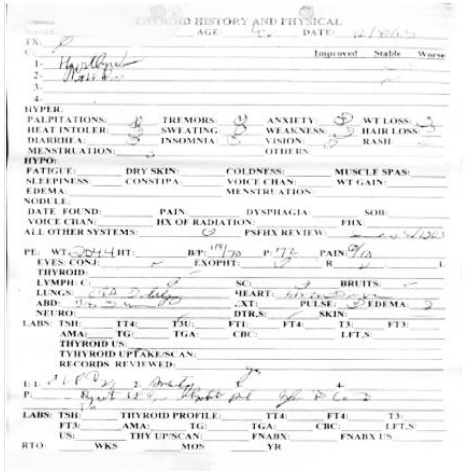
Building tools to empower patient experts

Orion Buske
@orionbuske

Structured data data

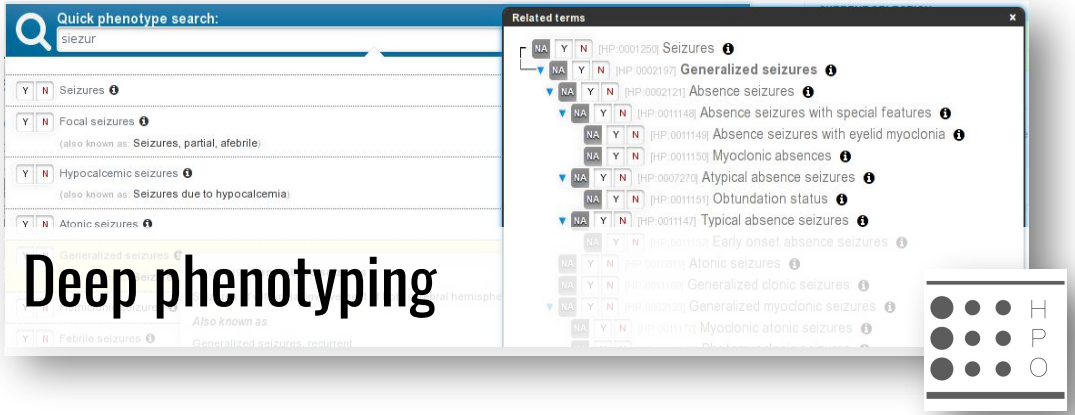


ataxia, developmental delay

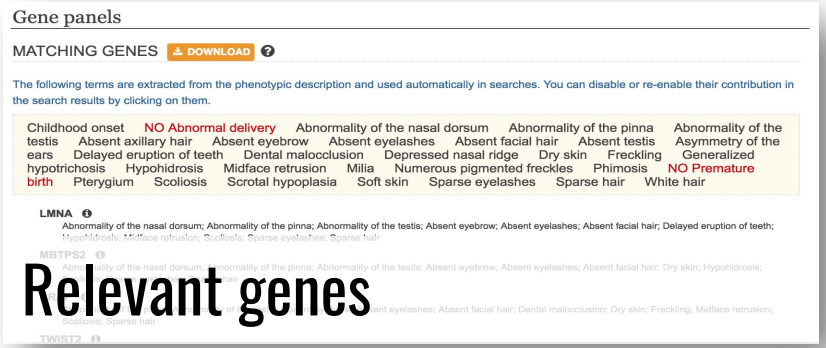
HP:0001251
HP:0001263

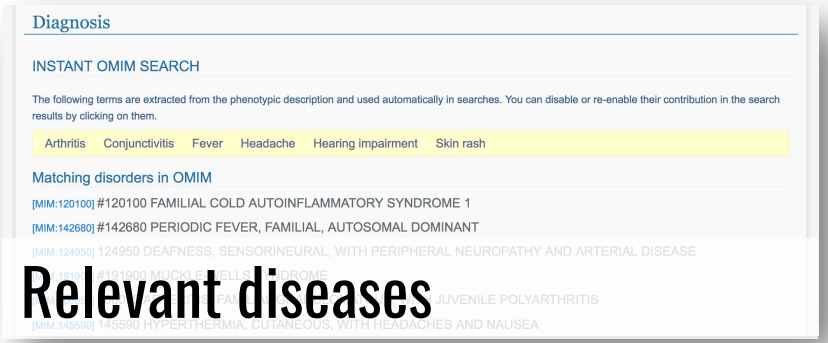
faster (sometimes) → **better**

Tools for doctors and researchers

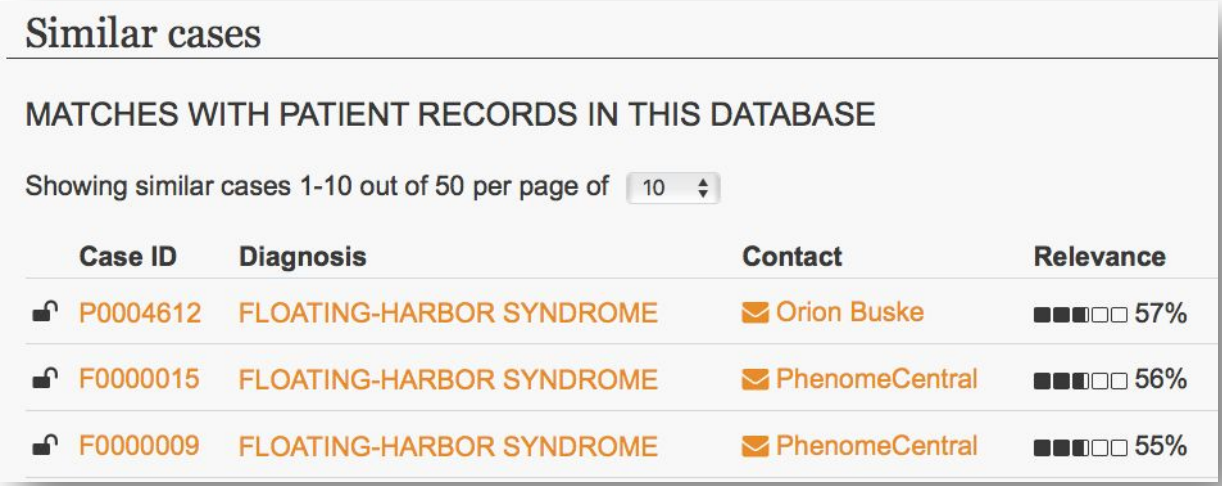


PHENOTIPS®





Finding similar cases



Similar cases

MATCHES WITH PATIENT RECORDS IN THIS DATABASE

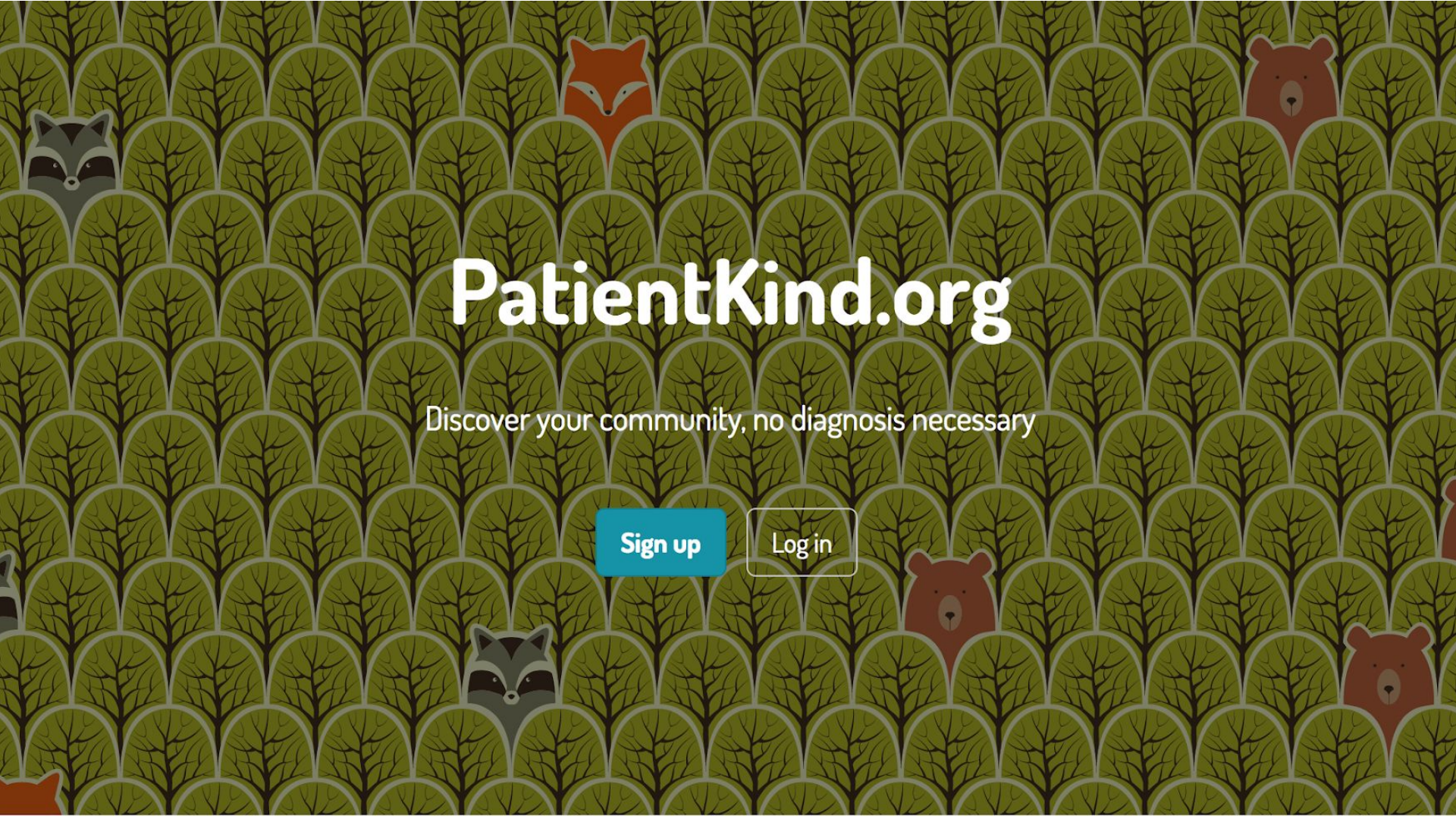
Showing similar cases 1-10 out of 50 per page of 10

Case ID	Diagnosis	Contact	Relevance
P0004612	FLOATING-HARBOR SYNDROME	Orion Buske	57%
F0000015	FLOATING-HARBOR SYNDROME	PhenomeCentral	56%
F0000009	FLOATING-HARBOR SYNDROME	PhenomeCentral	55%

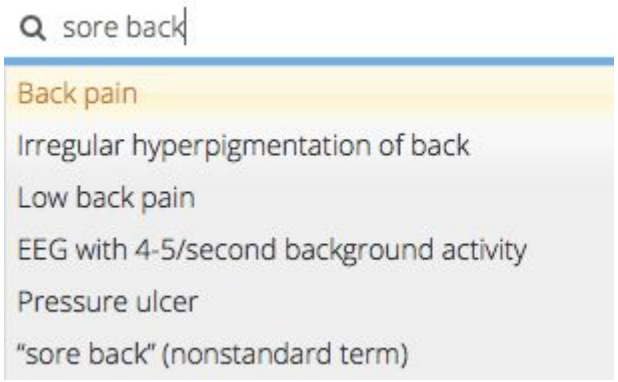
PHENOME CENTRAL

Matchmaker Exchange

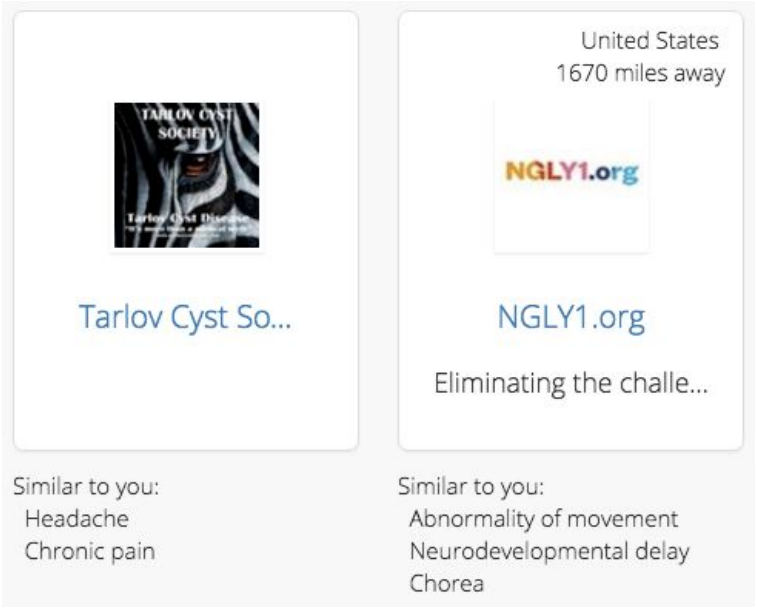
How do we make such tools accessible to patients?


PatientKind.org
Discover your community, no diagnosis necessary
Sign up
Log in

1. Add symptoms, genes, diagnoses



2. Find people and advocacy organizations that are similar



Lessons learned

- Patients and advocates were incredibly generous with their time and energy
- The tool required more expertise than many patients had
  - patient-friendly symptom terminologies would have helped (now available)
- This pilot would have been more successful if I had involved patients in the process sooner and more deeply
- I was a grad student and had no idea how to engage with patients as a researcher #HowNotToDoPtEngagement

The birth of the **new** RareConnect.org

Denis Costello

Ana Rath

RareConnect.org
A PARTNERSHIP OF EURORDIS AND NORD

PatientKind.org

slide by Dorin Manase

The birth of the **new** RareConnect.org

slide by Dorin Manase

The birth of the **new** RareConnect.org

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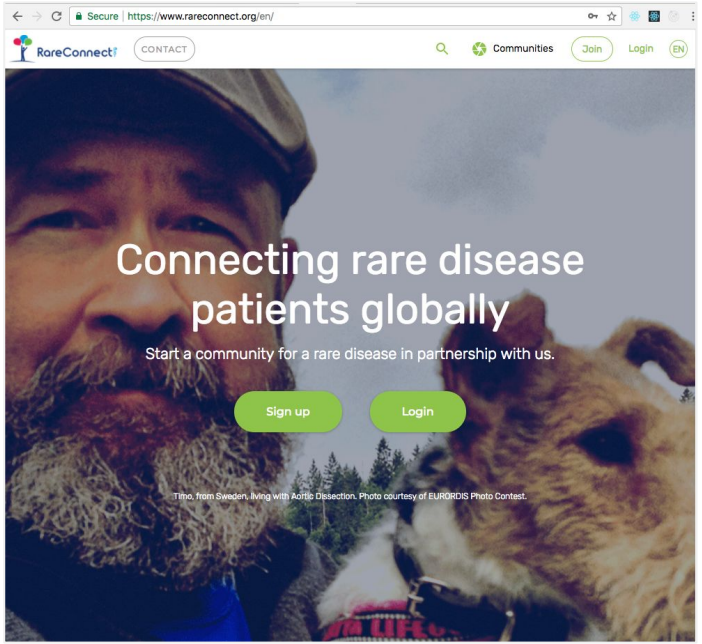
Goals

- Modernize the platform
- Mobile-friendly
- APIs to enable data interoperability
- Enable patients to share data with researchers
- Provide more support for undiagnosed patients through matching

What is RareConnect.org?

A **moderated** support platform where rare disease patients, families, and patient organizations can develop **online communities** across continents and languages



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How does it work?

- Create a profile
- Join communities
- Start a discussion
- Share your story
- Meet and connect with others

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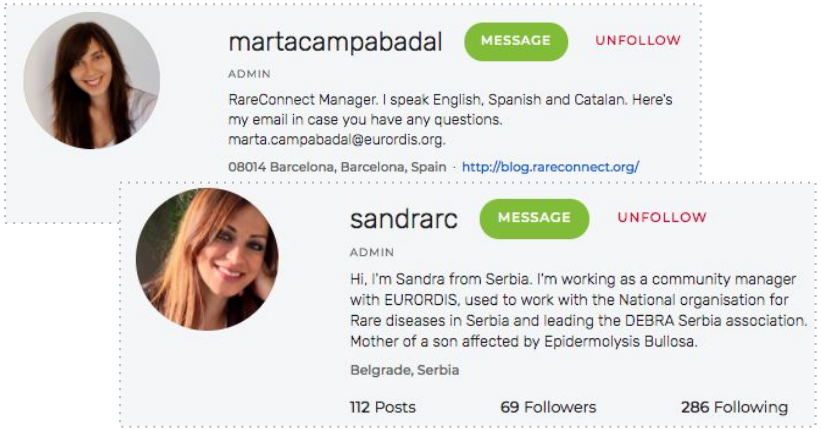
Moderators and managers

RareConnect Managers



Community Moderators

\+ **380**

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By the numbers

150
global communities

900
advocacy organizations

34K
members

12
languages

2K
patient testimonials

52K
posts & comments

CZ, DE, EN, ES, FA, FR, JA, IT, PT, SH, RU, UA

Goals

√ Modernize the platform

√ Mobile-friendly

√ APIs to enable data interoperability

... Enable patients to share data with researchers

X Provide more support for undiagnosed patients through matching

Now extending to support research

https://www.rareconnect.org/en

Consent to de-identified data sharing

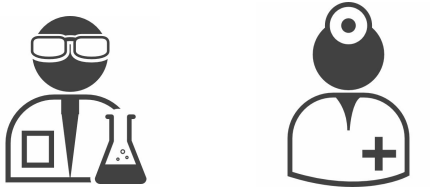
Research RareConnect.org

https://research.rareconnect.org/

Patients

Caregivers

Advocacy Organizations



Researchers

Clinicians

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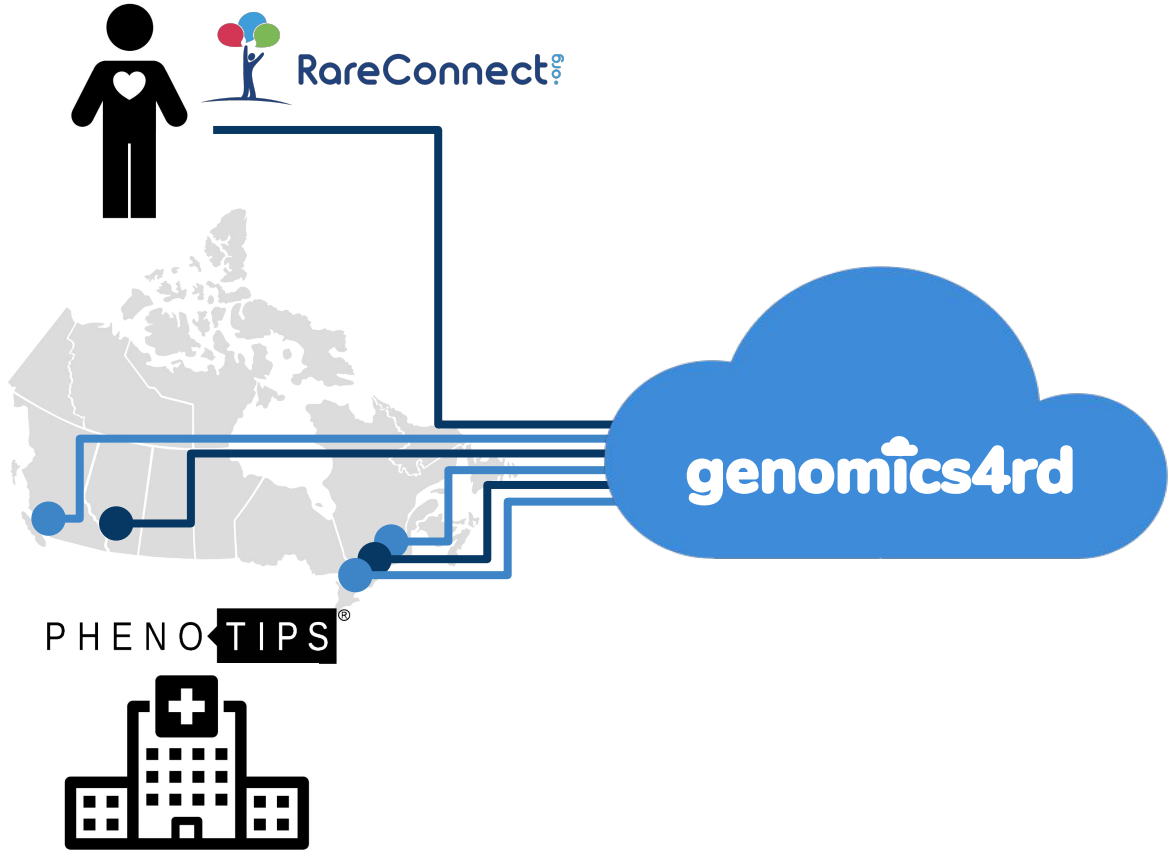
The two arms of a Canada-wide RD data repo



Special thanks

Dorin Manase, Amanda Silva

Michael Brudno and the SickKids teams (PhenoTips, PhenomeCentral, RareConnect)

Kym Boycott and the Care4Rare team

Denis Costello, Marta Campabadal, Sandra Pavlovic, and the original RareConnect team

Ana Roth

The patients and advocates

The funders