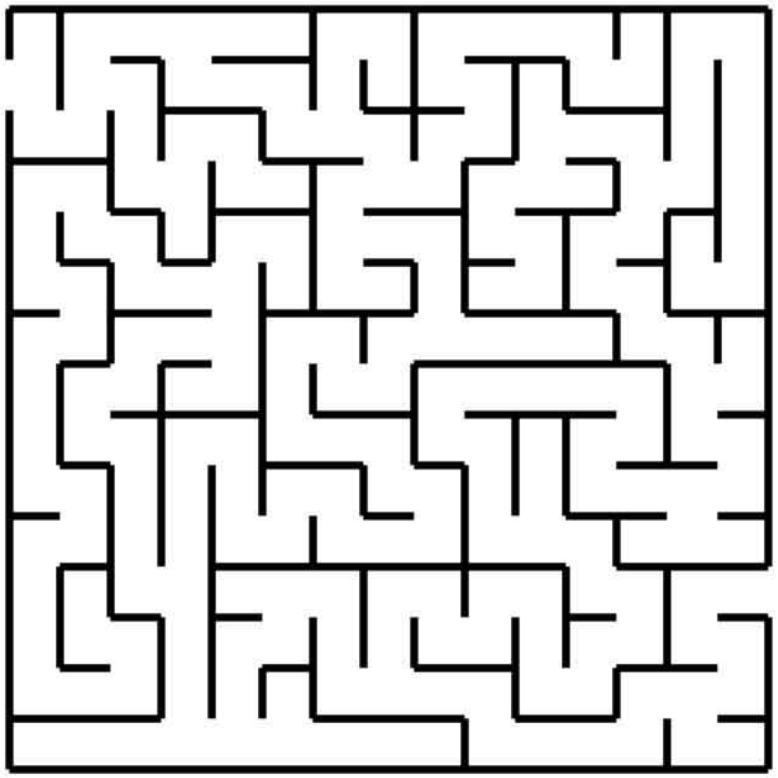


Diagnostic Breakthroughs:

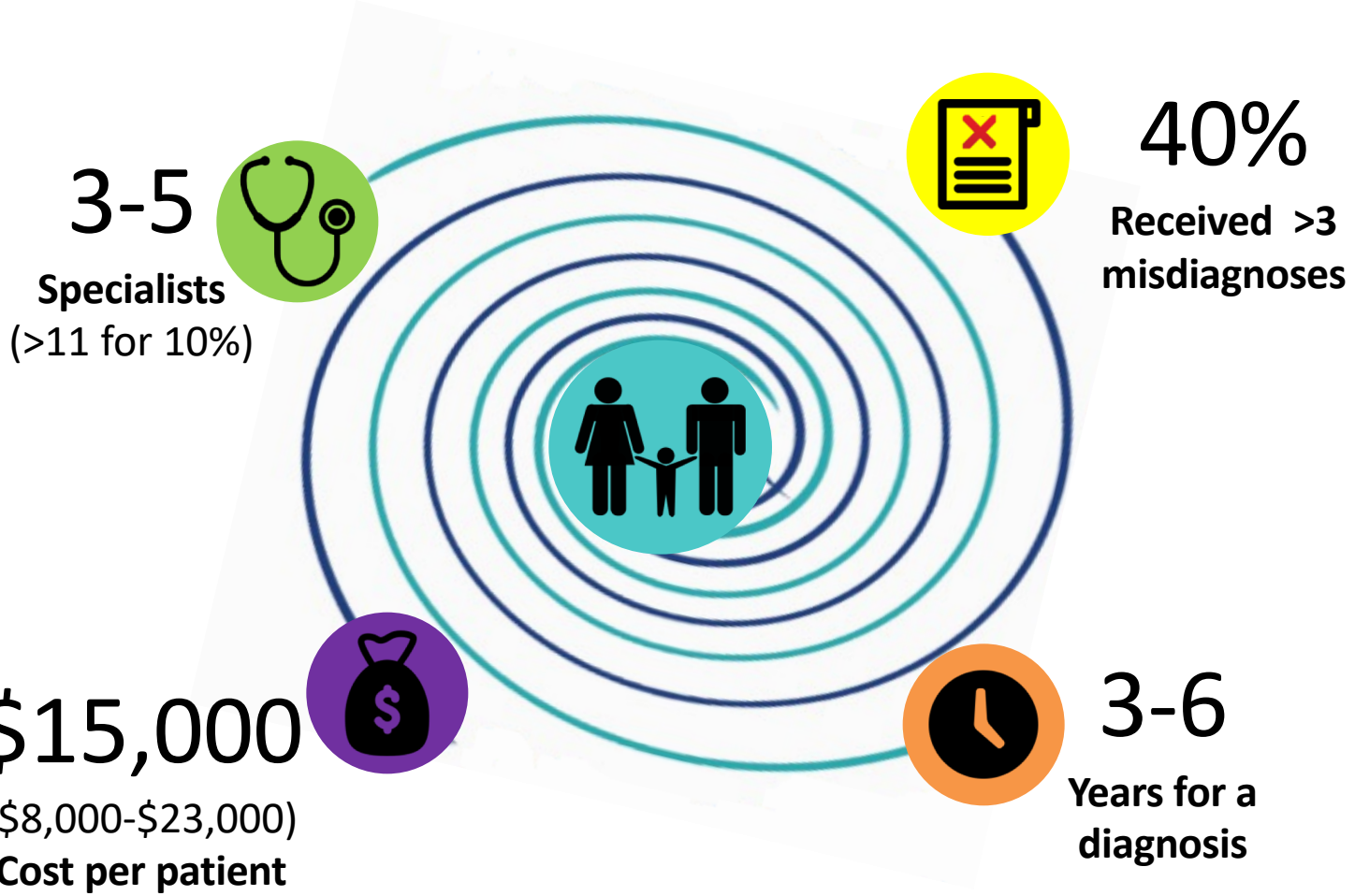
the path to access to the right test at the right time

Kym Boycott, **MD, PhD, FRCPC, FCCMG**

Professor of Pediatrics, University of Ottawa

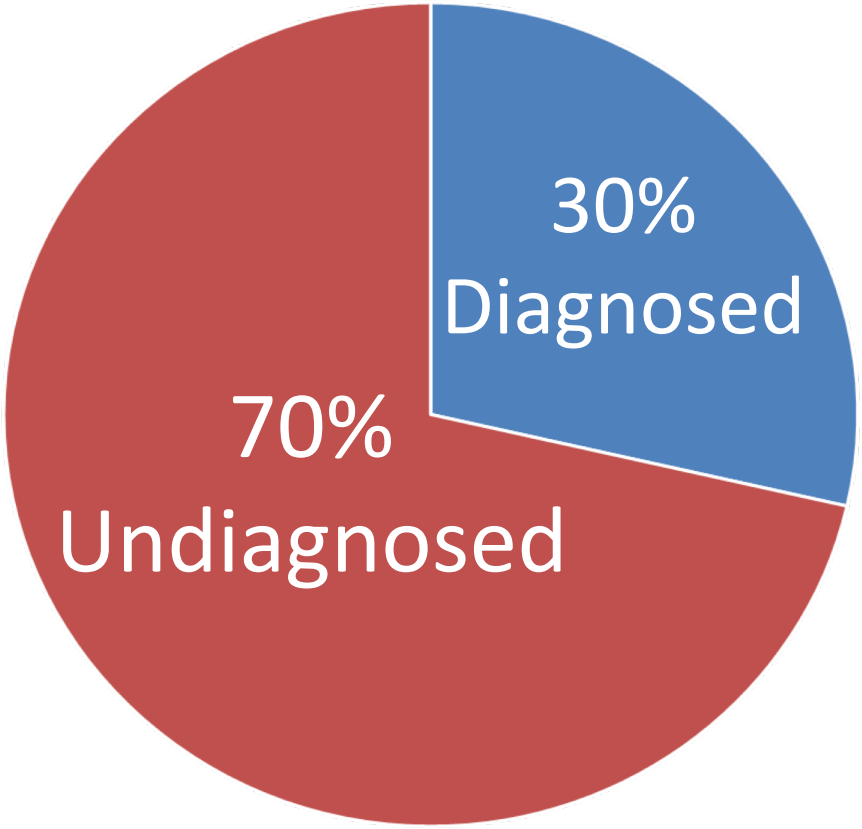
Unmet Clinical Need

Access to appropriate testing can be challenging
Our understanding of the causes of RDs is incomplete



Diagnostic Breakthrough

More than 200,000 rare disease patients now clinically sequenced world-wide



Clinical genome-wide sequencing

So along comes the best test we've ever had…

Diagnostic translation

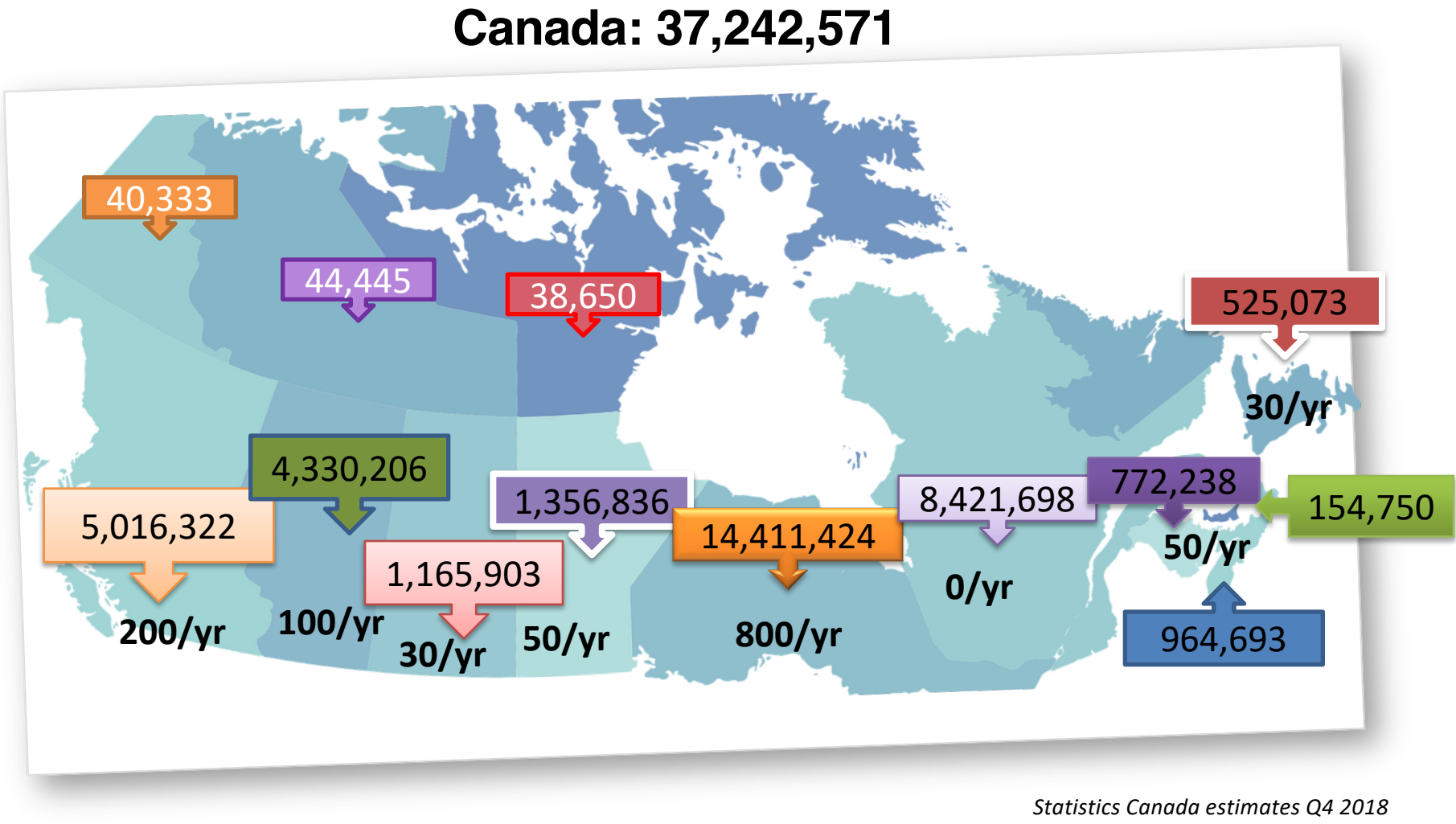
CCMG guidelines:

ORIGINAL ARTICLE

The clinical application of genome-wide sequencing for monogenic diseases in Canada: Position Statement of the Canadian College of Medical Geneticists

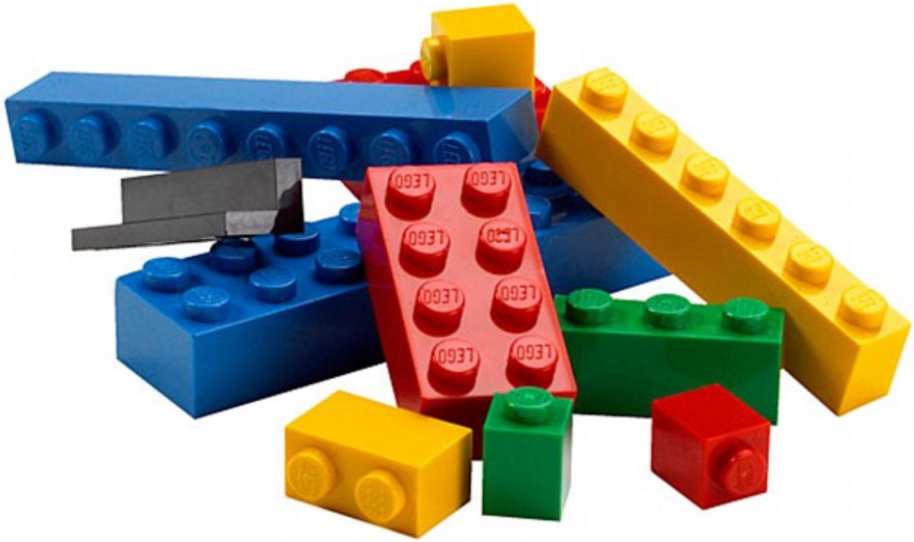
Kym Boycott,[1] Taila Hartley,[1] Shelin Adam,[2] Francois Bernier,[3] Karen Chong,[4,5] Bridget A Fernandez,[6] Jan M Friedman,[2] Michael T Geraghty,[1] Stacey Hume,[7] Bartha M Knoppers,[8] Anne-Marie Laberge,[9] Jacek Majewski,[10] Roberto Mendoza-Londono,[4] M Stephen Meyn,[4,11] Jacques L Michaud,[9] Tanya N Nelson,[12] Julie Richer,[1] Bekim Sadikovic,[13] David L Skidmore,[14] Tracy Stockley,[15] Sherry Taylor,[7] Clara van Karnebeek,[2] Ma'n H Zawati,[8] Julie Lauzon,[3] Christine M Armour,[1] on behalf of the Canadian College of Medical Geneticists

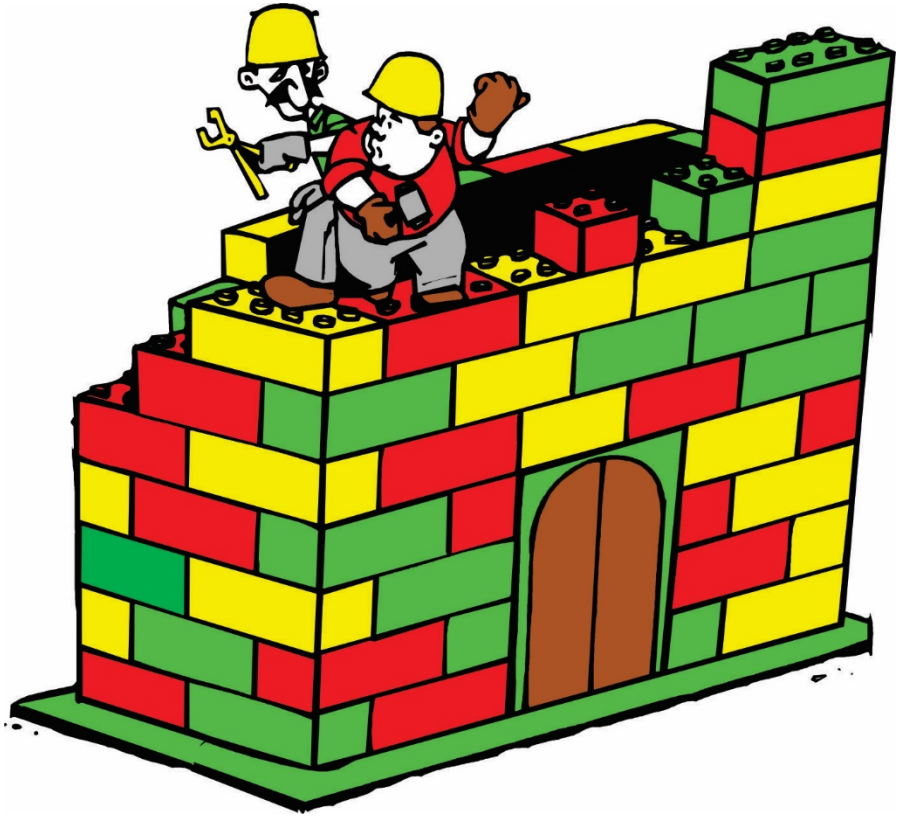
Journal of Medical Genetics 2015

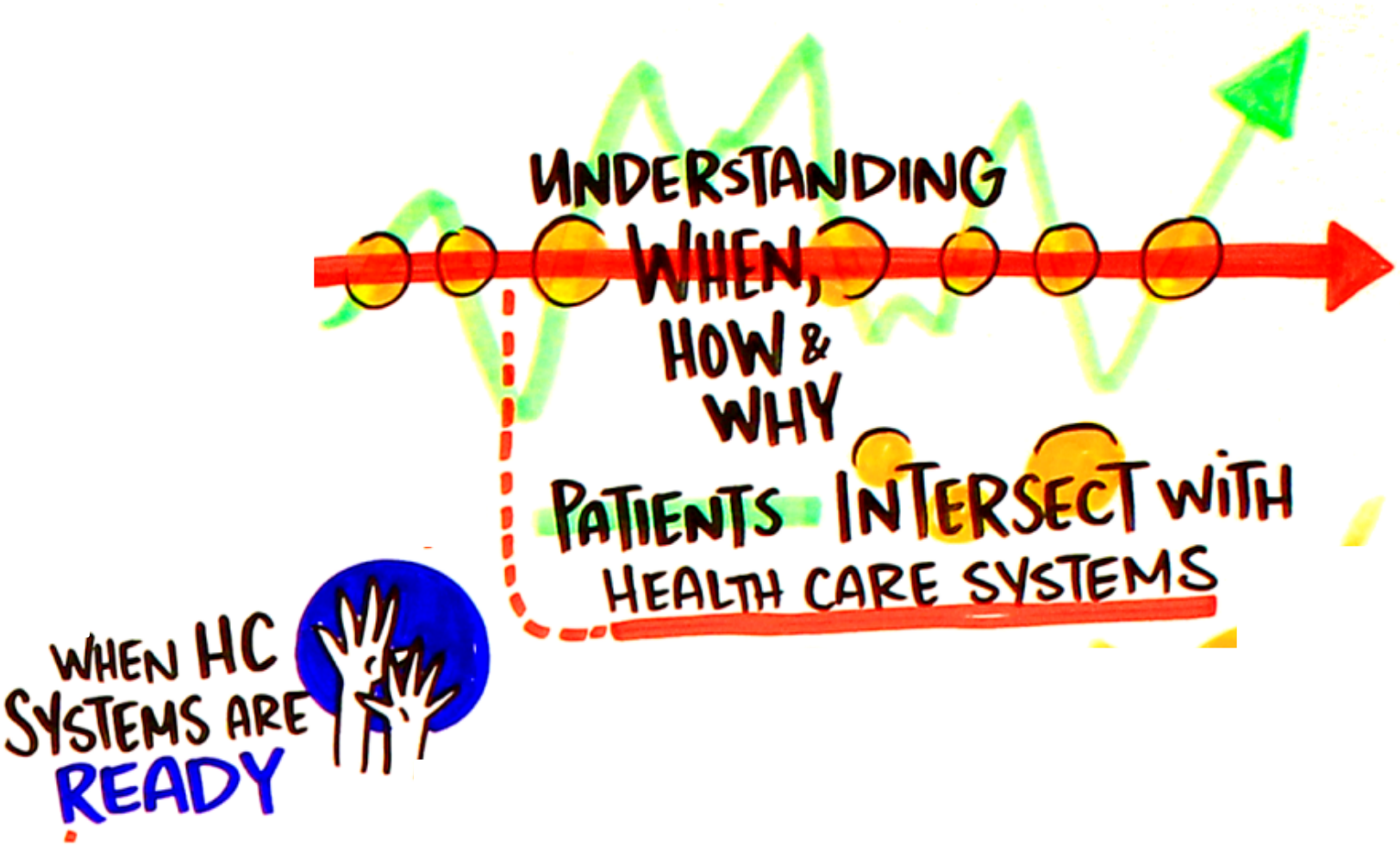


Access to clinical exome sequencing in 2018


FITTT!
FITTT!
FTTTT!

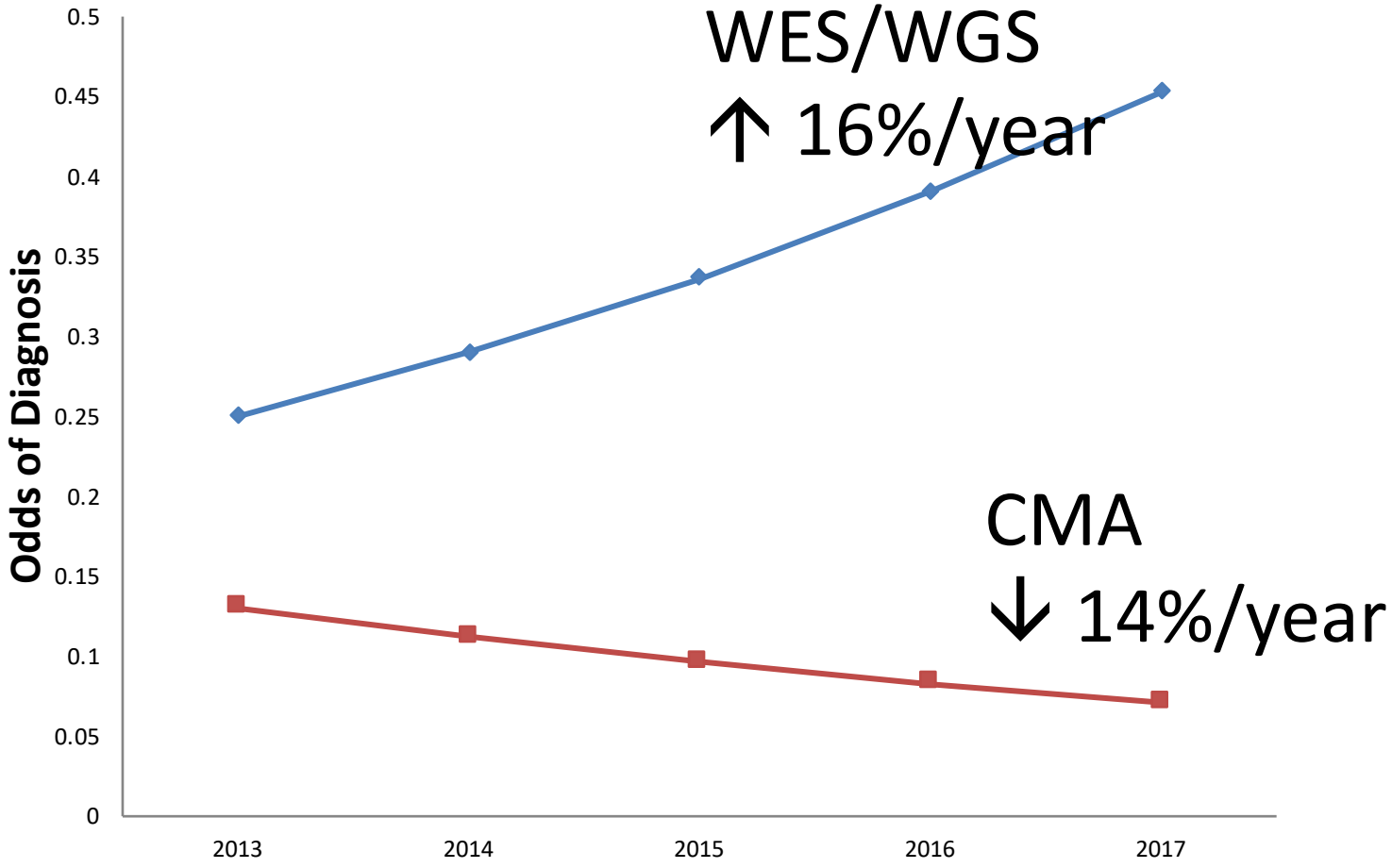




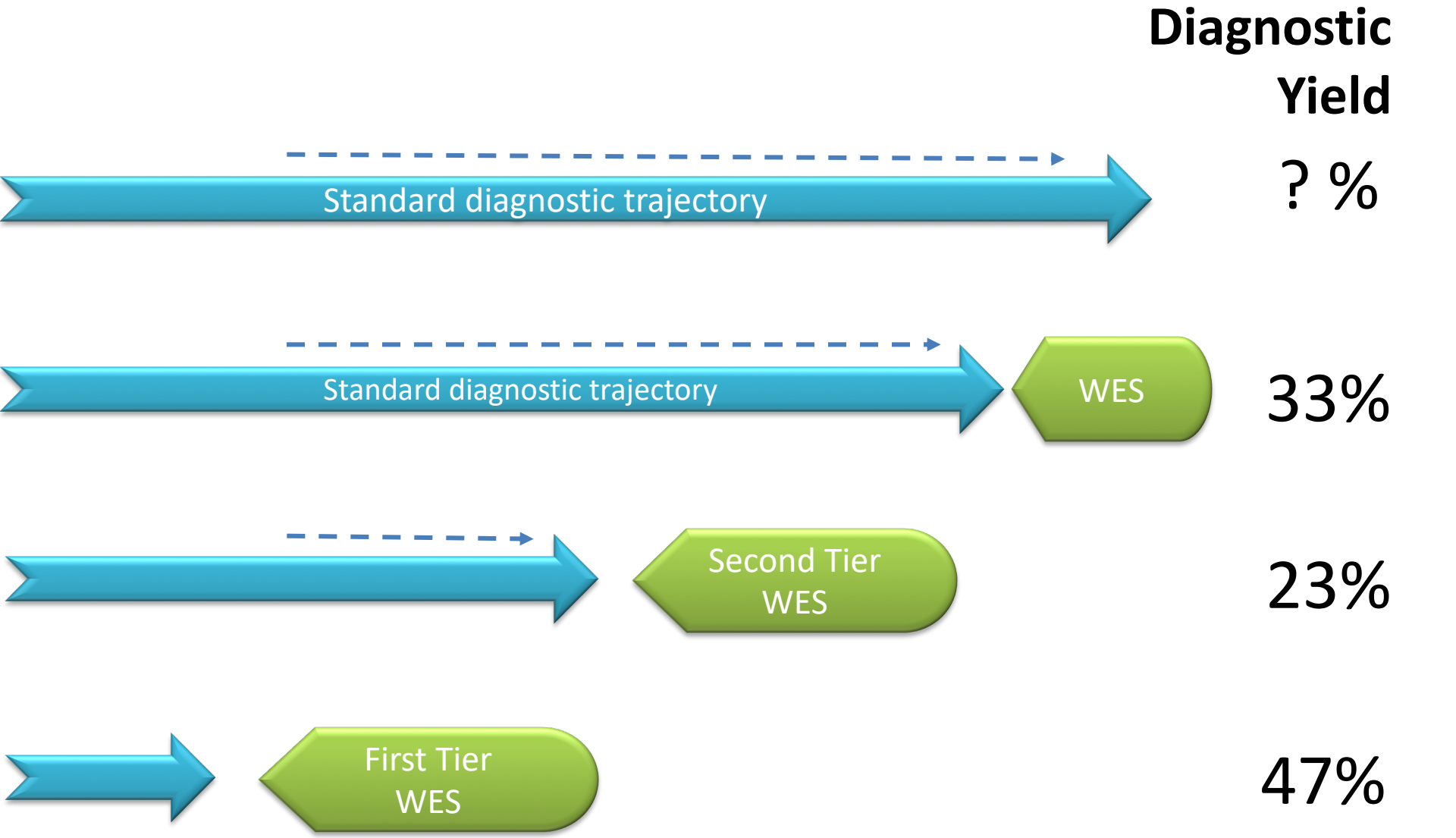


Diagnostic translation

Available to the right patient



At the right time



200 families

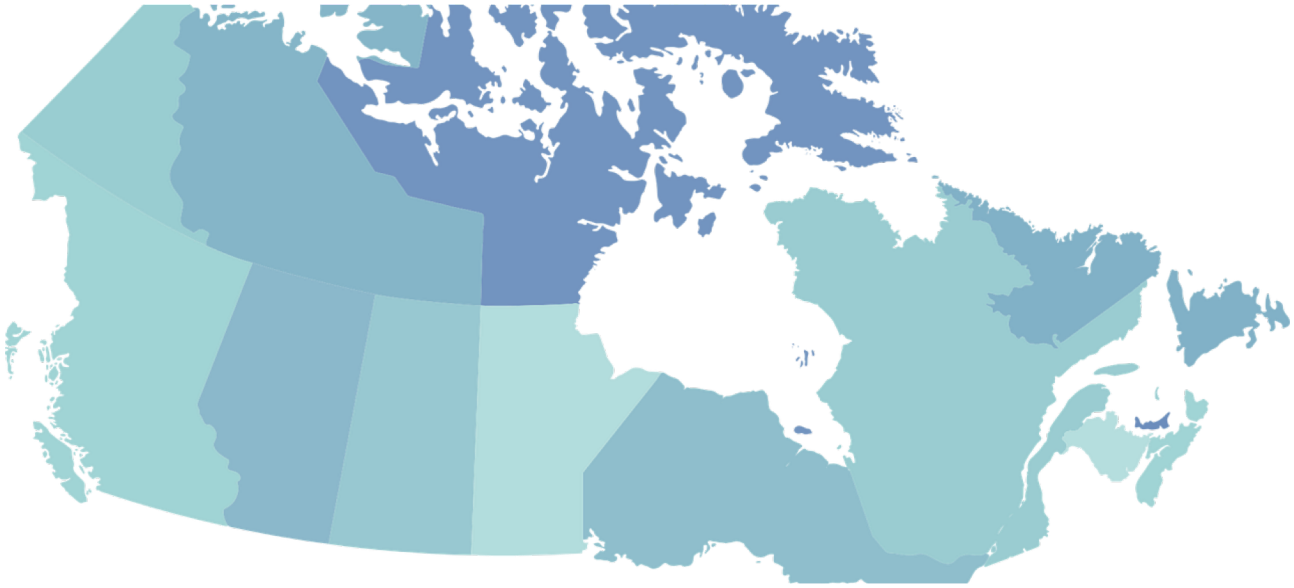
GenomeCanada

FUNDING OPPORTUNITIES

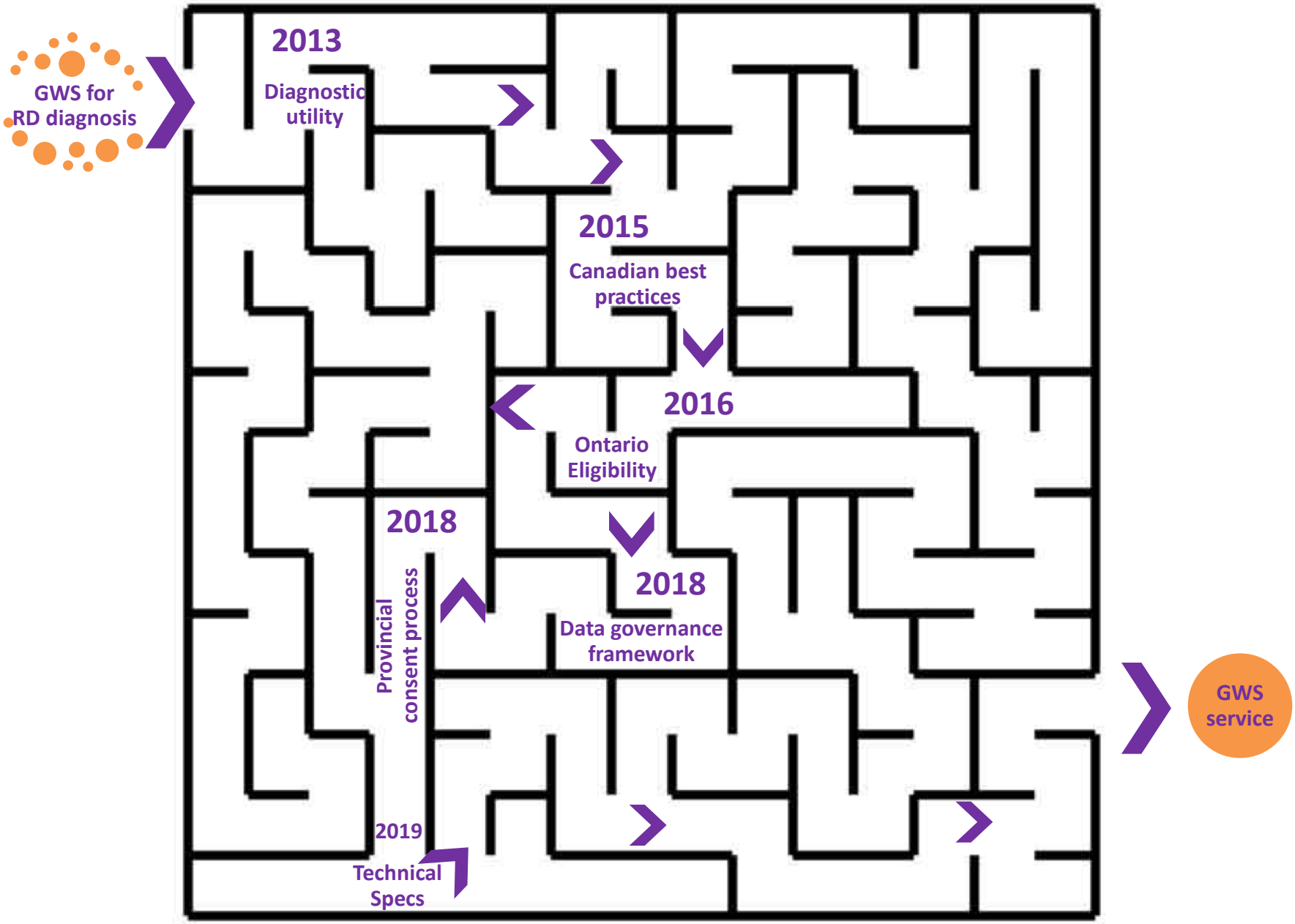
PRECISION HEALTH INITIATIVE – RARE DISEASES: CLINICAL IMPLEMENTATION PROJECTS

GENOMIC APPLICATIONS PARTNERSHIP PROGRAM

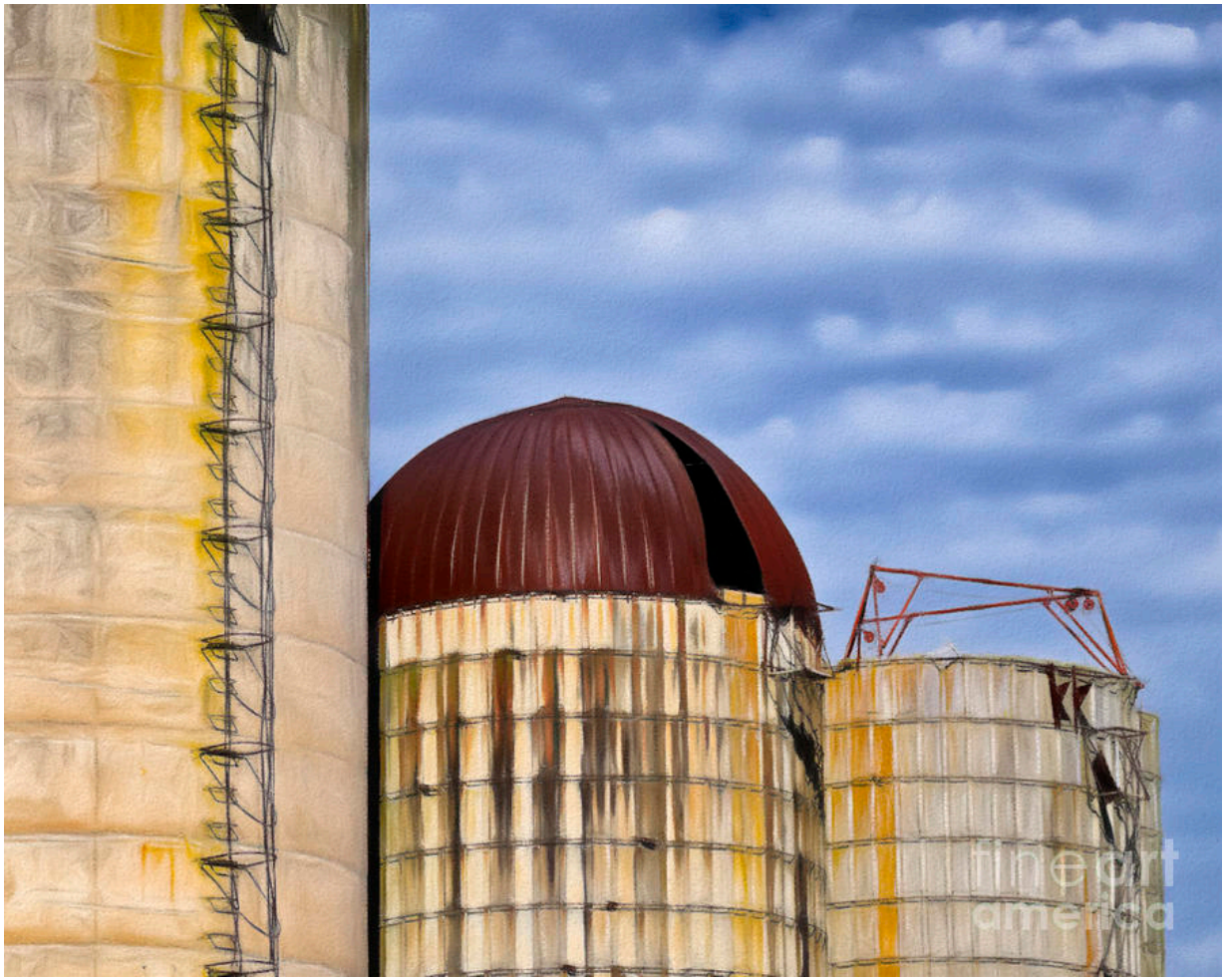
Access for all Canadians



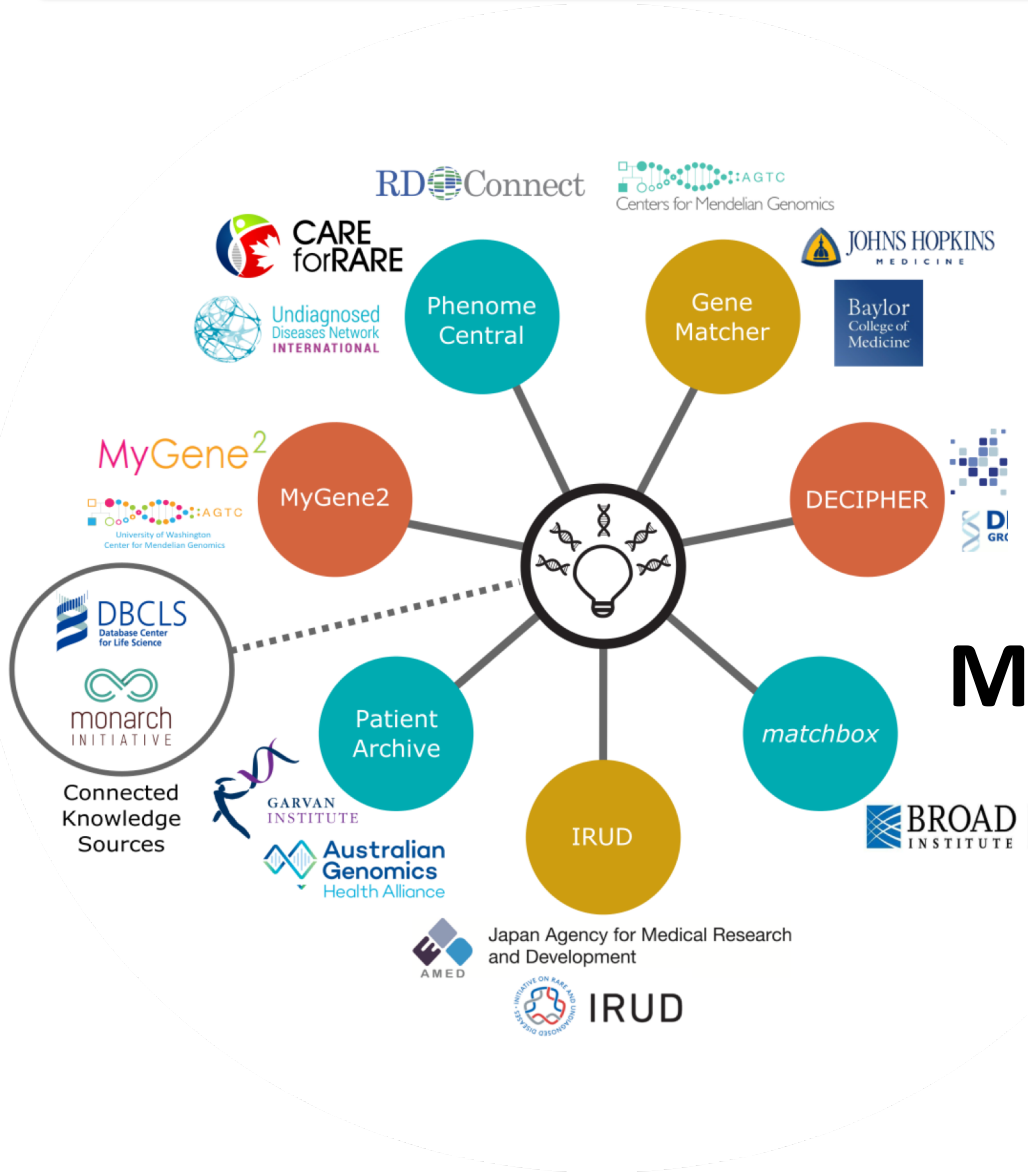
Clinical implementation for RD precision health


GWS for RD diagnosis
2013
Diagnostic utility
2015
Canadian best practices
2016
Ontario Eligibility
2018
Provincial consent process
2018
Data governance framework
2019
Technical Specs
GWS service

The risk for rare diseases



Global Collaboration



Matchmaker Exchange

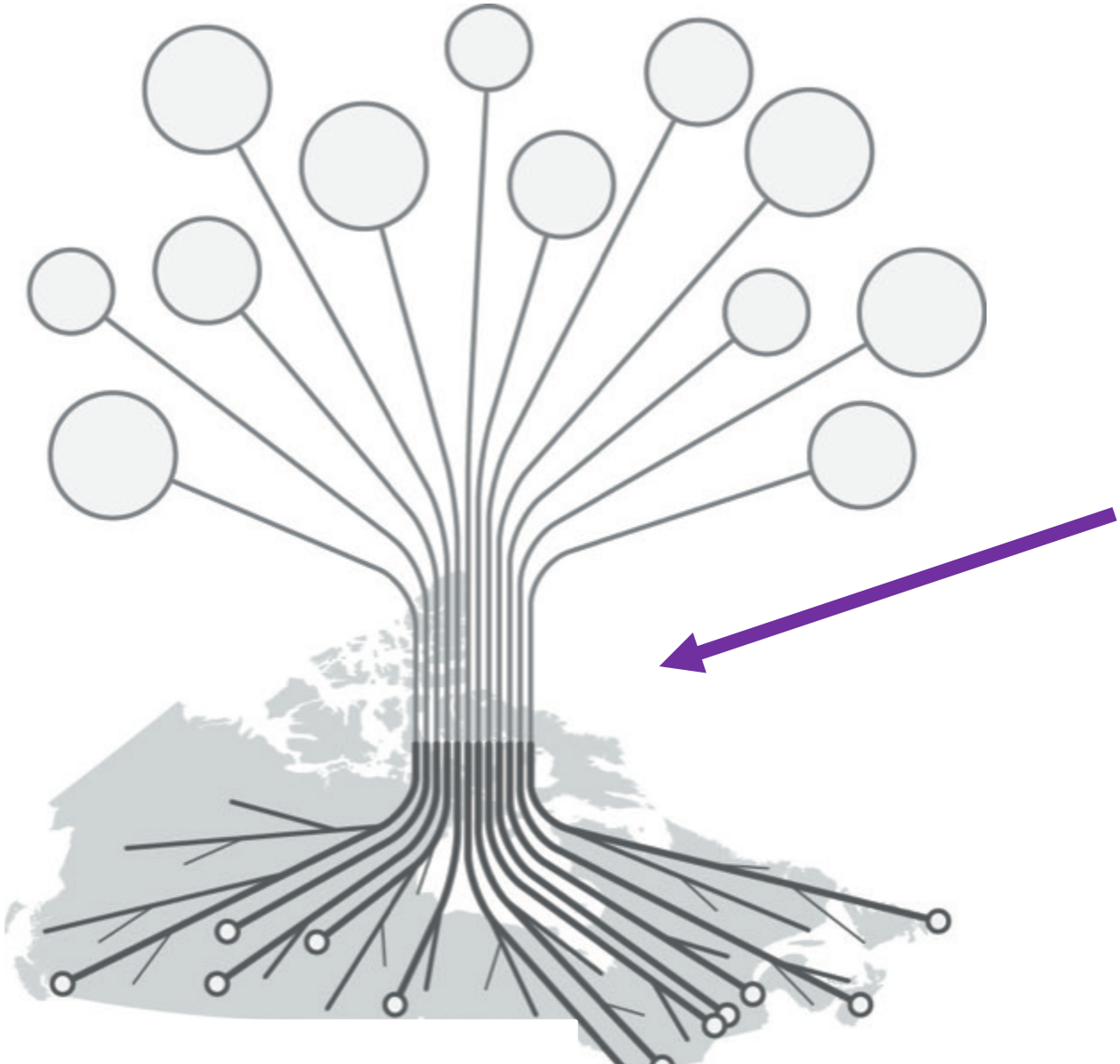
Matchmaker Exchange

Data from **>100,000** patients

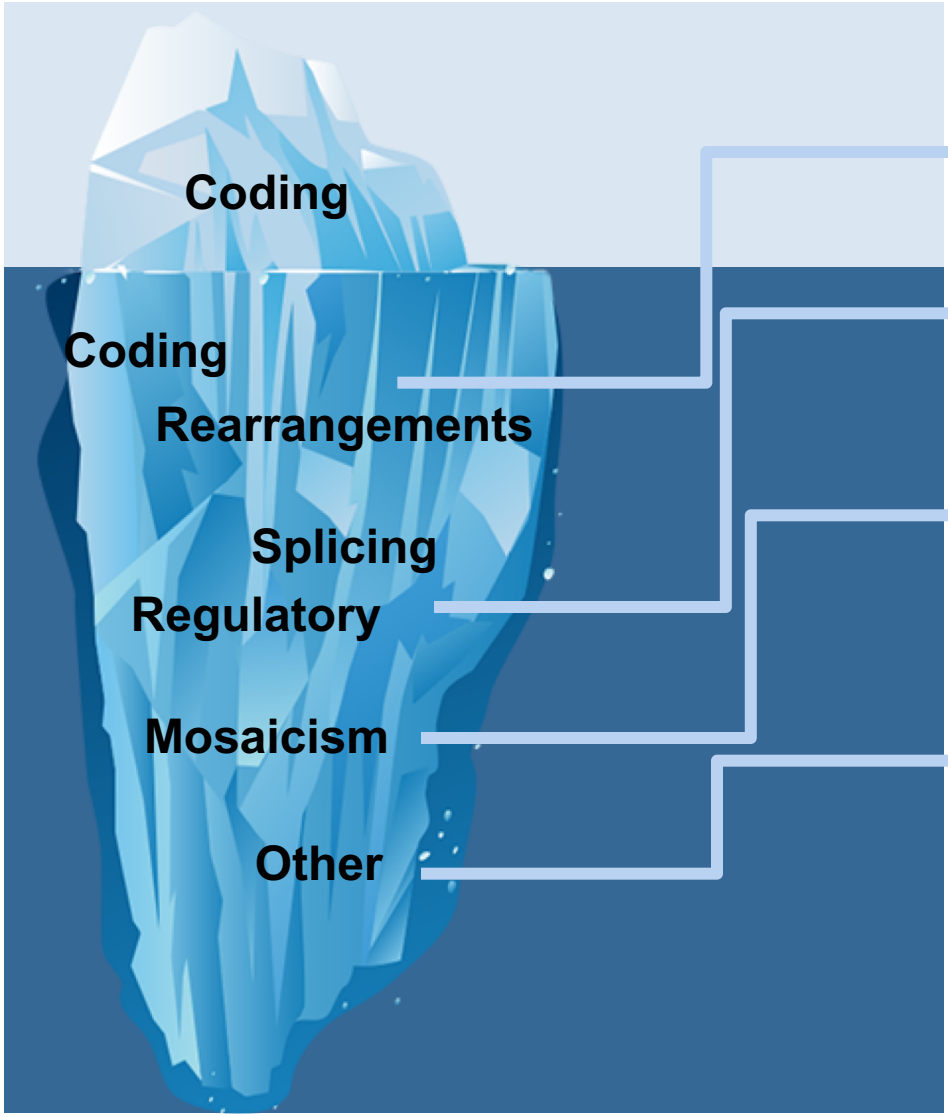
from **>4000** contributors

from **>84** countries

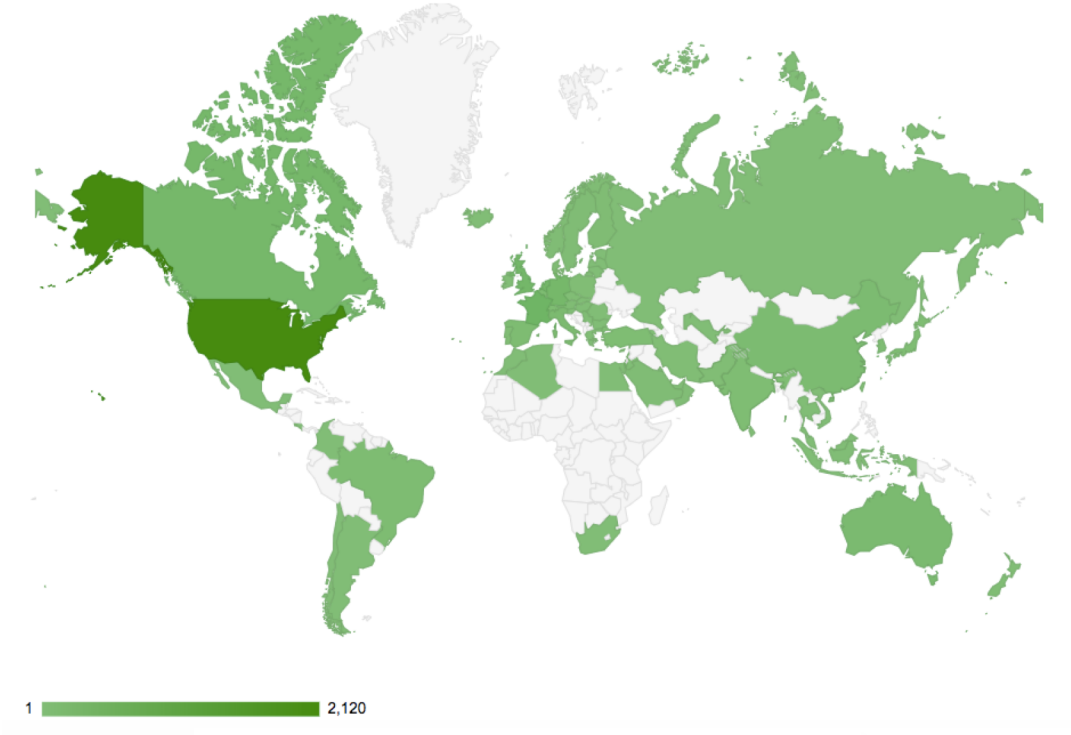
The opportunity



Emerging technologies



A means to diagnose most RDs



COMMENTARY

International Cooperation to Enable the Diagnosis of All Rare Genetic Diseases

Kym M. Boycott,[1,*] Ana Rath,[2] Jessica X. Chong,[3] Taila Hartley,[1] Fowzan S. Alkuraya,[4,5] Gareth Baynam,[6] Anthony J. Brookes,[7] Michael Brudno,[8] Angel Carracedo,[9] Johan T. den Dunnen,[10] Stephanie O.M. Dyke,[11] Xavier Estivill,[12,13] Jack Goldblatt,[6] Catherine Gonthier,[2] Stephen C. Groft,[14] Ivo Gut,[15] Ada Hamosh,[16] Philip Hieter,[17] Sophie Höhn,[2] Matthew E. Hurles,[18] Petra Kaufmann,[19] Bartha M. Knoppers,[11] Jeffrey P. Krischer,[20] Milan Macek, Jr.,[21] Gert Matthijs,[22] Annie Olry,[2] Samantha Parker,[23] Justin Paschall,[18] Anthony A. Philippakis,[24] Heidi L. Rehm,[24] Peter N. Robinson,[25,26] Pak-Chung Sham,[27] Rumen Stefanov,[28] Domenica Taruscio,[29] Divya Unni,[2] Megan R. Vanstone,[1] Feng Zhang,[30,31] Han Brunner,[32,33] Michael J. Bamshad,[3,34] and Hanns Lochmüller[35]

IRDiRC
INTERNATIONAL RARE DISEASES RESEARCH CONSORTIUM

Boycott at al., 2017: Am J Hum Genet 100; 695–705