

What is Netherton Syndrome?

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Outline

- Motivation
- About Netherton syndrome
- Text mining approach
- Results

What is Netherton syndrome?

- Rare hereditary disorder of skin
- Autosomal recessive trait
- Signs and symptoms
 - Dry, red, scaling of skin, hair anomalies (“bamboo hair”)
- Misdiagnosed as atopic dermatitis or psoriasis



Image source: <https://hotlifestylenews.com/world-news/boy-10-who-looks-scalded-has-to-carry-cards-explaining-his-rare-condition-to-stop-cruel-taunts-from-strangers/>

More on Netherton syndrome

- Age of onset: Neonatal or Infancy
- Netherton syndrome is estimated to affect 1 in 200,000 newborns
- High post-natal lethality
- Complications: Dehydration, recurrent infections

- Cause

- Mutation of SPINK5 gene (chromosome 5)
- Encodes for proteases -> Increased protease action -> too few layers of the outer skin

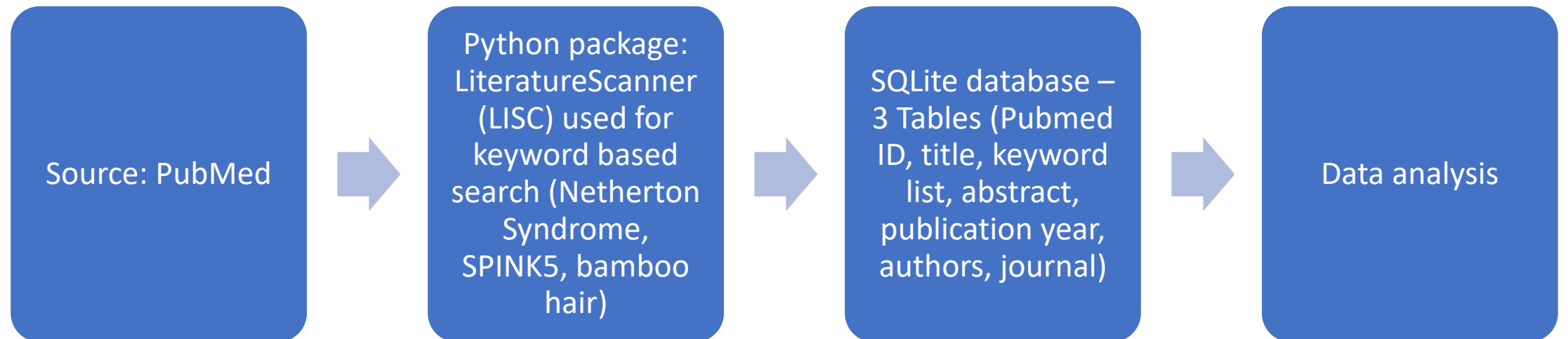
- Remedy

- Application of lotions, moisturizing creams (with care – sepsis), oral anti-histamines, oral and topical steroids for anti-inflammation

Text mining approach

- PubMed literature survey
 - When were they published?
 - What did it include?
 - Which are the countries contributing to this field?

Process



LISC Author: Thomas Donoghue

Data analysis approach

- Count Frequency metrics
 - Year of publication
 - Labs involved
- Association of terms
- Word frequency analysis
 - Keywords

Results

1) Search results

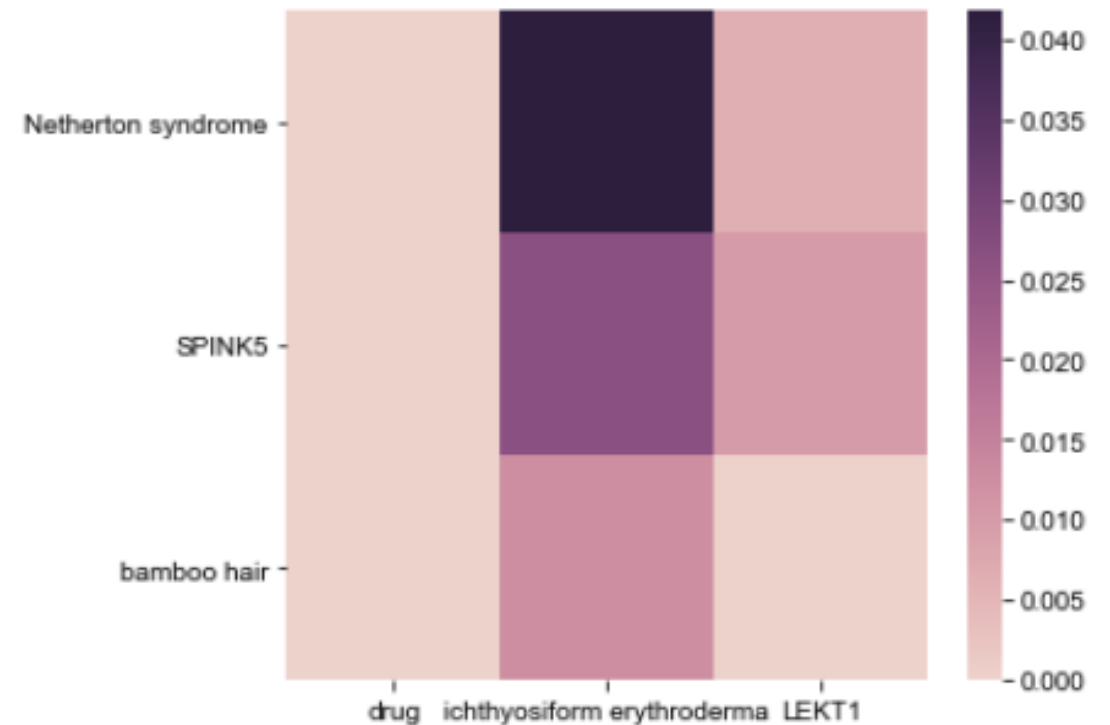
	Netherton Syndrome	SPINK5	Bamboo hair
No. of articles	311	198	24

Threshold for articles: 1500

2) Co-occurrence of terms in A and B (Jaccard similarity index)

Terms A=Netherton syndrome, SPINK5,
bamboo hair

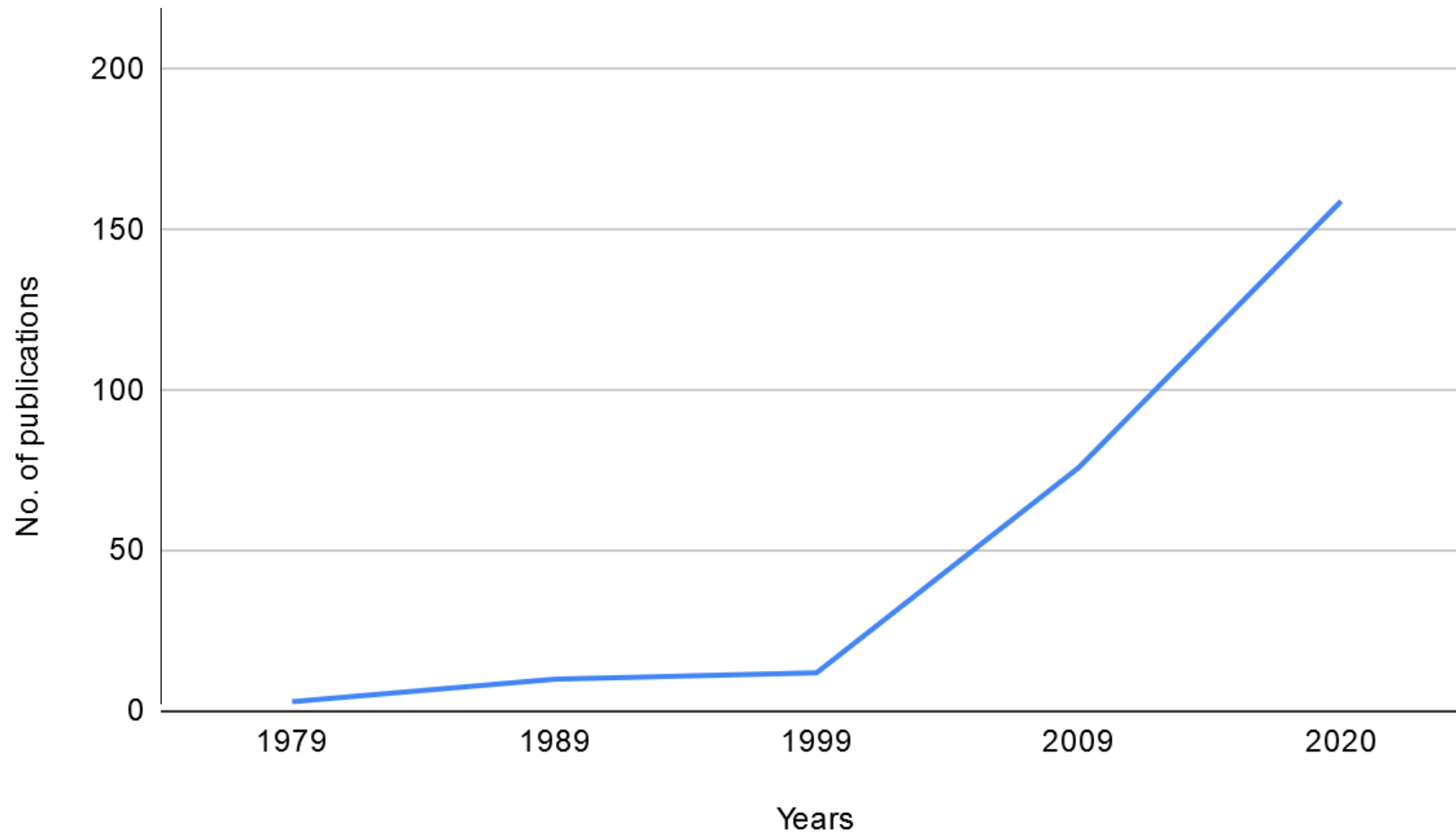
Terms B = drug, ichthyosiform
erythroderma, LEKT1



Snapshot of database

File Edit View Tools Help							
New Database Open Database Write Changes Revert Changes Open Project Save Project Attach Database Close Database							
Database Structure Browse Data Edit Pragmas Execute SQL							
Table: ns_pubmed_articles New Record Delete Record							
	pubmed_id	title	pubmed_keyword_list	abstract	author	journal	publication
	Filter	Filter	Filter	Filter	Filter	Filter	Filter
1	32347581	The matriptase-prostasin proteolytic cascade in dermatologic diseases.	['Ichthyosis', 'Netherton syndrome', 'epidermis', 'genetics', 'kallikrein']	normal regulation of desquamation. An increasing amount of research describing this pathway has recently become available, and the importance of this pathway is exhibited by the association of genetic defects in this pathway with human diseases of the skin and hair. Given the relevan...	Institute of Molecular Medicine, Thomas Jefferson University, Philadelphia, PA, USA.], ['Niaziorimi', 'Fateme', 'F', 'Department of Dermatology and Cutaneou...	['Experimental dermatology', 'Exp. Dermatol.']	2020
2	32265319	Skin barrier lipid enzyme activity in Netherton patients is associated with protease activity and ceramide abnormalities.	['Activity Based Probe labeling', 'Ceramides', 'Enzyme Expression', 'Fluorescence microscopy', 'Ichthyosis linearis circumflexa (Netherton Syndrome)', 'In situ Zymography', 'Mass spectrometry', 'Skin', 'Sphingolipids']	expression and activities with i) stratum corneum ceramide profiles, ii) in situ serine protease activity, and iii) clinical presentation of patients. Using activity-based probe labeling, we visualized and localized active, epidermal GBA, and a newly developed in situ zymography method enabled us to ...	['Stephens', 'Nicole', 'N', 'Leiden Academic Centre for Drug Research, Netherlands.'], ['Absalah', 'Samira', 'S', 'Leiden Academic Centre for Drug Research, ...	['Journal of lipid research', 'J. Lipid Res.']	2020

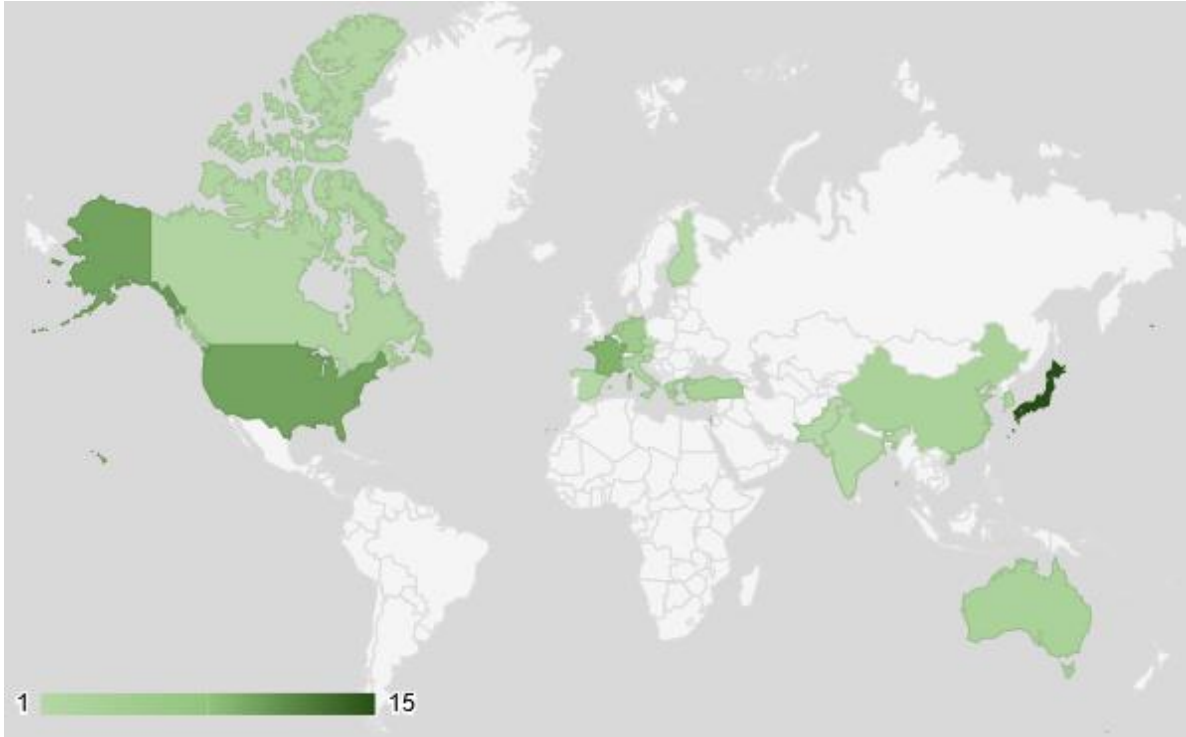
Number of 'Netherton syndrome' publications over the years



Word cloud of publications in year 2020
(Netherton syndrome and SPINK5)



Mapping of countries with publications



- Data based on last author of publications affiliated to country
- Colour intensity based on number of publications

Conclusion

Q1) When were they published?

- Netherton syndrome
 - Spike in publications from 1999
 - Maximum in 2020

Q2) What did it include?

- Latest research articles discuss about molecular level information (Kallikrein related peptidases, MALT1)

Q3) Which are the countries contributing to this field?

- Japan, USA, France, Germany top contributing countries

Thank you 😊