



The **UNDn** Undiagnosed Diseases Network

Anastasia Wise, PhD

Division of Genomic Medicine, NHGRI

May 21, 2018



NIH > Objectives of the UDN

1. **Improve the level of diagnosis and care** for patients with undiagnosed diseases
2. **Facilitate research** into the etiology of undiagnosed diseases
3. **Promote an integrated and collaborative community** to investigate these difficult to diagnose diseases





Undiagnosed Diseases Program

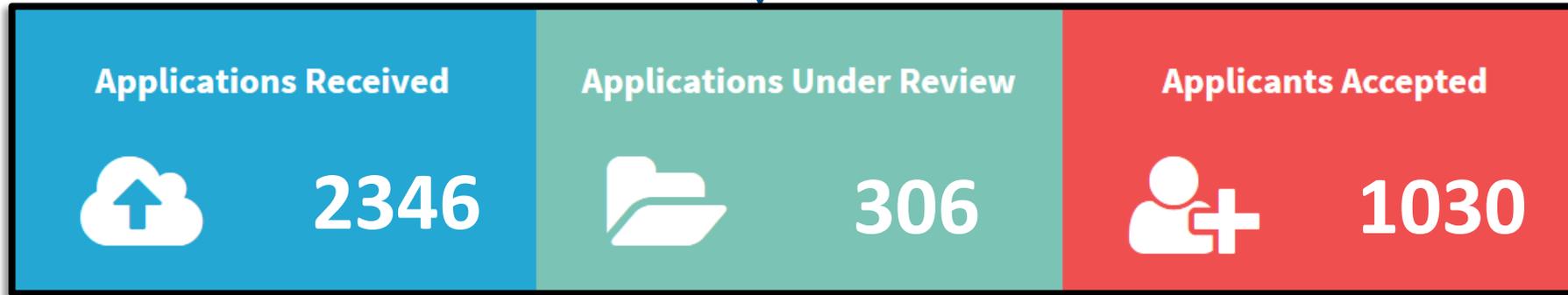
- Began in May 2008
- NIH Clinical Center
- Thorough, one-week evaluations
- No cost to patients
- Un-insured and under-insured patients welcomed











735 Clinical Evaluations



Sequencing
(exome and genome)



Model Organisms
(fly and fish)

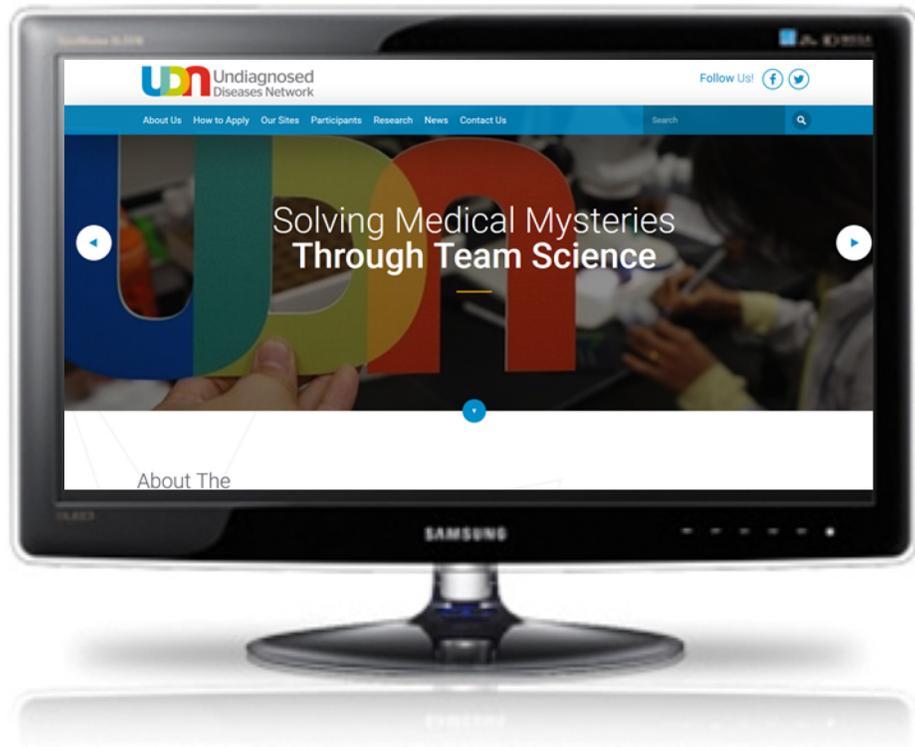
Metabolomics



Working Together 198 Diagnoses



UDN Data Sharing and Outreach



UDN Manual of Operations (MOO)

- Network-wide Protocol
- Single (Central) IRB
- Data Sharing and Use Agreement

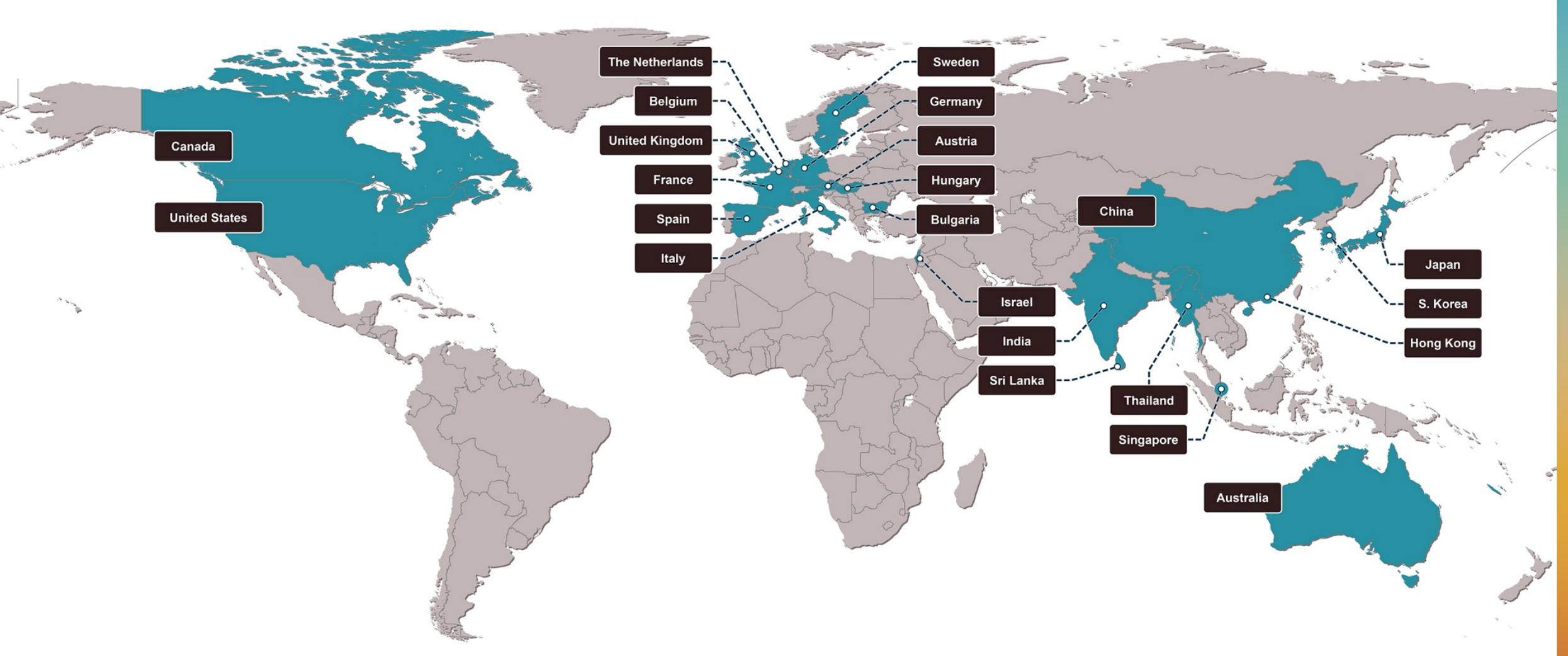


Matchmaker
Exchange



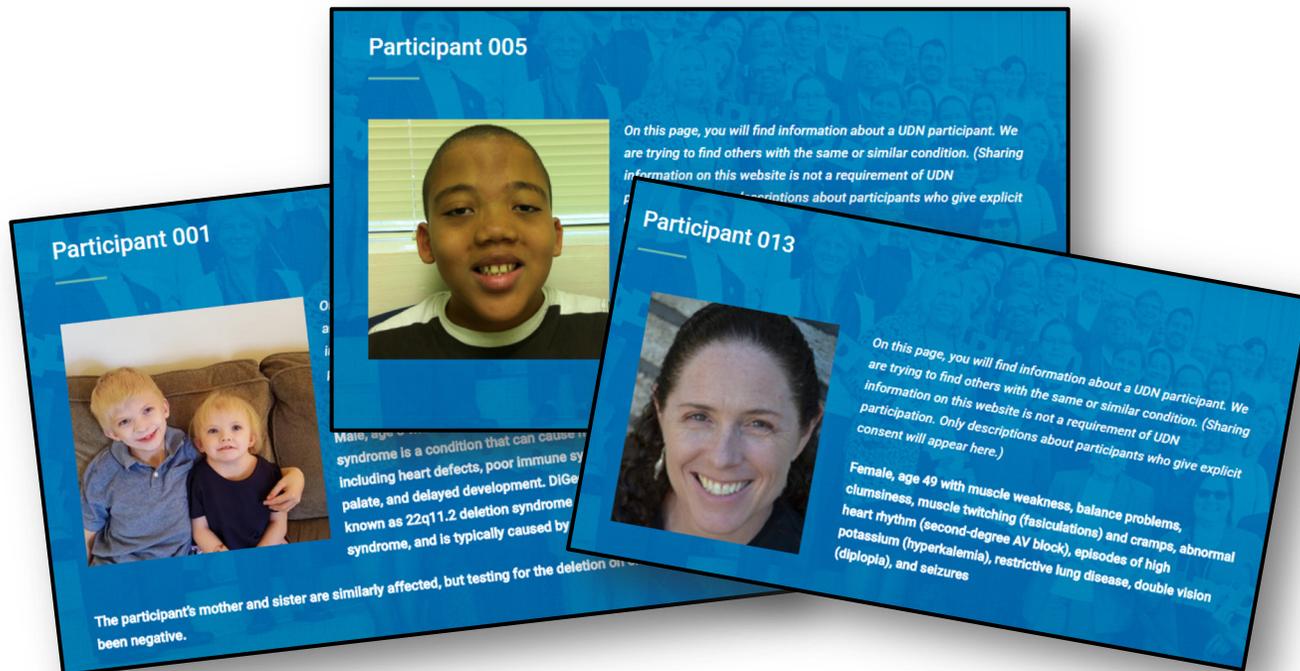
Manual of Operations Available!

[https://undiagnosed.hms.harvard.edu/
udn-manual-of-operations](https://undiagnosed.hms.harvard.edu/udn-manual-of-operations)





UDN Participant Pages



Not Required for UDN Participation



To date 63 participants!

<https://undiagnosed.hms.harvard.edu/participants>



Why Matching Matters: NACC1



Male, age 20 months

Symptoms:

- Seizures
- Cataracts
- Small head size (microcephaly)
- Global developmental delay

If any of these participants sound like you or someone you know, please contact us!

[CONTACT US](#)



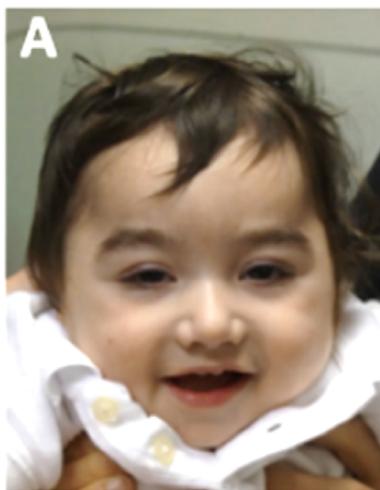
Why Matching Matters: NACC1



GeneMatcher



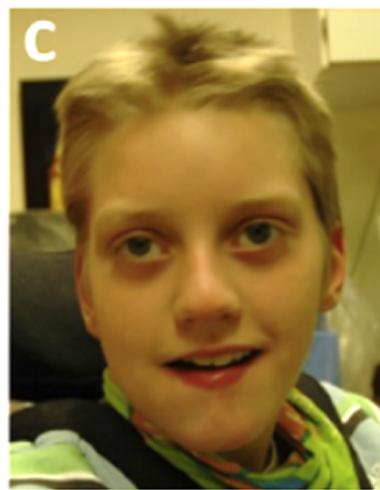
Participant Pages



Individual 1



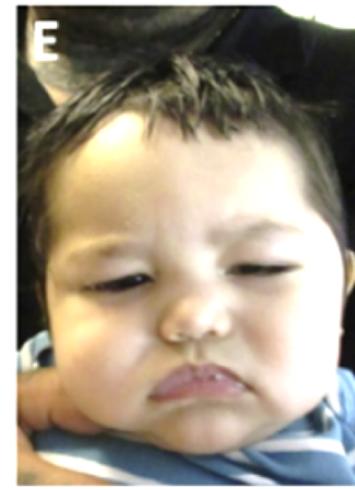
Individual 2



Individual 3



Individual 4



Individual 6



Individual 7

A Recurrent De Novo Variant in NACC1 Causes a Syndrome Characterized by Infantile Epilepsy, Cataracts, and Profound Developmental Delay

January 26, 2017

The American Journal of Human Genetics



UDN Data Sharing and Outreach



ClinicalTrials.gov

NCBI

ClinVar



To date 28 publications!

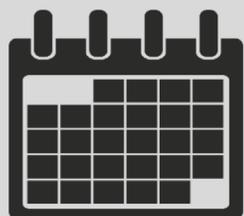
https://undiagnosed.hms.harvard.edu/research/publications_page

NIH **Status of UDN Phase II**



Renewed for another 5 years!

- Coordinating Center
- Clinical Sites
- Metabolomics Core
- Model Organisms Screening Center
- Sequencing Core



Upcoming Council Review in **May 2018**
Anticipate award **Fall 2018**



NIH Acknowledgements



Thank you to all the UDN patients and their families!



<http://undiagnosed.hms.harvard.edu/>

NIH Acknowledgements



Thank you to all the UDN investigators!



<http://undiagnosed.hms.harvard.edu/>

NIH Acknowledgements

Common Fund UDN WG

Co-Chair: Eric Green (NHGRI)

Co-Chair: Walter Koroshetz (NINDS)

Co-Coordinator: **William Gahl (NHGRI)**

CF Program Leader: **Mary Perry (OD)**

For More Information on the UDN

<https://commonfund.nih.gov/Diseases>

<http://www.genome.gov/27550959>

<http://rarediseases.info.nih.gov/undiagnosed>

David Adams (NHGRI)

Gabriel Batzli (NHGRI)

Heather Colley (NHGRI)

Jyoti Dayal (NHGRI)

David Eckstein (OD)

Sheena Faherty (NHGRI)

Sarah Gould (NHGRI)

Holli Hamilton (NIDCR)

W. Keith Hoots (NHLBI)

Ellen Howerton (NHGRI)

Donna Krasnewich (NIGMS)

Laura Mamounas (NINDS)

Teri Manolio (NHGRI)

Aron Marquitz (OD)

Pamela M. McInnes (NCATS)

John J. Mulvihill (NHGRI)

Melissa Parisi (NICHD)

Mary Purucker (NCATS)

Daniel Rotrosen (NIAID)

Averell H. Sherker (NIDDK)

Cyndi Tifft (NHGRI)

Lois Tully (NINR)

Tiina Urv (NCATS)

Carol Weil (NCI)

Yong Yao (NIMH)

THANKS!

anastasia.wise@nih.gov

 Undiagnosed
Diseases Network

<http://undiagnosed.hms.harvard.edu/>