

Building tools to empower patient experts

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RareConnect.org



Gene42

Structured data data

MEDICAL HISTORY AND PHYSICAL
NAME: John Doe AGE: 45 DATE: 12/10/20
1. Improved Stable Worse
2. None
3. None
4. None
HYPER: PALPITATIONS 0 TIREMORSE 0 ANXIETY 0 WT LOSS 0
HEAT INTOLER 0 SWEATING 0 WEAKNESS 0 HAIR LOSS 0
DIARRHEA 0 INSOMNIA 0 VISION 0 RASH 0
MENSTRUATION: 0 OTHERS: 0
HYPO: FATIGUE 0 DRY SKIN 0 COLDNESS 0 MUSCLE SPAS 0
SLEEPINESS 0 CONSTIPA 0 VOICE CHAN 0 WT GAIN 0
EDEMA 0 MENSTRUATION 0
NODULE 0
DATE FOUND: 0 PAIN 0 DYSPHAGIA 0 SOB 0
VOICE CHAN 0 RX OF RADIATION 0 FHX 0
ALL OTHER SYSTEMS 0 PSFHX REVIEW 0
PE: WT: 180 HT: 70 BP: 120/80 P: 72 PAIN 0
EYES: CONJ: 0 EXOPHT: 0 R 0 L 0
THYROID: 0
LYMPH C: 0 SC: 0 BRUTES: 0
LUNGS: 0 HEART: 0
ABD: 0 XT: 0 PULM: 0 EDEMA: 0
NEURO: 0 DTRS: 0 SKIN: 0
LABS: TSH: 0 TT4: 0 FT4: 0 T3: 0 FT3: 0
AMA: 0 TG: 0 TGA: 0 CBC: 0 LFTS: 0
THYROID US: 0
THYROID UPTAKE SCAN: 0
RECORDS REVIEWED: 0
F1: 0
F2: 0
LABS: TSH: 0 THYROID PROFILE: 0 TT4: 0 FT4: 0 T3: 0
FT3: 0 AMA: 0 TG: 0 TGA: 0 CBC: 0 LFTS: 0
US: 0 THY US SCAN: 0 FNAB: 0 FNAB US: 0
RTO: 0 WKS: 0 MOS: 0 YR: 0

ataxia, developmental delay

HP:0001251
HP:0001263

faster (sometimes)

better

Tools for doctors and researchers

Quick phenotype search:
seizur

☒ Seizures ⓘ
(also known as: Seizures, partial, afebrile)

☒ Focal seizures ⓘ
(also known as: Seizures due to hypocalcemia)

☒ Hypocalcemic seizures ⓘ
(also known as: Seizures due to hypocalcemia)

☒ Atonic seizures ⓘ

Deep phenotyping

Related terms

- ☒ [HP:0001250] Seizures ⓘ
- ☒ [HP:000197] Generalized seizures ⓘ
- ☒ [HP:000121] Absence seizures ⓘ
- ☒ [HP:001148] Absence seizures with special features ⓘ
- ☒ [HP:001149] Absence seizures with eyelid myoclonia ⓘ
- ☒ [HP:001150] Myoclonic absences ⓘ
- ☒ [HP:000727] Atypical absence seizures ⓘ
- ☒ [HP:001151] Obtundation status ⓘ
- ☒ [HP:001147] Typical absence seizures ⓘ
- ☒ [HP:000125] Early onset absence seizures ⓘ
- ☒ [HP:000125] Atonic seizures ⓘ
- ☒ [HP:000125] Generalized clonic seizures ⓘ
- ☒ [HP:000125] Generalized myoclonic seizures ⓘ
- ☒ [HP:000125] Myoclonic atonic seizures ⓘ

PHENOTIPS®

Gene panels

MATCHING GENES [DOWNLOAD](#) ⓘ

The following terms are extracted from the phenotypic description and used automatically in searches. You can disable or re-enable their contribution in the search results by clicking on them.

Childhood onset **NO Abnormal delivery** Abnormality of the nasal dorsum Abnormality of the pinna Abnormality of the testis Absent axillary hair Absent eyebrow Absent eyelashes Absent facial hair Absent testis Asymmetry of the ears Delayed eruption of teeth Dental malocclusion Depressed nasal ridge Dry skin Freckling Generalized hypochidrosis Hypochidrosis Midface retrusion Milia Numerous pigmented freckles Phimosis **NO Premature birth** Pterygium Scoliosis Scrotal hypoplasia Soft skin Sparse eyelashes Sparse hair White hair

LMNA ⓘ

Abnormality of the nasal dorsum; Abnormality of the pinna; Abnormality of the testis; Absent eyebrow; Absent eyelashes; Absent facial hair; Delayed eruption of teeth; Hypochidrosis; Midface retrusion; Scoliosis; Sparse eyelashes; Sparse hair

MBTPS2 ⓘ

Abnormality of the nasal dorsum; Abnormality of the pinna; Abnormality of the testis; Absent eyebrow; Absent eyelashes; Absent facial hair; Dry skin; Hypochidrosis; Hypochidrosis; Midface retrusion; Scoliosis; Sparse eyelashes; Sparse hair

Relevant genes

TWIST2 ⓘ

Abnormality of the nasal dorsum; Abnormality of the pinna; Abnormality of the testis; Absent eyebrow; Absent eyelashes; Absent facial hair; Dental malocclusion; Dry skin; Freckling; Midface retrusion; Pterygium; Scoliosis; Scrotal hypoplasia; Soft skin; Sparse eyelashes; Sparse hair; White hair

Diagnosis

INSTANT OMIM SEARCH

The following terms are extracted from the phenotypic description and used automatically in searches. You can disable or re-enable their contribution in the search results by clicking on them.

Arthritis Conjunctivitis Fever Headache Hearing impairment Skin rash

Matching disorders in OMIM

[OMIM:120100] #120100 FAMILIAL COLD AUTOINFLAMMATORY SYNDROME 1

[OMIM:142680] #142680 PERIODIC FEVER, FAMILIAL, AUTOSOMAL DOMINANT

[OMIM:124950] 124950 DEAFNESS, SENSORINEURAL, WITH PERIPHERAL NEUROPATHY AND ARTERIAL DISEASE







Relevant diseases

[OMIM:103400] 103400 MUKO-POLLY-OSSEOUS SYNDROME

[OMIM:103400] 103400 MUKO-POLLY-OSSEOUS SYNDROME

[OMIM:103400] 103400 MUKO-POLLY-OSSEOUS SYNDROME

Finding similar cases

Similar cases			
MATCHES WITH PATIENT RECORDS IN THIS DATABASE			
Showing similar cases 1-10 out of 50 per page of <input type="text" value="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%



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

🔍 sore back

Back pain

Irregular hyperpigmentation of back

Low back pain

EEG with 4-5/second background activity

Pressure ulcer

"sore back" (nonstandard term)

2. Find people and advocacy organizations that are similar



Tarlov Cyst So...

Similar to you:
Headache
Chronic pain

United States
1670 miles away

NGLY1.org

NGLY1.org

Eliminating the challe...

Similar to you:
Abnormality of movement
Neurodevelopmental delay
Chorea

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



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



slide by Dorin Manase

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



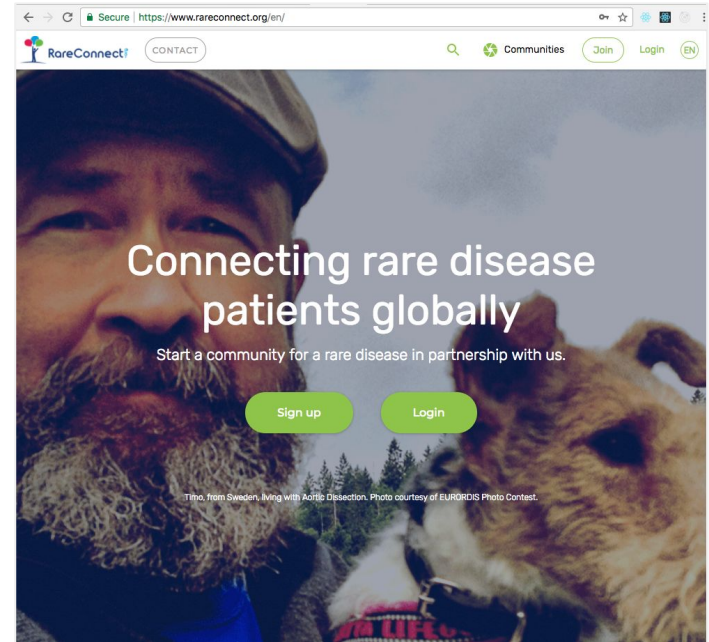
slide by Dorin Manase

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



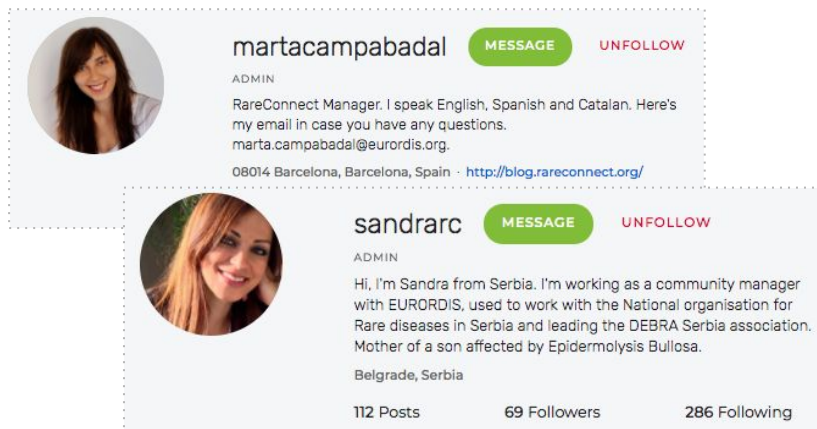
slide by Dorin Manase

How does it work?

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

Moderators and managers

RareConnect Managers



Community Moderators

+

380

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
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- ... Enable patients to share data with researchers
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Now extending to support research



<https://www.rareconnect.org/en>



Patients



Caregivers



Advocacy
Organizations



Consent to
de-identified
data sharing

**Research
RareConnect.org**

<https://research.rareconnect.org/>



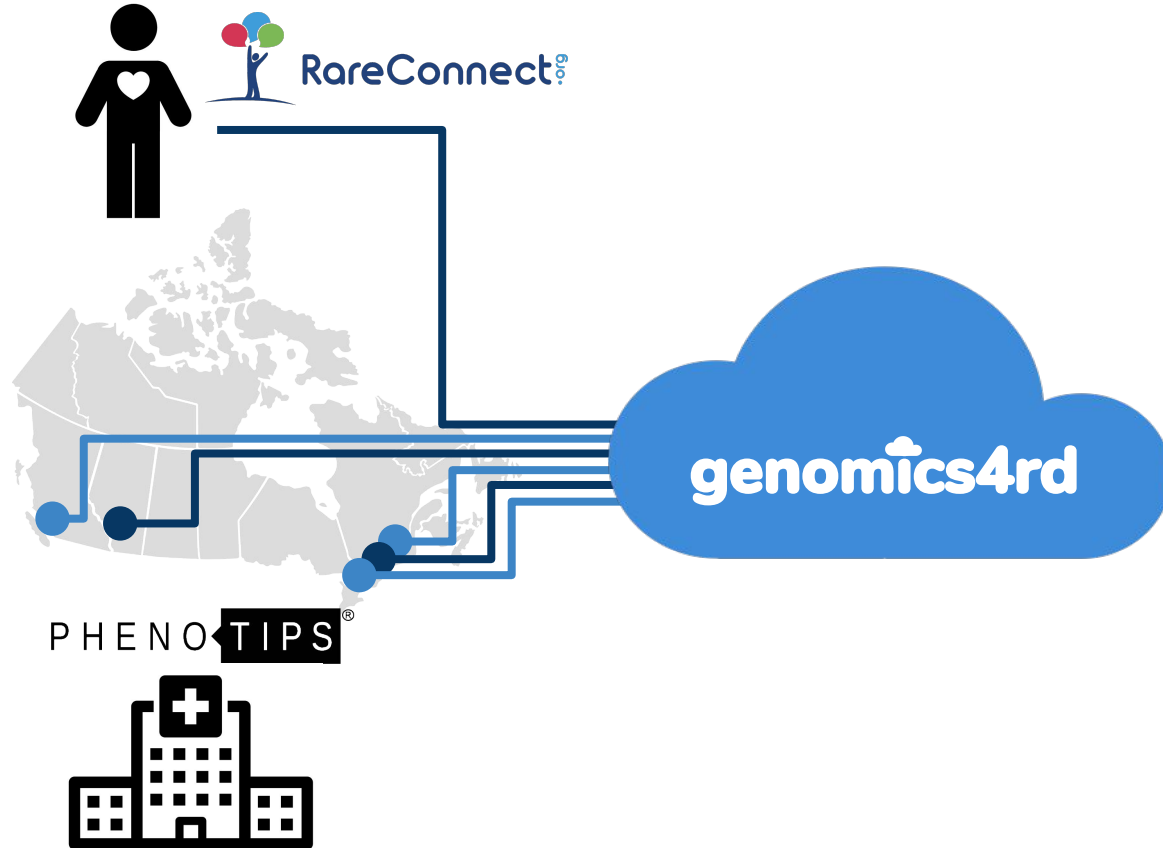
Researchers



Clinicians

slide by Dorin Manase

The two arms of a Canada-wide RD data repo



Special thanks

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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