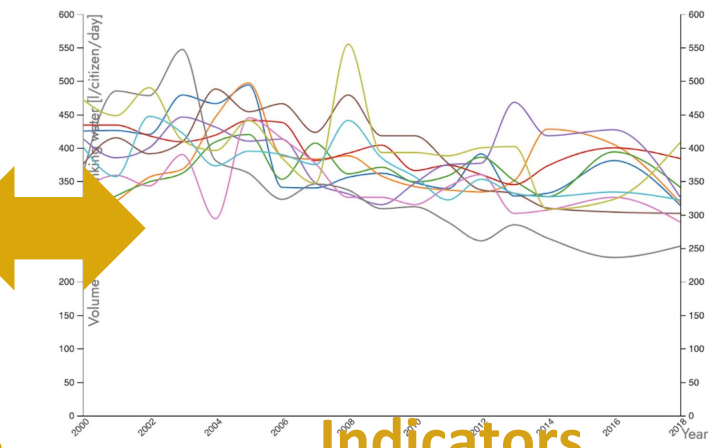
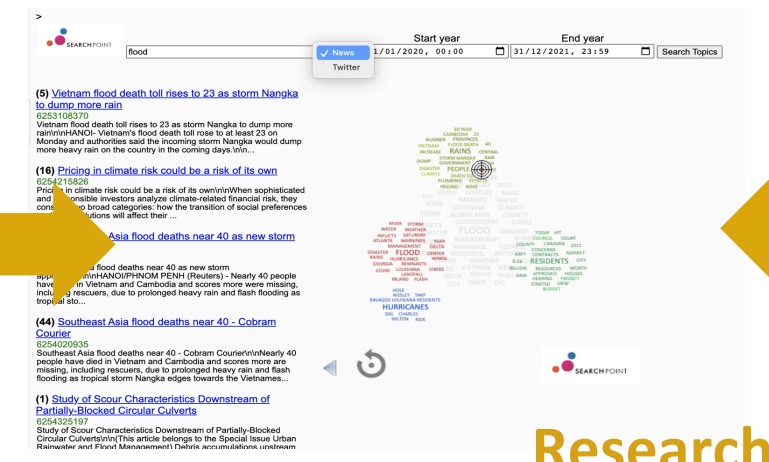
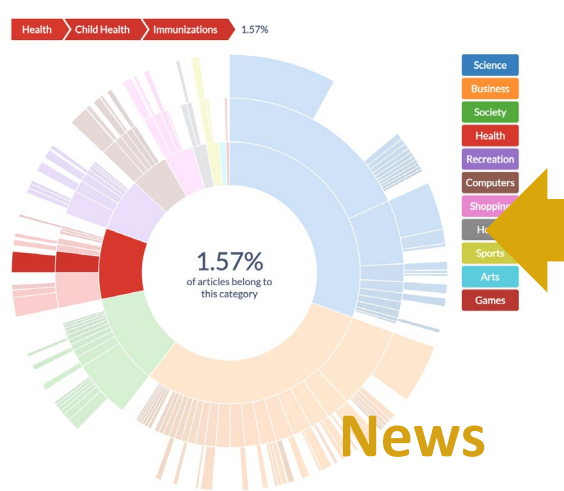


# EXPLORING RARE DISEASES ACROSS DIFFERENT PERSPECTIVES

Support  
Community  
Priorities



Explore  
Worldwide  
Practices



Research

< Back

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

SWForum.eu / Online SW Forum / Software Technology /

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

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

Recent comments

<https://swforum.eu/online-sw-forum/software-technology/6/hope-dark-how-ai-unlocking-new-possibilities-rare-disease>



**International Research Centre on Artificial Intelligence  
under the auspices of UNESCO (Category II)**  
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E: [info@ircai.org](mailto:info@ircai.org) | W: <https://ircai.org/>

*With the Support from the African partners in our NAIXUS Network*

*Laboratory at the University of the Witwatersrand, Masakhane  
Foundation, Tanzania AI Lab & Community, Research ICT  
Africa, University of Cape Coast, University of Pretoria,  
Kabarak University*