Canadian networks linking research to care

Kym Boycott
Professor of Pediatrics,
CHEO, University of Ottawa
Identify the causes of rare diseases
Facilitate access to testing
>1500 rare diseases studied
>5000 families recruited
50% of families diagnosed
100s of novel discoveries
USP7

TMEM106B

Matchmaker Exchange

International data sharing for discovery
Connecting 50,000 undiagnosed patients
From 67 countries
Partner every new rare disease gene we discover with a scientist to study its function.
81 partnerships and counting
514 scientists registered
Connecting now to Europe, Japan and Australia
Partnering with patient organizations
Connecting patients and families globally

Enabling patients and families as research partners
“We have found a community”
-Quinlan’s dad

40,000 users
179 patient communities
950 patient organizations

12 languages
30 new users a day
2 studies
Unprecedented international cooperation to:

- The Grand Challenge
- History & Leadership of Gene Mapping
- Patient Experiences & Stories
- Research
- Relationships with patients
- Understanding when, how, and why
- Patients intersect with health care systems
- Creating notifications for innovation
- What is my role in improving the patient experience?