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Sjældne Diagnoser

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The Danish Haemophilia Society

IML
Innovative Mindre Life Science Bolag

Timely Diagnosis Why does it matter?

A clinical perspective on timely diagnosis

Arvid Heiberg
Senior consultant, Prof (em)
Dept of medical genetics. Oslo University Hospital, Norway

Nordic Rare Disease Summit
12 April 2021 – Virtual Conference

Disclosures

- Participated in meetings in Norway and Europe arranged by various companies in the Rare Disorder field as Takeda/Shire, Sanofi-Genzyme, Alexion, Biomarin, Amicus and others.
- No honoraria.

Rare disorders

- Various definitions:
- EU 1 in 2000, Denmark 1 in 10 000.
- Sweden and Norway 1: 10 000, now switching to EU definition
- Estimated to be 7000 different possible genetic diagnoses, in practice 1500? - new entities coming each week
- 80% thought to be genetic and more than 50% affect children

Wright CF (2018) Nature Reviews ,doi:10.1038/nrg.2017.116

Rare disorders and severity

- Multisystem disorders affecting many organs and thereby »specialities«
- Phenotypically heterogeneous
- Responsible for 1/3 of deaths in first year of life
- 1/3 of children will not reach age 5 years

Wright CF (2018) Nature Reviews ,doi:10.1038/nrg.2017.116

Diagnosis

- Phenotype: Symptoms and signs obtained by medical history and physical examination
- Genotype: Obtained by laboratory techniques
- Sometimes easy- often difficult, time and procedure- and cost consuming.
- »Diagnostic Odyssé« taking many years

To what purpose?

What is in an accurate diagnosis

- Etiology: Why did it happen?
- Phenotype: What disorder is it-Information?
- Prognosis: What is going to happen?
- Complications: How to follow and what to be aware of?
- Can it be treated/cured?
- Will it happen again? - Implications for the extended familiy

Diagnosis

- Wrong diagnosis - sometimes very difficult to remove!
- Specter within diagnostic category: genotype- phenotype-correlations
- Exclusion of other diagnostic possibilities sometimes usefull

Periods in genetics

- Monogenic: Search for single genes: Until 2000, and later - Sanger sequencing
- Improved chromosome techniques: 1970-
- GWAS (Genome Wide Association Screening): 1995-2000 onwards
- Genome sequencing: 2010 onwards - various techniques

Genome sequencing

- From expensive to »cheap«
- Removes need for other diagnostic sometimes costly and »unpleasant«time consuming procedures as MRI(anesthesia) or muscle biopsy, serial hospital appointments etc
- Diagnostic yield 25-40% - depending on…

Impact on treatments

- Few, but important treatments p.t:
- Gene therapies increasingly promising:
  SMA (Nusinersen and Zolgensma)
- Blood disorders - (Sickle cell and Thalassemia, Hemophilia)
- Eye: Leber (LHON), Retinitis Pigmentosa,
- Immune diseases: SCID-ADA
- Enzyme replacement therapies

New Born Screening-NBS

- Norway: increased from 2 to 24 diagnoses (2012), now 26, including SCID- 200 genes in panel
- SMA to be added soon - HTA-Health Technology Assessment
- Genome sequencing in severely ill newborn children - panel of 800 genes
- 700 genes for immunodeficiencies
- Great importance for next child.
- Differences between Nordic countries

Management guidelines

- How to follow up?
- Screening for complications - sometimes very time consuming - Guidelines in Cassidy and Allanson - Syndromes
- Chromosome disorders: UNIQUE-www.rare chromosomes.org
- Contact with other parents: Local organisations for rare disorders, Facebook groups

Genetic counselling

- Am I to blame? (drinks, diet, occupation, various cultural health beliefs etc)
- Next child?
- Prenatal diagnosis?
- Extended family? - Inbreeding?
- Need for further knowledge - new visit?
- Recounselling for children when growing up?

Transition from clinical expertice to laboratory work on developmental disorders-A 50 year perspective:

A few well defined phenotypes: Cornelia de Lange, Noonan, Prader-Willi and Williams, but no molecular diagnosis

A few chromosomal disorders: Down, Turner, Cri du chat, trisomy 13 and 18.

Later: Angelmann , Rett, Fragile-X, Rubinstein-Taybi – but «iceberg concept»

Breaking Bad News

- Simple findings may imply serious prognosis
- Timing and setting - gradual process or all in one?

The Global Commission to End the Diagnostic Odyssey for Children with a Rare Disease

Clifford Goldsmith
US Chief Medical Officer at Microsoft

@clifgold

clifgold

350M affected by rare disease

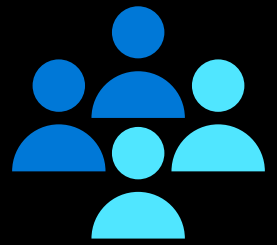
6–8yrs average time to be diagnosed

>40% are initially misdiagnosed

Background: Forming the Global Commission

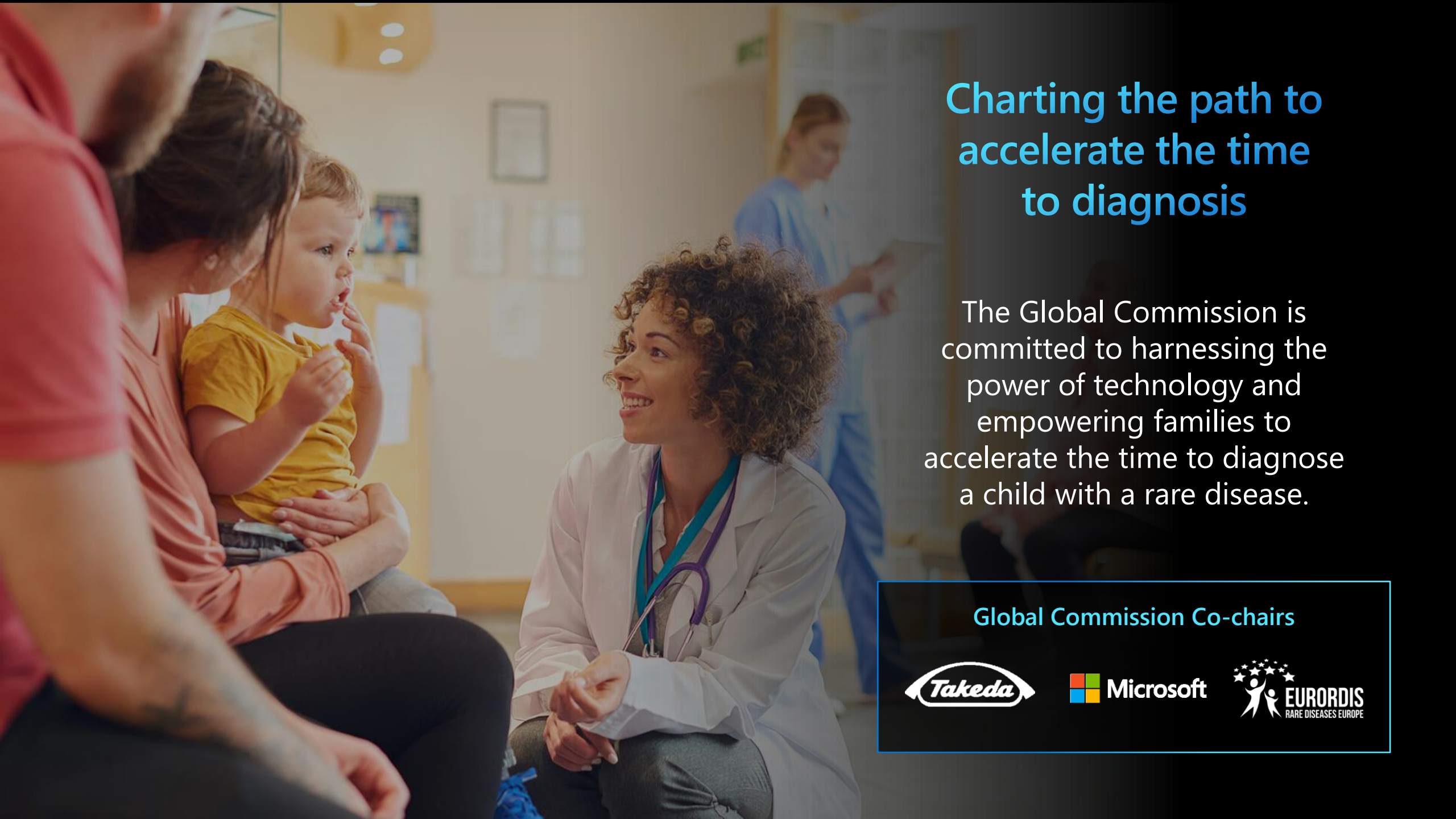
There are more than 6,000 known rare diseases worldwide. An estimated **300 million people are living with a rare disease,** with 50% typically occurring in children.

Today, it takes an **average of 5 years to get an accurate rare disease diagnosis,** even in countries with sophisticated health systems. Additionally, up to **40% of rare disease patients are misdiagnosed** more than once.



In 2018, Takeda, Microsoft and EURORDIS-Rare Diseases Europe **joined forces to launch the Global Commission** and bring the rare disease and technology communities together to solve this problem.

Our goal is to inspire concerted action and mobilize **diverse actors—within and outside the health field—**to work collaboratively toward a shared ambition.



Charting the path to accelerate the time to diagnosis

The Global Commission is committed to harnessing the power of technology and empowering families to accelerate the time to diagnose a child with a rare disease.

Global Commission Co-chairs

Takeda

Microsoft

EURORDIS
RARE DISEASES EUROPE

Global Commission Report Recommendations

Empower families to play an active role

Equip frontline providers with tools for diagnosis and referral

Reimagining the genetic consultation

GLOBAL POLICY RECOMMENDATIONS

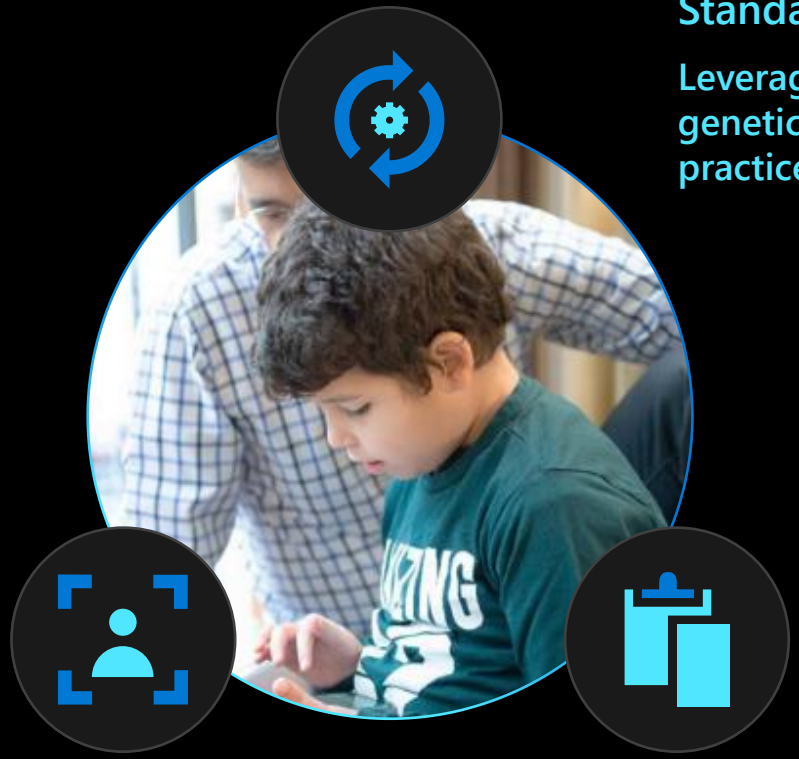
Where We Are Headed

The Global Commission aims to establish a clear path to a timely, accurate diagnosis for children around the world

Global Commission Key Priorities

GLOBAL COMMISSION
to End the Diagnostic Odyssey for Children with a Rare Disease

The **Global Commission's mission is to harness the power of technology and advances in genetics to accelerate the time to diagnose a rare disease.** The Global Commission also focuses on **two supporting areas**—family/caregiver empowerment and policy/advocacy—to ensure success.



Standards of Practice

Leverage advances in technology and genetics to develop **new standards of practice** for accelerating the time to diagnosis

Awareness & Empowerment

Empower families and caregivers to accelerate the diagnostic journey and **broaden awareness** about the urgent need to decrease the time to diagnose a rare disease

Policy & Advocacy

Advocate for local and global policy change to support an **enabling environment** for accelerating the time to **rare disease diagnosis**

Global Commission: A Clear Tie to Health Equity

- Access to comprehensive healthcare is important for promoting and maintaining health and preventing and managing diseases.
- However, in rural settings and for patients with a rare disease, the issues related to quality healthcare access are greater and more daunting.
- Challenges faced by patients with rare diseases range from the distance and time it takes to get to their HCP, to lack of access to specialists or specialist care for their disease.

The Opportunity: Ensure the Global Commission uses an equity lens in existing and future activations to prioritize access to rare disease diagnosis for even the most vulnerable and marginalized populations.

Global Commission and Technology Pilots

Global Commission Report Early Successes

Empower families to play an active role

EARLY SUCCESSES

Initial pilot launch in San Diego, U.S. in early 2021

Equip frontline providers with tools for diagnosis and referral

EARLY SUCCESSES

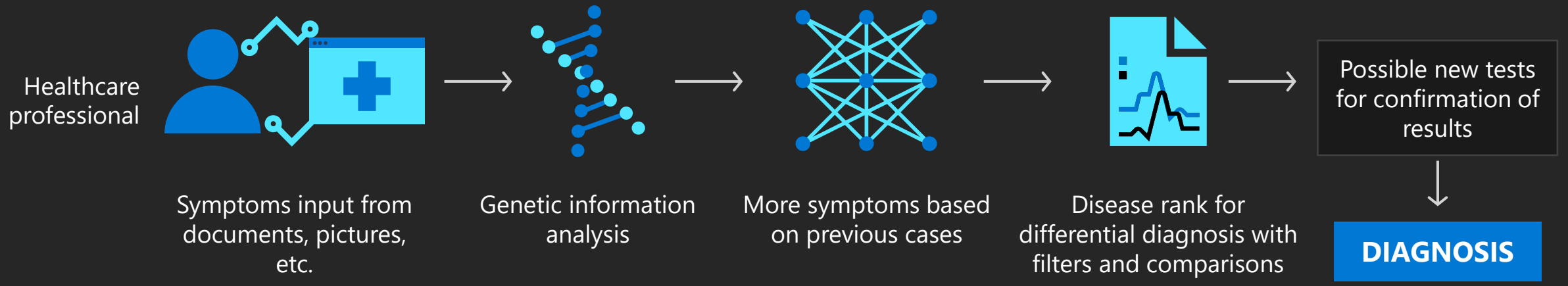
Dx29 correctly predicted diagnoses for **79%** of previously studied cases. The tool has been tested among **400** healthcare providers

Reimagining the genetic consultation

EARLY SUCCESSES

The average wait time for patients to see a clinical geneticist is estimated to **decline by 50%**

Phase 1: Dx29 for clinicians



Where is Dx29 being used?

380 users and 720 cases

Validation study in 3 healthcare systems

Global Commission Pilot 2: Genetics Opinion App

VISION

Leverage technology and communication tools to create a virtual collaboration platform that remotely connects genetic clinics and primary care physicians in order to rapidly deliver an initial opinion.

VALUE PROPOSITION: Faster times to diagnosis

PATIENTS

- Faster engagement with specialists
- Shorter wait times for full appointments
- Provides expert opinions

COMMUNITY PROVIDERS

- Better triage strategy for patients
- Identifies “urgent need” patients sooner
- Increases ease in obtaining an opinion
- Connects patients to the right specialist more quickly

SPECIALISTS

- Better triage of referrals
- Exposure to increased patient pool
- Higher value full visits
- More efficient use of limited clinical space

Inspiring Other Pilots (Example): Patient-reported phenotyping as a tool to bridge the inequity gap in rare disease diagnosis

CHALLENGE

Genome-wide sequencing (GWS) has become the best test for establishing a diagnosis for rare disease (RD) patients. However, GWS requires comprehensive description of a patient's signs and symptoms (**phenotyping**) to be properly interpreted. **RD patients are often experts in their own condition and** may be able to provide critical information for interpreting their GWS and ultimately improving the diagnostic rate for RDs.

SOLUTION

- **OBJECTIVE:** Evaluate patient-reported phenotyping as an approach to facilitate RD diagnosis from GWS data
- **PARTNER(S):** Care4Rare SOLVE, Takeda
- **PROGRAM DURATION:** 2020-2021
- **LOCATION:** Ottawa, Canada

STATUS UPDATE

Early Successes/Tasks Accomplished:

- Self phenotyping survey has been designed and hosted on RareConnect
- Survey has been piloted on RD patients, and feedback is currently being incorporated

Next Steps:

- Patient recruitment will begin once survey revisions are complete

Global Commission Patient Empowerment and Awareness Campaign

Patient Empowerment and Awareness Campaign

The challenge

Patients, caregivers, and frontline **providers often do not suspect that a patient may be suffering from a rare disease,** thus extending the diagnostic journey. Repeated visits to the emergency room, consultations with various specialists, and internet searches are "dead-end" paths common to many families. **Greater public awareness** of rare diseases and an **understanding of how to best engage** with health care providers is the **first step on the path to a quicker diagnosis.**

Our goal

The goal of the campaign is to **increase awareness of rare disease as a consideration** among parents and/or caregivers searching for a diagnosis for their child and **empower them to work with their physicians to find a diagnosis** for their child.

GLOBAL COMMISSION
to End the Diagnostic Odyssey for Children with a Rare Disease

RareNavigator: What it is

rare navigator

RareNavigator is an online, interactive guide to help parents and caregivers find the tools, resources and communities they need to get answers about their child's health and explore the possibility of a rare disease.

RareNavigator will generate a customized report with the following:

Tools
Discussion guides for parents, common signs of rare disease, links to third-party tools to educate and help track their child's health

Resources
Links to relevant third-party resources to help parents find answers based on where they are in their journey

Community
Links to connect parents with other parents and caregivers who may be on a similar journey, sharing advice, support and a sense of community

RareNavigator is being piloted in San Diego, Dublin and Perth in 2021

Policy & Advocacy

Global Policy Recommendations

Centers of Excellence

National healthcare systems should issue guidance on collaboration between prima care centers and centers of excellence to ensure consistent, effective and efficient diagnostic and referral protocols. The guidance should address coordinating ca laboratory resources, and knowledge sharing across country borders.

Data Sharing

To fully leverage the global benefit of cloud-based data storage – of particular value to countries with limited patient da (common in the case of rare disease) – health policies should encourage data sharing across borders to increase the likelihood of a match to determine a diagnosis.

Genetic Screening

As countries develop policies around genetic screening, these should incorporate next generation sequencing given its declining cost and potential to more quickly pinpoint a diagnosis, thus generating savings in unnecessary provide visits and diagnostic tests.

Privacy

In encouraging patients to provide medical and other information about their symptoms to help expedite diagnosis, it's critical that countries implement adequate privacy safeguards.

The Path Forward

The Path Forward

The Global Commission will strive to have a **far-reaching impact**—benefiting not only those **living with a rare disease and their caregivers,** but ultimately for all patients.

The **technological, scientific, and behavioral changes** the Global Commission hopes to generate are exactly what is needed to **improve patient-centered care** across health systems.

Learn More

GLOBAL COMMISSION

to End the Diagnostic Odyssey for Children with a Rare Disease

Website: https://www.globalrarediseasecommission.com/
Email: questions@globalrarediseasecommission.com

Thank you!

Clifford Goldsmith
US Chief Medical Officer at Microsoft

@clifgold

clifgold

Appendix

The Global Commission is Conducting Three Pilots

PILOT 1

Multifactorial machine learning to recognize symptom patterns

PILOT 2

Genetics Opinion App

PILOT 3

Patient Empowerment and Awareness Pilot Campaign

rare navigator

Cross-Section of Leaders to Find Solutions

Yann Le Cam

EURORDIS RARE DISEASES EUROPE

Wolfram Nothaft

Takeda

Gregory Moore

Microsoft

Moeen AlSayed

Gareth Baynam

IRDiRC

Kym Boycott

CHEO RESEARCH INSTITUTE

Pamela Gavin

NORD National Organization for Rare Disorders

Roberto Giugliani

UFRGS UNIVERSIDADE FEDERAL DO RIO GRANDE DO SUL

Kevin Huang

Derralynn Hughes

Royal Free London NHS Foundation Trust

Maryam Mohd. Fatima Matar

دبي العطاء Dubai Cares

Dau-Ming Niu

Anne O'Donnell-Luria

Boston Children's Hospital Until every child is well

BROAD INSTITUTE

Mike Porath

The MIGHTY

Richard Scott

Genomics england

Marshall Summer

Children's National

Durhane Wong-Rieger

CORD Canadian Organization for Rare Disorders

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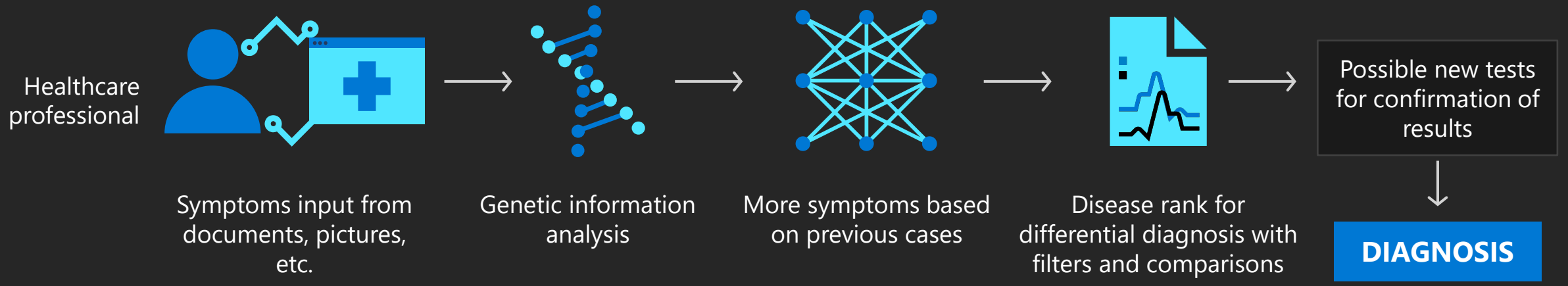

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Global Commission Report Recommendations

Empower families to play an active role

OBJECTIVE: Develop tools that empower families and caregivers to become more proactive in getting a diagnosis as quickly as possible.

Equip frontline providers with tools for diagnosis and referral

OBJECTIVE: Equip frontline providers with the knowledge and tools to quickly and effectively identify patients who may have a rare disease and take appropriate action.

Reimagining the genetic consultation

OBJECTIVE: Develop innovative ways to enable medical geneticists to see priority patients more quickly—especially given the growing shortage of geneticists.

GLOBAL POLICY RECOMMENDATIONS

Policy guidance at a global level that can be adapted to meet differentiated regional needs and work with national and local governments

GLOBAL COMMISSION to End the Diagnostic Odyssey for Children with a Rare Disease

Background: Forming the Global Commission

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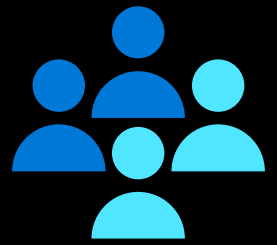

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Pilot 2: Genetics Opinion App

PROBLEM STATEMENT

There are several barriers hindering early rare disease diagnoses for children, notably the severe shortage of Clinical Geneticists and Centers. Additionally, HCPs/PCPs lack the right pre-screening tools which is a significant time/cost burden for patients and parents. These barriers contribute to the average 5+ year delay in diagnosis for children.

PILOT OVERVIEW

PROGRAM DURATION: 3+ years (2019-2022)

PARTNER: Children's National Hospital, Washington, DC (CNMC-DC)

OBJECTIVE: Leverage technology and communication tools to create a virtual collaboration platform that remotely connects genetic clinics and primary care physicians in order to rapidly deliver an initial opinion.

ABOUT THE APP

GENETICS OPINION APP (BETTER REFERRAL)

- Allows pediatricians to submit a referral to a specialist electronically
- Patient information is entered on a child (e.g., prenatal history, birth history, family history) including pictures, images or files)
- By providing the necessary information needed *upfront,* specialists are able to turn around a pre-diagnosis much faster

Genetics Opinion App

Enabling collaboration tool for Primary Care Physicians/Pediatricians/GPs with Specialists

GENETICS OPINION APP

Solution aims to reduce bottlenecks at Genetics centers by providing opinion via the app

App provides an easy and intuitive way to connect pediatricians with rare disease specialists (Clinical Geneticists)

The app is currently hosted by NORD (National Organization for Rare Disorders, in the US)

APPROACH

Pediatrician answers genetic questionnaire and can upload further information

Specialists determines next steps based on data (do nothing, refer, get more tests)

Currently collaborating with Children's National Hospital (Washington, DC, US)

Potential to expand to other English-speaking countries and beyond after the testing and enhancing phase

DELIVERABLES

RDO App: A Pediatrician's Electronic Referral for Children with Possible Rare Diseases

Website: www.rarediseaseconsults.com

FAQs, user manual, quick reference guides, patient/ caregiver handout, value proposition for Primary Care and Clinical Geneticists

Creative:

Visually compelling materials to support surround-sound approach




Innovative technologies and the future of diagnostics

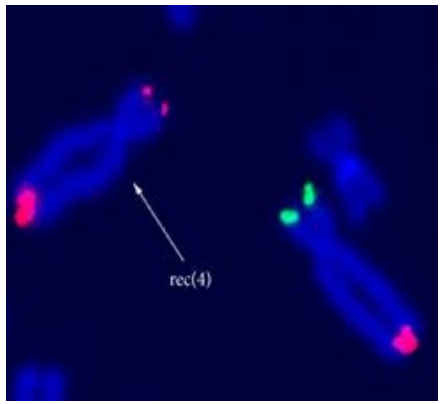
Helena Kääriäinen

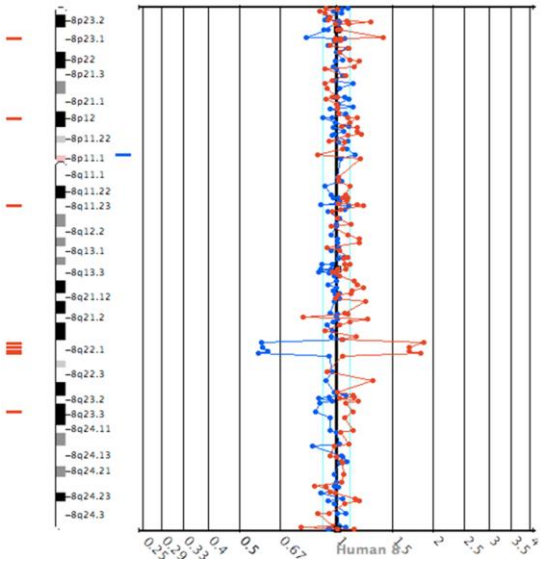
Finnish Institute for Health and Welfare

- Affiliations/Conflict of Interest
  - Norio - Centre of Rare Diseases (consultant)
  - Finnish Institute for Health and Welfare (research professor)
  - Blueprint Genetics Laboratory (consultant)

Picture: Norio Centre

Innovative technologies during my career





Innovative technologies during my career

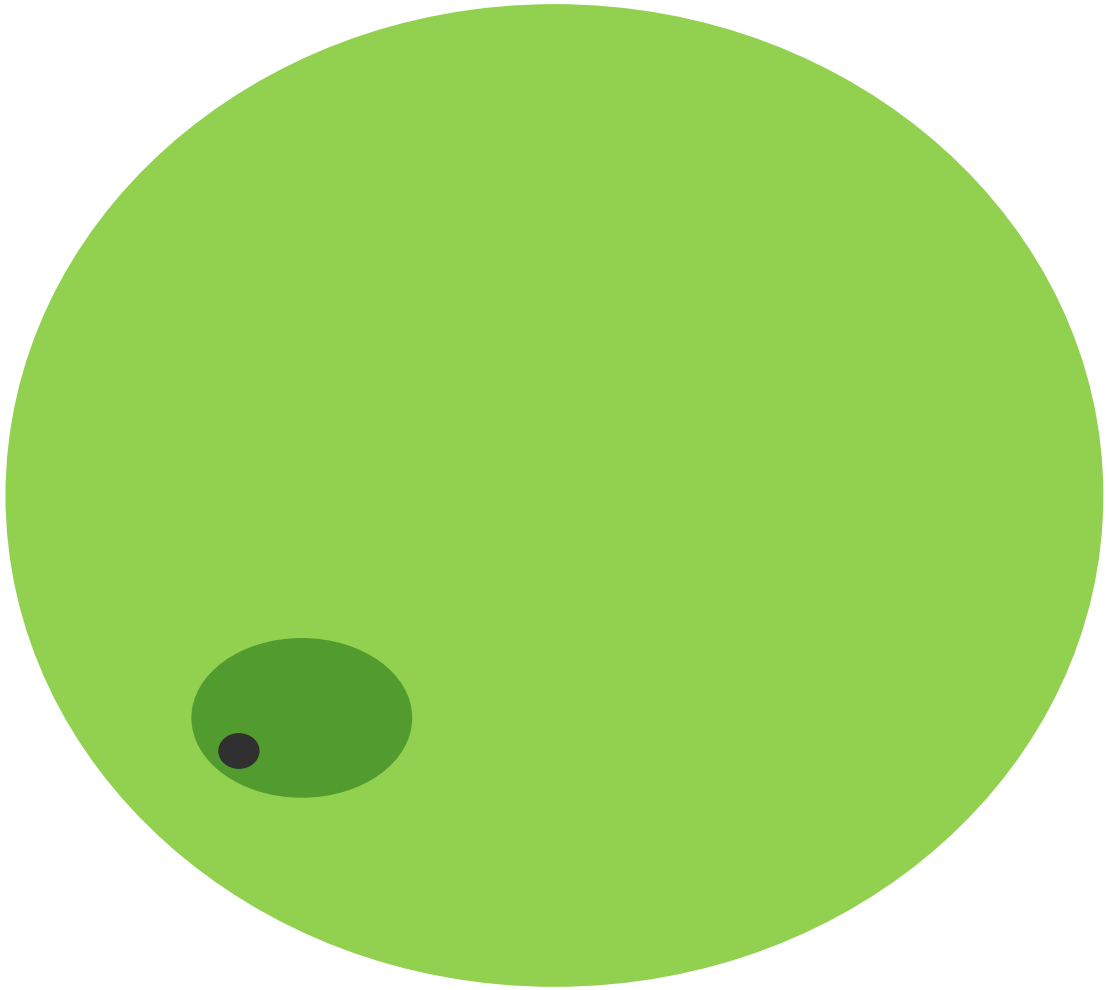
- Diagnostics based on linkage studies
- Detecting known mutations
- Sequencing the gene(s) of interest
- Next generation sequencing
  - Genes, gene panels,exomes (= all (known) genes)
- Whole genome sequencing
  - Also areas between genes, copy number variations, balanced translocations
- Cell-free DNA

What is found in the exome study?

Some 20 000 – 50 000 variants detected;
95% can be ruled out by automatic algorithms

700 – 1000 variants left;
Most can be ruled out (nearly) automatically

Some 100-200 variants left;
These will be judged individually



Classification of variants

- Pathogenic = known to cause the disease
- Likely pathogenic = most likely causes the disease
- Variant of unknown significance (VUS) = impossible to judge with the present knowledge
- Likely benign = most likely harmless
- Benign = known to be harmless

Classification of variants

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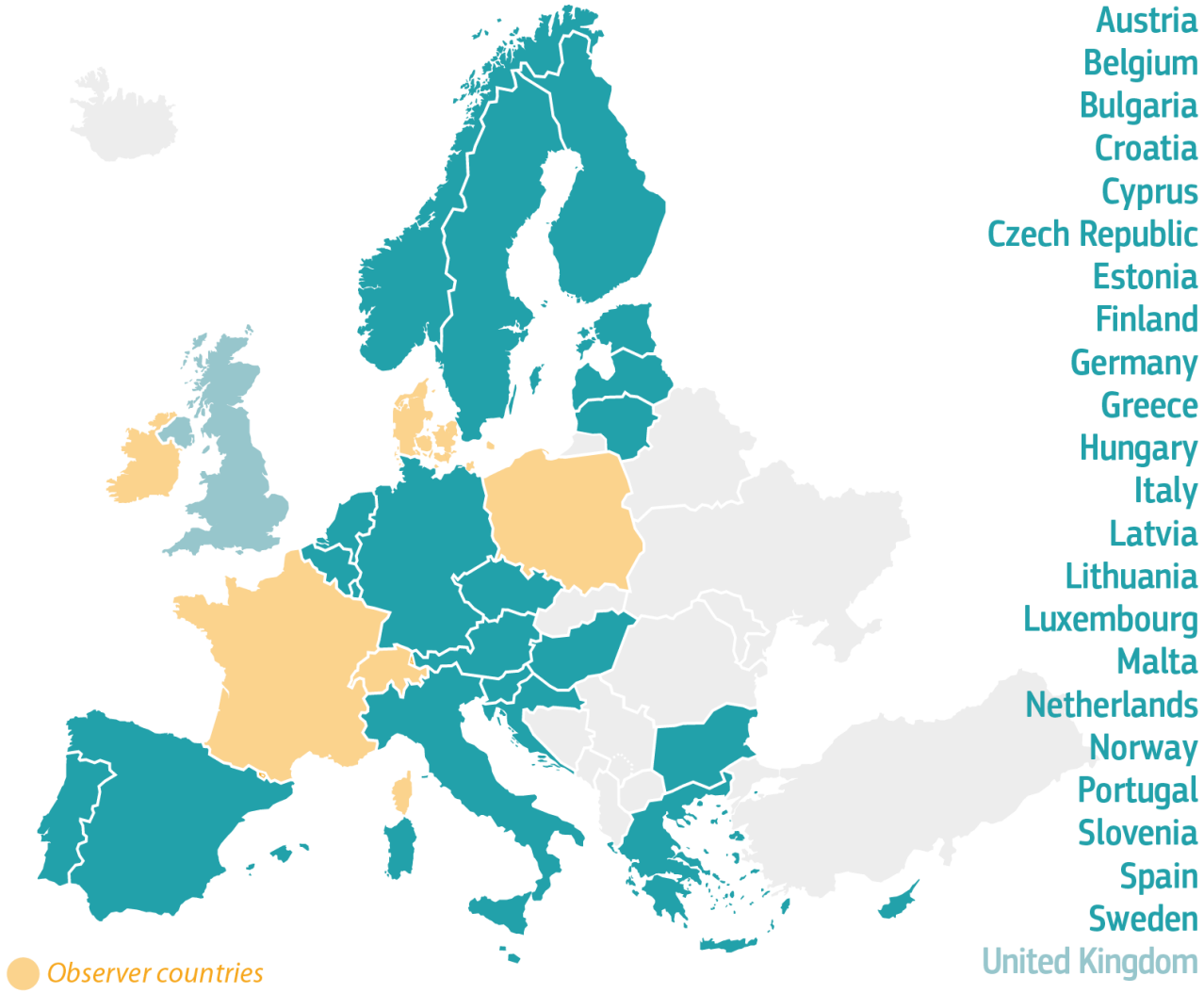
VUS-variants are a difficult problem

- For the laboratory:
  - to report or not?
- For the doctor:
  - to believe or continue diagnostic work?
- For the patient and family:
  - to react relating to family planning, to inform relatives?

How to overcome VUS-problem?

- Better, detailed referrals to the lab
- Regular reporting of such variants to the relevant databases
  - By the lab? Or by the clinician?
- To collect and share more genomic data from population cohorts, biobanks etc.
  - E.g. “1+ Million Genomes” initiative

Countries that have signed the 1+MG Declaration since 2018



Austria
Belgium
Bulgaria
Croatia
Cyprus
Czech Republic
Estonia
Finland
Germany
Greece
Hungary
Italy
Latvia
Lithuania
Luxembourg
Malta
Netherlands
Norway
Portugal
Slovenia
Spain
Sweden
United Kingdom

updated on 03 Decembre 2020

Whole genome sequencing?

- Interpreting the results is still very difficult
  - Whole genome sequencing creates much more data (and also much more variants to judge)
  - The reference data is still unsufficient

The future of diagnostics

- We will know more about normal variation
- We will understand more about the consequences of different variants
  - This improves both exome and whole genome diagnostics
- The diagnostics will be quicker and cheaper
- ”Genome first” –thinking?

Summary

- RD diagnostics have improved in an unprecedented way!
- The costs of diagnostics have remained reasonable (when compared with other diagnostic tools).
- In spite of this, still about half of the patients who most likely have a genetic RD remain without exact diagnosis.
- The future developments are difficult to predict.

C-ANPROM/FI/RDG/0001

thl

THE IMPORTANCE OF EMPOWERMENT FOR PATIENTS, RELATIVES AND SOCIETY

Nordic Rare Disease Summit

2021

Terkel Andersen

Eurordis – Rare Diseases Europe

EURORDIS.ORG

C-ANPROM/DK/TAKH/0013

"EURORDIS recommends that the EC and all European countries implement specific mechanisms that empower people living with a rare disease and their carers, in co-creation and co-delivery with organisations representing people living with a rare disease:

- Opportunities to foster peer-to-peer support **between people living with a rare disease** should be available and supported. " *

What is "patient empowerment"?
What are the roots of the concept?
Why insist on "empowerment" in the context of PLWRD?
What does it mean in practice and how does it play out on different levels?
What is the value creation in pt. empowerment among PLWRD?

*) EURORDIS Position paper on "Achieving Holistic Person-Centred Care to Leave No One Behind"
Full paper at www.eurordis.org/carepaper

Patient empowerment:

Reaffirms autonomy and integrity at individual (micro) and community (macro) level

Connects individual and collective experience and action

Allows to voice community views and interest with other stakeholders and to influence a policy level

A dynamic phenomenon

Driver of innovative change and transformation

Patient empowerment (2):

Can be a process and an outcome

Can be achieved at individual, community and policy level

Plays out in the medical realm and outside of it (everyday life).

Is liberating and transformative

Provides opportunities for value creation for all stakeholders

Roots of patient empowerment:

Often linked to the AIDS activism of the 1980's:

- Strong advocacy groups to fight discrimination, support prevention efforts and work for accelerating research, therapy development and shortening the road to access

But other patient communities had already embarked on this journey

High social impact and marginalisation

Rare diseases are chronic, progressive, degenerative, disabling and frequently life-threatening

- Patients are few, geographically scattered and therefore often isolated
- Patients are sometimes undiagnosed, misdiagnosed or wait years for a diagnosis
- Are sometimes met with misunderstanding and prejudice leading to stigmatisation
- Often experience difficulties to find and interpret available information
- Have to face challenges to access adequate care
- Heavy psychosocial burden: Families can find it hard to cope with the consequences, uncertainties and prognosis of a rare disease and not always meet adequate societal understanding and support

-> Need to find ways to regain control

To regain a level of “control”

(Empowerment at individual/micro level)

Acquire the skills / ability to

Achieve insights and knowledge enabling you to manage / ease the impact of illness (physical, emotional and social) and develop coping strategies

- obtain sufficient understanding (health literacy) to make informed choices e.g. about treatment options
- (co-)design as well as negotiate workable solutions for you / your child
- learn to navigate health care and social systems
- gain confidence to become “co-managers” of your condition in partnership with health professionals / caregivers

Mutual support
(Empowerment at community/macro level)

Develop common resources and structures to

- generate collective learning, knowledge and experience sharing
- organize and sustain peer to peer support
- transform tacit knowledge into shared information (e.g. through surveys)
- create adequate services (e.g. helplines)

Awareness raising and advocacy
(Empowerment at policy level)

Engage with mutually agreed positions and ensure representation to

- cocreate solutions, “asks” and policies
- mobilize resources, e.g. for campaigning or research
- speak with a common voice, for advocacy and partnering: Influence priorities
- obtain representation on relevant boards etc.
- reach out to other stakeholders

Nothing about me without me

- **Patient empowerment**
  - Community building & networking
  - Capacity building
- **Patient engagement**
  - In research
  - In therapy development
  - In health care
  - In social care
- **Patient advocacy and representation**
  - Influencing priorities
  - Patient-perspective included in policy making

Value creation through patient empowerment – dynamic examples:

Individual/micro level: Joining a group of RareConnect.org to learn about ways to manage the disease -> helps establish a knowledge base

Community/macro level: Setting up Community Advisory Boards to agree patient relevant outcomes in clinical trials, advice on design of trials -> speeds up clinical development

Society (policy) level: Rare Disease Day -> Developing a more favourable structural, legal and economic framework through awareness raising


#RareDiseaseDay
rarediseaseday.org
RARE IS
MANY
RARE DISEASE DAY®
29 FEBRUARY 2020


#RareDiseaseDay
rarediseaseday.org
RARE IS
STRONG
RARE DISEASE DAY®
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#RareDiseaseDay
rarediseaseday.org
RARE IS
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RARE DISEASE DAY®
29 FEBRUARY 2020

Experiences of Web-based adaptation training for people with rare diseases

Sinikka Hiekkala, Adj. Prof. in Neurorehabilitation, PhD in Neuroscience, MSc in Health Science, PT

Research Director in the Finnish Association of People with Physical Disabilities

Web-based adaptation training for the target groups of the associations (2018-2021)

The purpose of the project was to develop and research web-based adaptation training, which was carried out by 12 organizations in collaboration

Leading: The Finnish Associaton of People with Physical Disabilities (FPD)

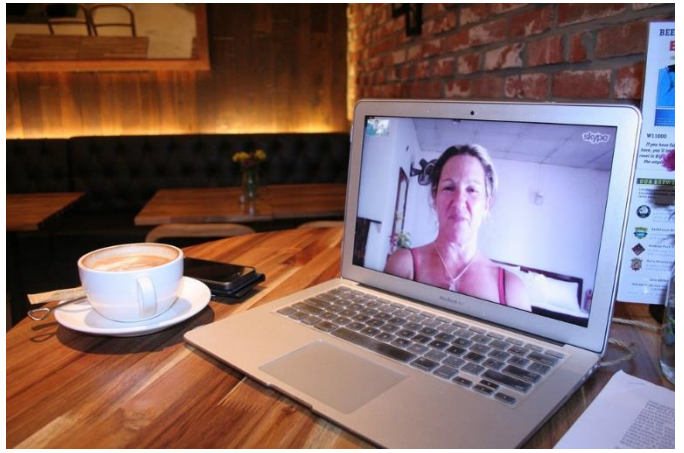
Funding: The Funding Centre for Social Welfare and Health Organisations (STEA), which operates in connection with the Ministry of Social Affairs and Health.

The aim of the courses was to produce well-being for the people involved

- Web-based group meetings
- 2 supervisors and a peer
- Courses aimed at people with a longterm disease or disability
- Implementation of courses across diagnostic boundaries
- Course duration approx. 2 months. The courses have two different implementation methods
  - 8 web-based groups meetings (2 hours once a week)
  - A face-to-face one-day meeting at start + 6 web-based groups meetings (2 hours once a week)

➔ in addition, for both course types, a follow-up meeting online 5 months after the first meeting (2 hours)

Group meetings



Applying for courses through the project website

Participant-specific conversation with the supervisor

Skype for Business and Teams for group meetings

Course platform hyväkysymys.fi –service for tasks and chat (hyvakysymys.fi/en/)

Goal Attainment Scale (GAS)

Service design produced 4 themes – 12 courses were performed

Rohkeasti mä!
(Boldly me!)

- 2 courses

Tukea työelämässä jaksamiseen
(Support for coping in working life)

- 4 courses

Keinoja omaan hyvinvointiin
(Ways to your own well-being)

- 3 courses

Harvinaisena joukossa
(Rare among)

- 3 courses

Woman, 43 years, rare disease

Peer support can be hard to find, as usually no one has heard of my disease

I was already looking forward to hearing from others and being able to laugh together at even difficult things

The implementation method suited me well, because sometimes it is difficult to leave home, but on the computer I can sit enough

I got quite a huge amount of energy for my everyday life, and I started to implement them independently. Definitely apply for a new course if the opportunity arises.

We talked about everyday life and future plans. We had a lot of good conversations that shed useful information that might not be found out elsewhere

Kuusenmäki (ed.) 2019. Tukea saa kotisohvalle. IT Invalidiliitto-lehti 2019:79(3).

Man, 50 years, rare disease

Yes, it strangely makes it easier for someone else to know this little concern

Yes, this moment goes before the blocks are back in the box, but yes it put its own values in order when you heard the stories of the other course participants and learned from their experiences

Kuusenmäki (ed.) 2019

Woman, 75 years, rare disease

I was able to attend the course from home - wool socks on foot and a comfortable seat on my own couch

Using Skype is not a barrier to participation. It's like a phone from which you can click another face to appear if you want

My physical condition didn't actually improve, but self-acceptance increased. I was able to better define the philosophical foundation for myself

The teachers were absolutely awesome. They had the job in their possession all the time, they helped with technical problems and encouraged us to continue

Kuusenmäki (ed.) 2019

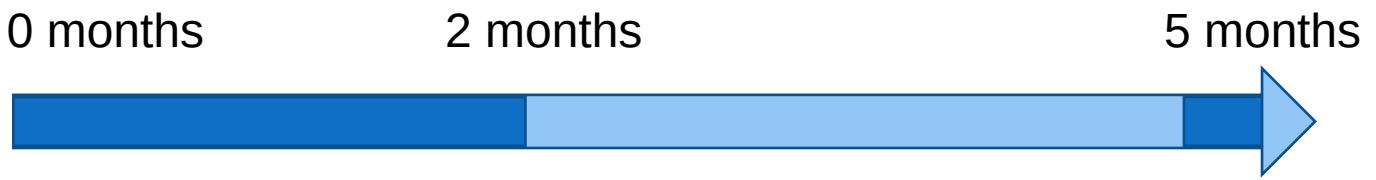
Outcomes

PROMIS®, Patient-Reported Outcomes Measurement Information System (healthmeasures.net):

- PROMIS Global Health – physical and mental health
- PROMIS Sleep disturbances short form 4a
- PROMIS Anxiety short form 4a
- PROMIS Depression short for 4a

Rosenberg Self-Esteem Scale (Rosenberg 1965)

Goal Attainment Scaling (Kiresuk & Sherman 1968)



Participants in all 12 courses

65 signed consents (9 men)
Outcomes from 44 participants (5 men)
Mean age 47.1 (SD 14.1, range 23-75)

The goals had been achieved on average at the target level:
Mean GAS T-Score 49.7, range 31-79

Pietilä et al. Etäsopeutumisvalmennuksella myönteisiä vaikutuksia koettuun hyvinvointiin (Remote adaptation coaching has a positive effect on perceived well-being). In Savinainen et al. (eds). Kuntoutus työelämän käännekohdassa - rehabilitation at a turning point in work life. Helsinki, Työterveyslaitos/Finnish Institute of Occupational Health, 2021, p. 55-56.

Changes in welfare

There were positive changes in the overall experience of health

Physical, $p = 0.016$, $n = 37$ and

Mental, $p = 0.025$, $n = 39$,

Anxiety, $p = 0.018$, $n = 39$,

Depression, $p < 0.001$, $n = 39$ and

Self-esteem, $p = 0.002$, $n = 39$

All of the positive changes were maintained for the follow-up. The perceived sleep difficulties were also noticeable parallel development, but the change was not statistically significant ($p = 0.094$, $n = 39$)

Pietilä et al. 2021

Summary of Web-based adaptation training

Service design produced 4 thematic web-based adaptation training
Patients with rare diseases had good experiences about the course ”Rare among”
Helped all participants to achvieve personal goals
Improved physical and mental health and self-esteem

New products need a lot of marketing communication

Thank you!

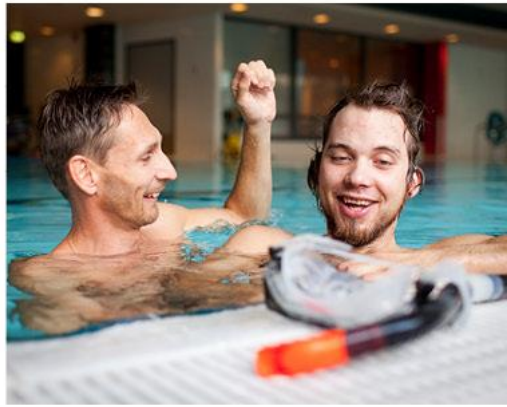
Sinikka.hiekkala@invalidiliitto.fi











INVALIDI LIITTO

Involved in creating an equal and accessible Finland

Sjældne Diagnoser

Rare Diseases Denmark: our Helpline in a societal perspective

Birthe Byskov Holm, President, Rare Diseases Denmark

WHY DOES RARE DISEASES DENMARK HOST A HELPLINE?

- **Because it is much needed**

Sjældne Diagnoser

WHY DID WE TAKE THE MATTER INTO OUR OWN HANDS?

- **Because it is an obvious solution**

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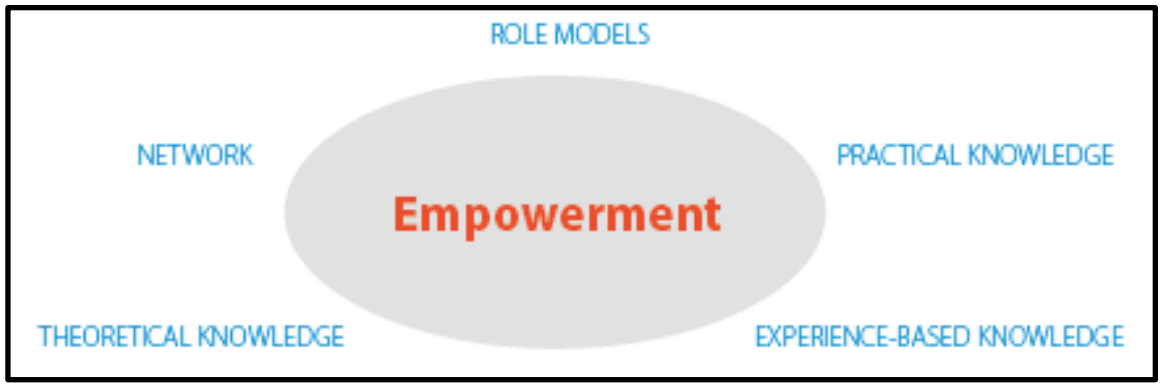
WHAT DOES HELPLINE DO?

- **Provides information, coping support and individual counseling**
- **Empowerment:**



- **Acknowledgement as an important factor**

HOW ABOUT VOLUME?

- **Initial dimension: 200 – 400 inquiries per year**
- **Actual dimension: 2.187 inquiries the first four years – 3.202 contacts**
- **418 different rare diagnosis**
- **At least 75 pct. satisfied or very satisfied, at leas 75 pct. empowered**
- **Evaluation 2020/2021:**

  https://sjaeldnediagnoser.dk/ja-helpline-opfylder-sit-formaal/

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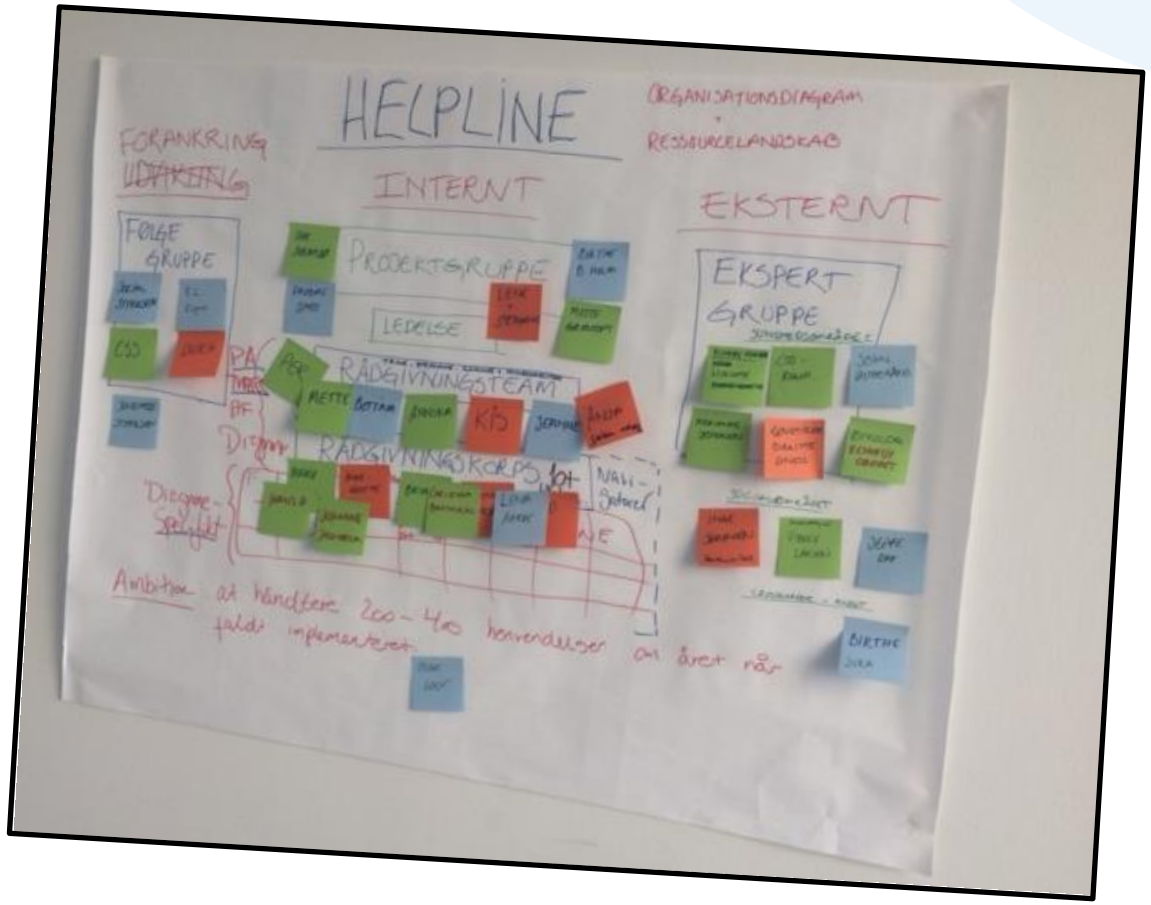
WHAT MAKES HELPLINE SPECIAL?

- **Help-to-self-help**
- **Multi disciplinarity, cross sectoral**
- **Builds up capacity**



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HOW DOES HELPLINE BUILD UP CAPACITY?

- **Helpline supports the empowerment of rare people**
- **Helpline built bridges**
- **Helpline create and share new documentation**

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IN CONCLUSION

- **Helpline is a recommendation of the Danish National Strategy of Rare Diseases**
- **Through Helpline, Rare Diseases Denmark provides a service of societal importance**
- **Helpline is much needed – wish us luck!**

Sjældne Diagnoser

Takeda

Presentation in accordance to the Nordic Rare Disease Summit held by Takeda Pharma AB

Presentation by Helene Cederroth, Founder and President of The Wilhelm Foundation

Takeda pays, in accordance with LER, lecturer fees

TAKEDA PHARMA AB

Contact: 08 731 28 00

Link: www.takeda.com

E-mail: infosweden@takeda.com

C-ANPROM/SE/RDG/0004 apr-21

The Undiagnosed diseases

An Undiagnosed disease is like a nightmare

The Undiagnosed diseases

No tests or exams match any disease that is known today

Why a diagnosis is important

- To understand the disease
- Know if a treatment works or makes it worse
- Have a prognosis
- Know if it’s a genetic disease


Without a diagnosis
relatives, friends, caregivers
often mistrust the family

In the worst-case scenario, a late diagnosis could lead to irreversible consequences and even be life threatening

The odyssey to reach an accurate diagnosis can take years

Undiagnosed diseases are divided into 3 groups:

1. Not yet diagnosed – but should be since they suffer from a known rare disease

2. Undiagnosed - since the disease is not discovered

3. Misdiagnosed

Patient Journey through diagnosis

> *"It's a waiting game, but you tell a mum to wait when she's waited 15 years. It's difficult. – Nuria*

> *"People began to ask which side of the family it came from...It was a difficult time for us as parents. – Alexa*

> *"A diagnosis may be bad news, it may be very bad news or it may be no news. But all of that's OK and there's help and support for whatever spectrum you end up on. – Peter*

> *"We went around, travelling across the entire city to find a nursery for our son. It was impossible to have him accepted. – Gaston*

Information and training for patient organizations

Helplines

Information on secondary findings

Next Steps Toolkit

EUROGENTEST Guidelines

Experience Based Co-Design

RareConnect .org

ENSERIO study. Time to Diagnosis

Undiagnosed Photo Project

Protocol to support ultra-rare diagnosis

Training for professionals

EURORDIS

SolveRD

CHEO

European Patient Advocacy Group

EuroGentest

GLOBAL COMMISSION

UNIAMO

WILHELM FOUNDATION

feder

SWAN

UDNI

Undiagnosed diseases are all around the globe

The variants don’t respect national borders.

Geneticists and researchers from all different parts of the world must collaborate to solve the undiagnosed diseases.

International Conferences on Undiagnosed Diseases

Undiagnosed Diseases Network INTERNATIONAL

Mission to share and collaborate

udninternational.org

UDNI Patient Engagement Group

Finding Answers through Sharing

Name and gender *(Only first name displayed. Max 70 characters)*	
Year of birth *(Max 4 characters)*	
Description - your Undiagnosed disease *(Max 1500 characters)*	
Symptoms/Signs *(Max 800 characters)*	
Current Treatments *(Max 300 characters)*	
Prior Treatments *(Max 300 characters)*	
Considered treatments *(Max 300 characters)*	
Previously Considered Diagnoses *(Max 300 characters)*	
Photo	Please send one photo of your child so we pair the name with Rick Guidotti's photos.
Genetic Variants of Interest *(Max 250 characters)*	
Bio *for public exhibitions without medical information. we want short description of the undiagnosed* *(Max 350 characters)*	
Siblings, age & gender *(Max 250 characters)*	
Contact information *Never displayed. We will contact you if there is an interested doctor.* *(Max 150 characters)*	

Please, send the form and the photo to Wilhelm Foundation info@wilhelmfoundation.org

Wilhelm Foundation - the Undiagnosed
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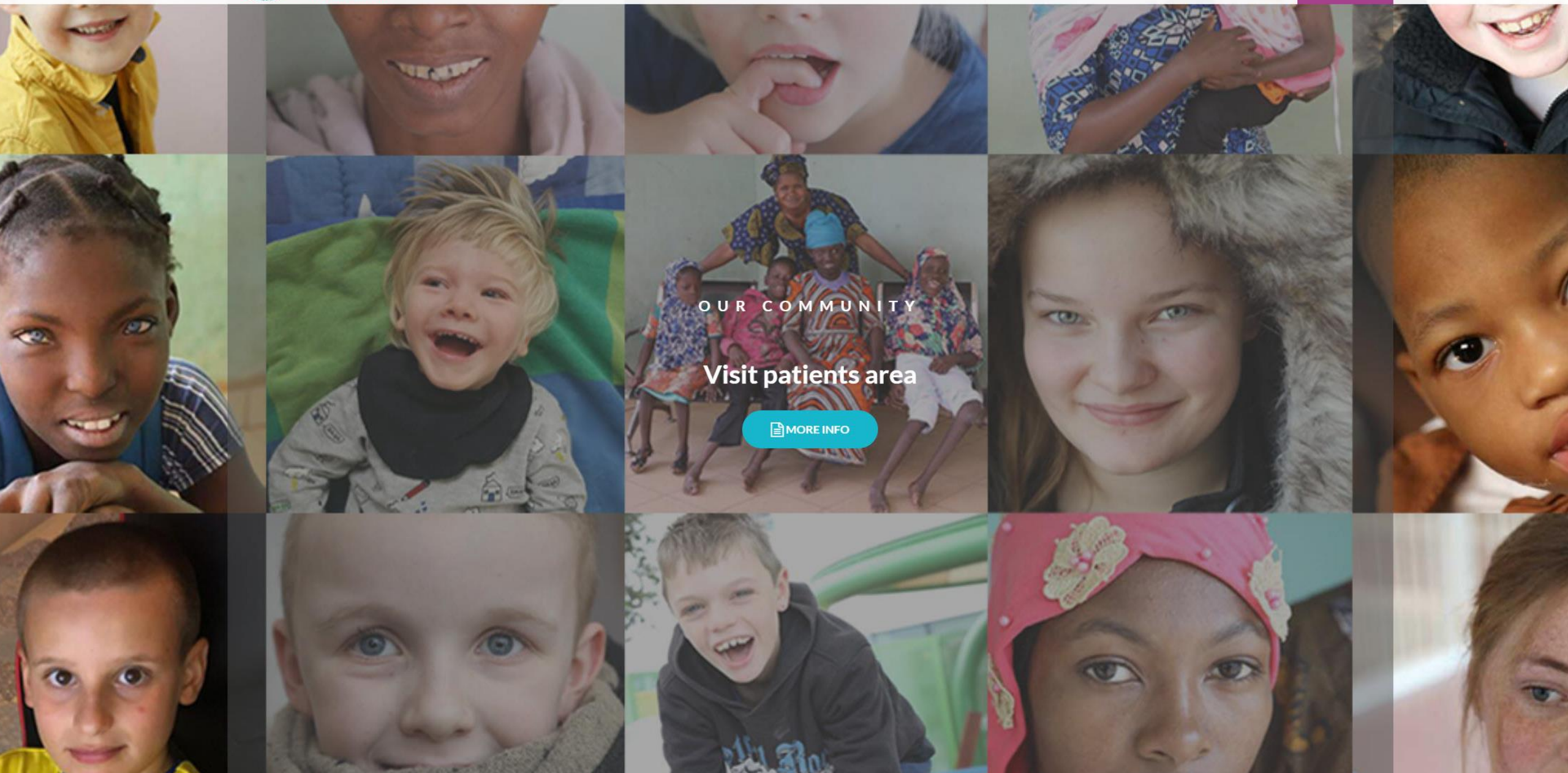
www.wilhelmfoundation.org • shop.wilhelmfoundation.org
www.udinternational.org

Takeda

UDNI
Undiagnosed Diseases Network INTERNATIONAL
ISTITUTO SUPERIORE DI SANITÀ

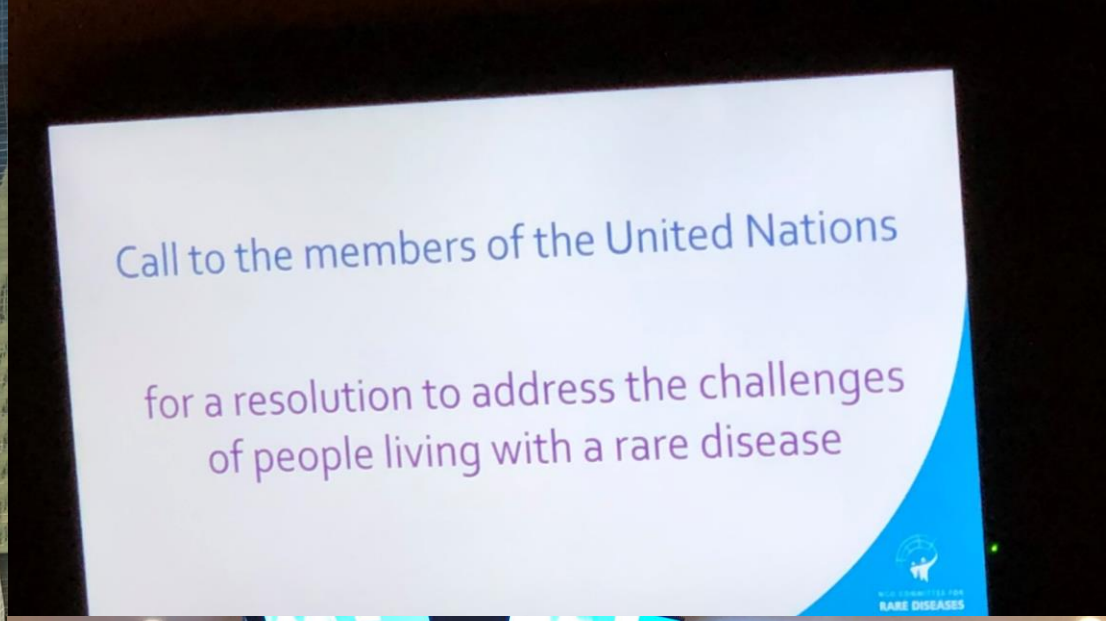


OUR COMMUNITY

Visit patients area

MORE INFO


UN’s the Sustainable
Development Goals 2030
Leave no one behind


Call to the members of the United Nations
for a resolution to address the challenges
of people living with a rare disease


The undiagnosed patients need all of us!

Thank you for listening!

Helene Cederroth
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helene@wilhelmfoundation.org