Finding Rare Disease Experts for the Undiagnosed Disease Network

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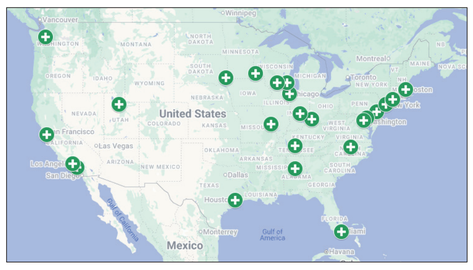
Abstract

The NIH-funded Undiagnosed Disease Network (UDN) brings together experts from healthcare organizations across the United States to provide answers for patients and families affected by mysterious conditions where clinicians have been unable to discover the cause. We used "Profiles", an open-source system for generating searchable online profiles of investigators, to create a website to help the UDN find these experts. Profiles imports and links data from PubMed, ClinicalTrials.gov, GeneReports, OMIM, and more.

Undiagnosed Disease Network

https://undiagnosed.hms.harvard.edu

The NIH-funded Undiagnosed Disease Network (UDN) helps provide answers for patients and families affected by mysterious conditions where clinicians have been unable to discover the cause. The UDN brings together clinicians and researchers from 20+ healthcare organizations across the United States. To date, the UDN has evaluated 3,094 participants (61% under age 18) and diagnosed 30% of them. A challenge for the UDN is finding and recruiting experts in rare diseases and genomics who can evaluate new participants.

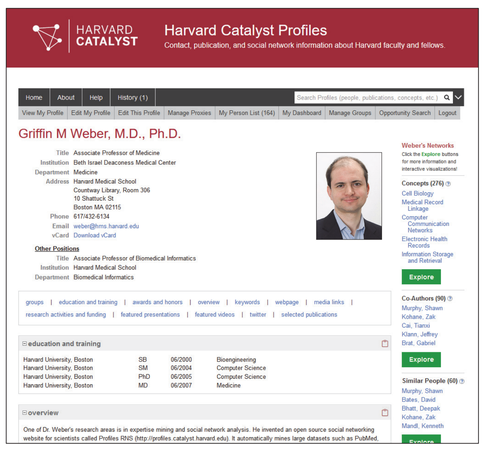


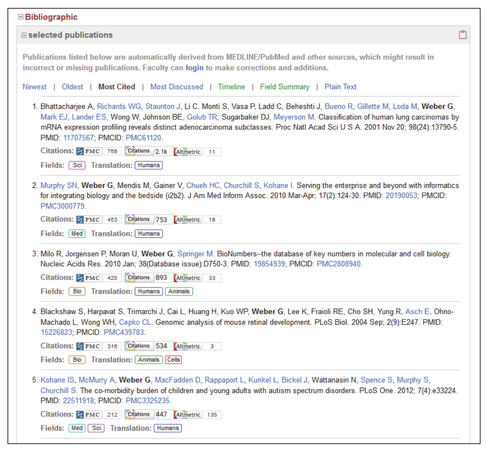
Profiles Research Networking Software

Team Science Tool for Finding Experts

Profiles Research Networking Software (RNS) is a free and open-source web-based expertise search and collaboration tool used at Harvard and other organizations. It creates online profiles of an organization's investigators by linking internal data from administrative systems with external data sources of publications (e.g., PubMed), grants, clinical trials, and more. Various bibliometric measures, such as article citation count, Altmetric score, fields, and translation are displayed on the website.

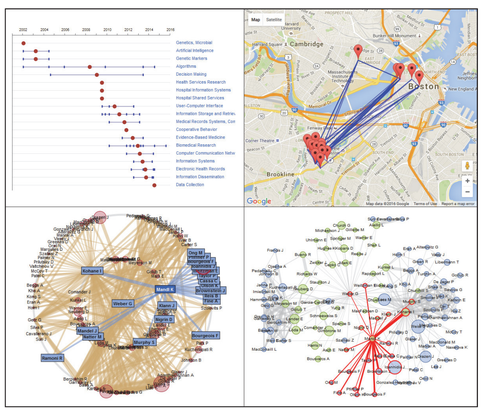
https://profiles.catalyst.harvard.edu

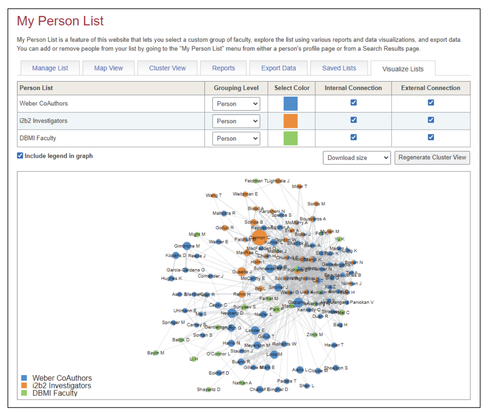




Network and Team Analysis in Profiles RNS

Data mining algorithms analyze the profiles to discover connections between investigators, such as previously co-authoring a publication, doing similar research, or having nearby offices. Data visualizations display these connections as interactive network graphs and social network analysis metrics are calculated by the software. The "My Person List" feature enables users to define arbitrary teams of people, generate reports about these teams, and visualize their overlap.



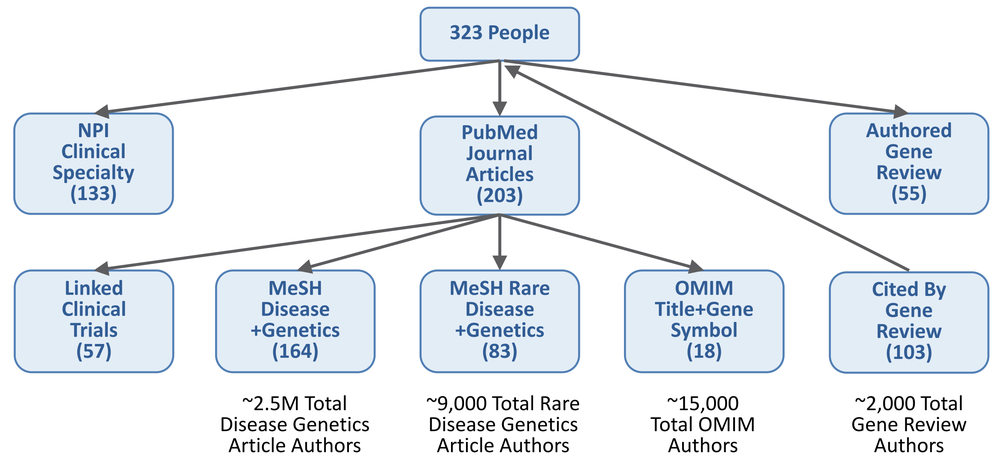


Methods

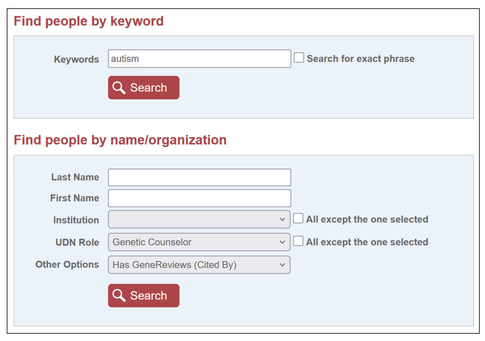
We created a private Profiles website for UDN staff to find relevant experts. The UDN provided an initial list of 323 clinicians and researchers from UDN sites who volunteered to join the network. We searched PubMed, GeneReviews (a book series about genetic conditions), and the Online Mendelian Inheritance in Man (OMIM) website for additional potential experts. People were linked to the National Provider Identifier (NPI) database to obtain information about their clinical expertise.

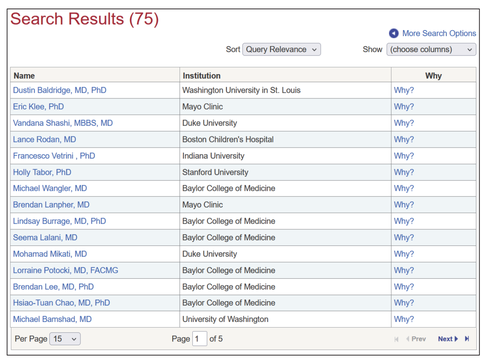
Results

The UDN Profiles website contains searchable profile pages of the initial 323 people. Profiles automatically found 12,487 journal articles authored by 203 of the individuals. These publications were associated with clinical trials for 57 people, MeSH terms related to disease genetics (164 people) and rare diseases (83 people). We extended Profiles to import OMIM records and GeneReview articles. We found 133 of the people in the NPI database to retrieve their clinical specialties. To find other experts that we could potentially add to the website in the future, we separately searched all of PubMed (9,000 rare disease genetics authors), OMIM (15,000 authors), and GeneReview (2,000 authors).



Search for UDN Experts





UDN Expertise Report

NPI Specialties
[NPI:1144204868] Medical Genetics - Clinical Genetics

Authored GeneReviews
[PMID:29369572] TANGO2 Deficiency
[PMID:39527683] Shashi-Pena Syndrome

GeneReviews That Cite This Person
[PMID:20301347] Congenital Myasthenic Syndromes Overview
[PMID:20301618] SALL1-Related Townes-Brocks Syndrome
[PMID:25275207] Cantú Syndrome
[PMID:25719193] 16p12.2 Recurrent Deletion
[PMID:29851452] GRIN2B-Related Neurodevelopmental Disorder

OMIM (Linked to Publications)
[OMIM:602568] METHIONINE SYNTHASE REDUCTASE; MTRR

Clinical Trials (Linked to Publications)
[NCT03721367] Chronic Liver Disease in Urea Cycle Disorders

Publication MeSH Terms with "Genetics" Qualifier
22q11 Deletion Syndrome; Anodontia; Anxiety; Anxiety Disorders; Arthrogryposis; Attention Deficit Disorder with Hyperactivity; Autism Spectrum Disorder; Autistic Disorder; Blepharoptosis

(This is an example report created by merging items from several people.)

References

1. Ramoni RB, et al. The Undiagnosed Diseases Network: Accelerating Discovery about Health and Disease. Am J Hum Genet. 2017 Feb 2;100(2):185-192.
2. Weber, GM, Yuan LA. The Power of Research Networking Systems to Find Experts and Facilitate Collaboration. Book chapter in *Strategies for Team Science Success* (pp. 541-562). 2019. Springer International Publishing.

Conclusions

The UDN Profiles site provides UDN staff a mechanism of easily searching for clinicians and researchers who can help evaluate new participant cases. We had to import several new types of data to capture expertise in rare diseases and genetics. We plan to add this new functionality to the Profiles open-source software.

For more information about this project contact **weber@hms.harvard.edu**.