EURORDIS
RARE DISEASES EUROPE

“It’s not my fault”

Webinar on the diagnosis Odyssey

Webinar, 21.03.2024

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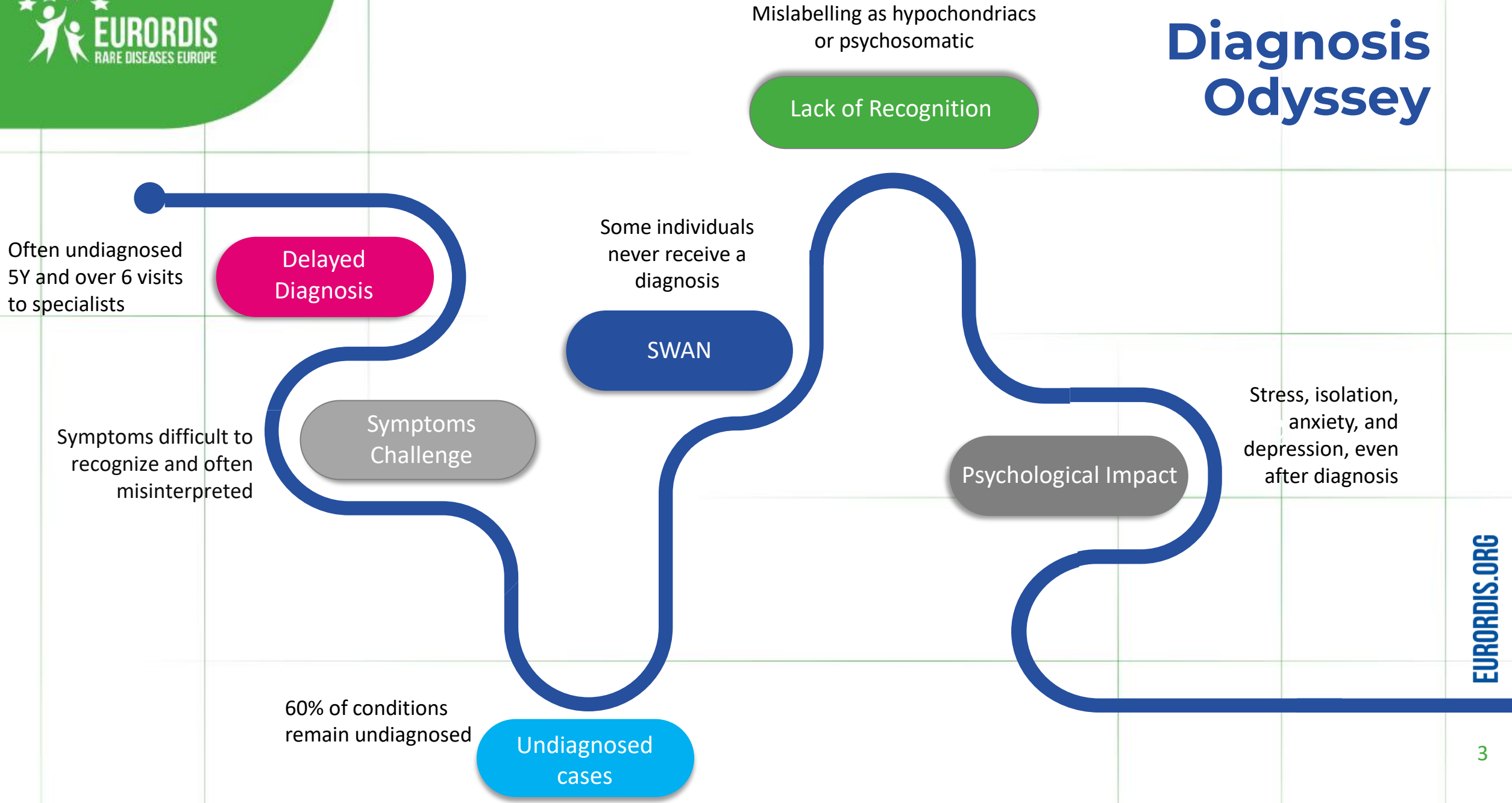
Philipp, osteogenesis imperfecta

Welcome & Opening Remarks

Diagnosis Odyssey



Psychosocial Support

Agenda

Time	Topic	Speaker
11.00 – 11.05	Welcome & Opening Remarks	Concha Mayo, EURORDIS
11.05 – 11.20	Keynote Speech: Impact of the diagnostic odyssey on mental health and wellbeing.	Helene Cederroth, Wilhelm Foundation
11.20 – 11.40	Advocacy in action, securing support for people traveling the diagnosed odyssey.	Gulcin Gumus, EURORDIS
11.40 – 12.25	Panel Q&A	Matt Bolz-Johnson, EURORDIS
12.25 – 12.30	Closing Remarks & Next Steps	Gulcin Gumus, EURORDIS

Misha, Duchenne muscular dystrophy

Impact on Mental Wellbeing of Rare Diseases

Helene Cederroth, Wilhelm Foundation

It is not my fault! – Webinar on the impact of the Diagnostic Odyssey on Mental Health.

Helene Cederroth
Founder President
Wilhelm Foundation

I have no conflicts of interest.

The photos are from Wilhelm Foundation’s activities and the Photo project together with photographer Rick Guidotti, Positive Exposure.
We have consent for all pictures.

Undiagnosed Diseases are a global health problem

Behind every undiagnosed disease
stands a family



Undiagnosed families ***are*** left behind.

Undiagnosed diseases are divided into 3 groups:

1. Not yet diagnosed
2. Undiagnosed, since the disease is not discovered yet
3. Misdiagnosed

Without a diagnosis

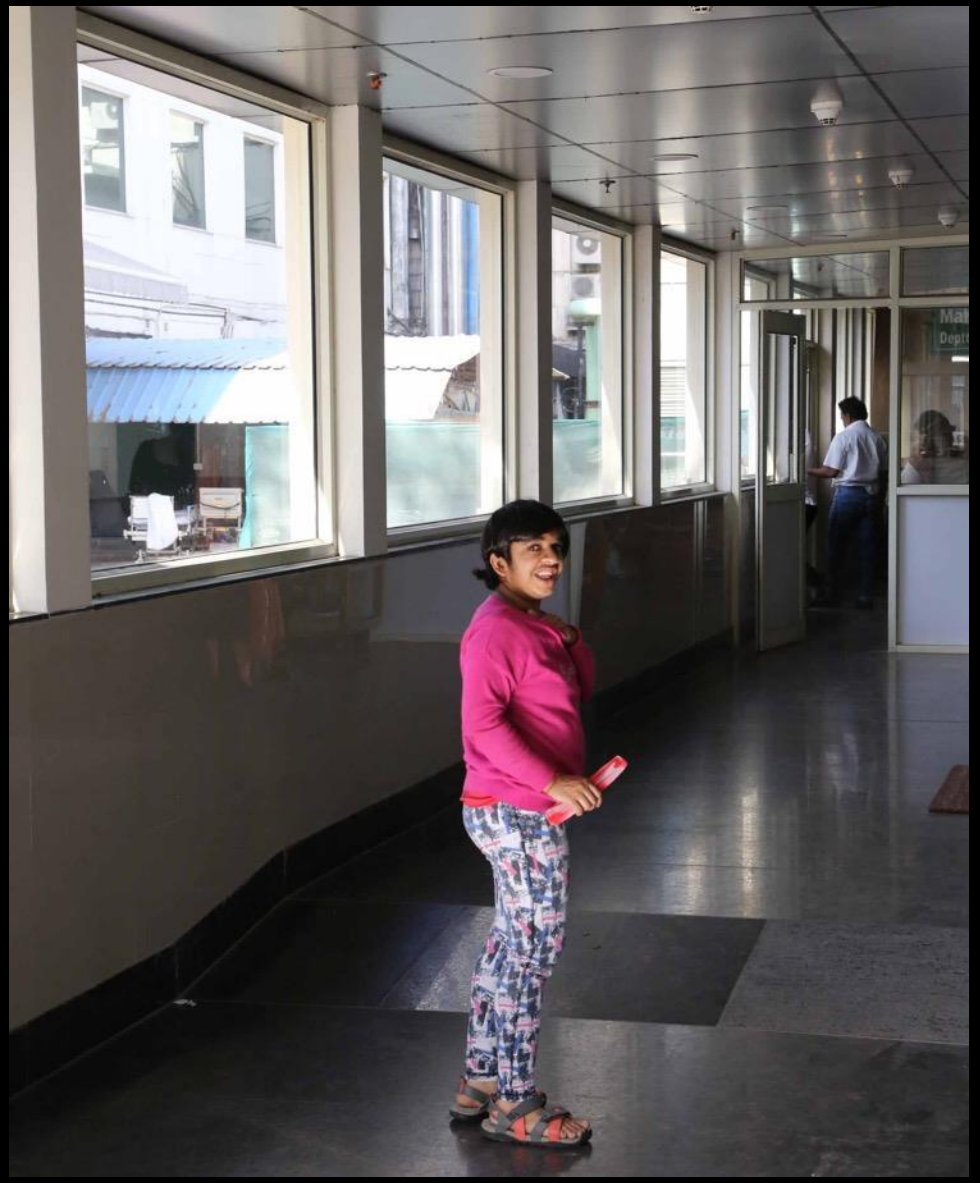
- no one know the cause
- no one understands the disease
- no treatment
- no prognosis

The undiagnosed diseases affect the whole family

Without a diagnosis – no one knows if it's a hereditary disease or not

Without a diagnosis – no one knows if it's a fatal disease or not

The odyssey to reach an accurate diagnosis can take years



- Epilepsy
- Intellectual disability
- Cerebral palsy
- Rare cancer
- Autoimmune diseases
- Overgrowth diseases
- Degenerative diseases
- Metabolic diseases

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*

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

> *"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*

Information and training for patient organizations

Helplines

Information on secondary findings

Next Steps Toolkit

EUROGENTEST Guidelines

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RareConnect .org

ENSERIO study. Time to Diagnosis

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Protocol to support ultra-rare diagnosis

Training for professionals

EURORDIS

SolveRD

CHEO

European Patient Advocacy Group

EuroGentest

GLOBAL COMMISSION

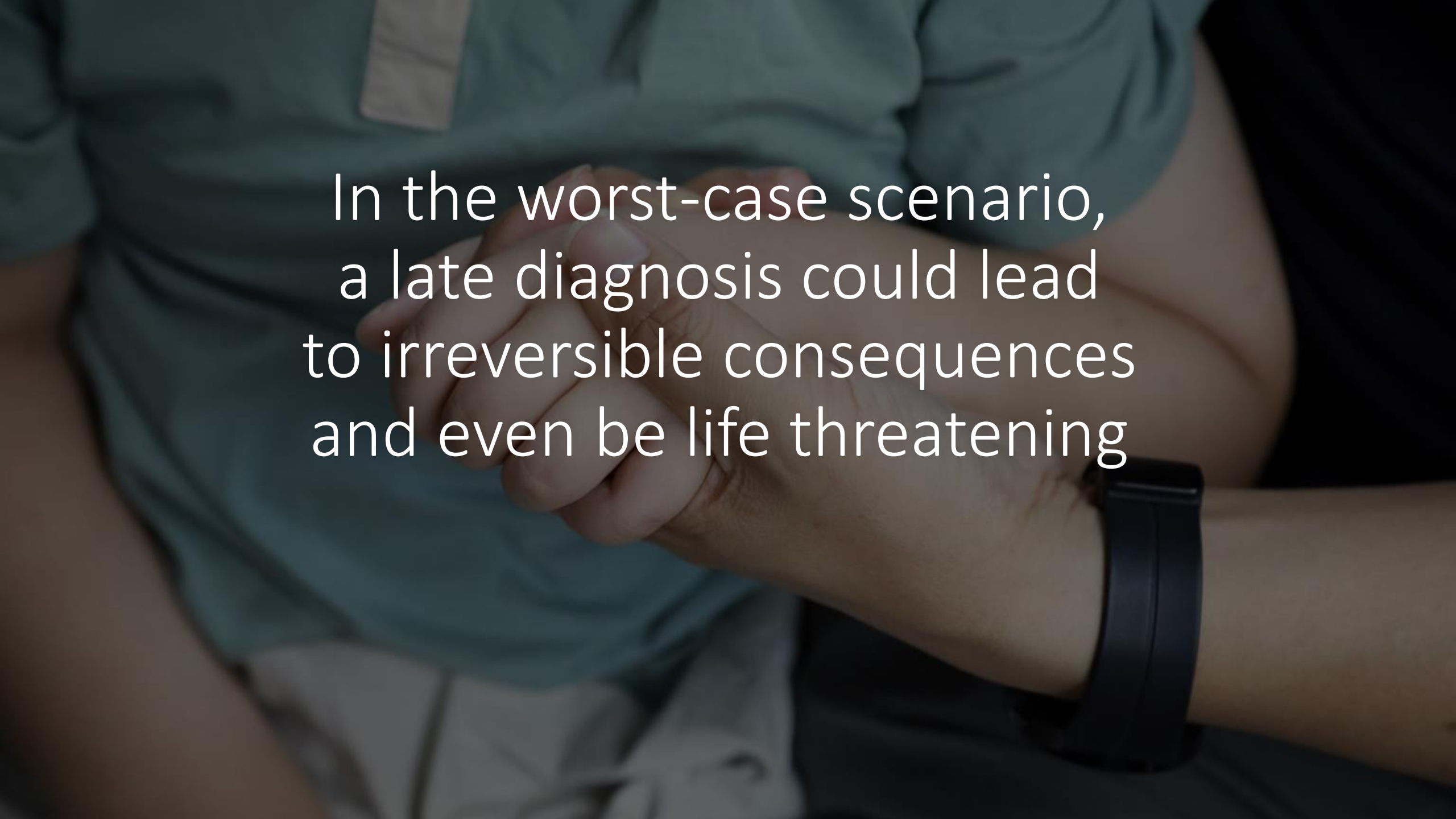
UNIAMO

WILHELM FOUNDATION

feder

SWAN

UDNI


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

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

Children with undiagnosed diseases and their sibling

“Your child is a mystery”

“We don’t know what it is, but it’s nothing dangerous ”

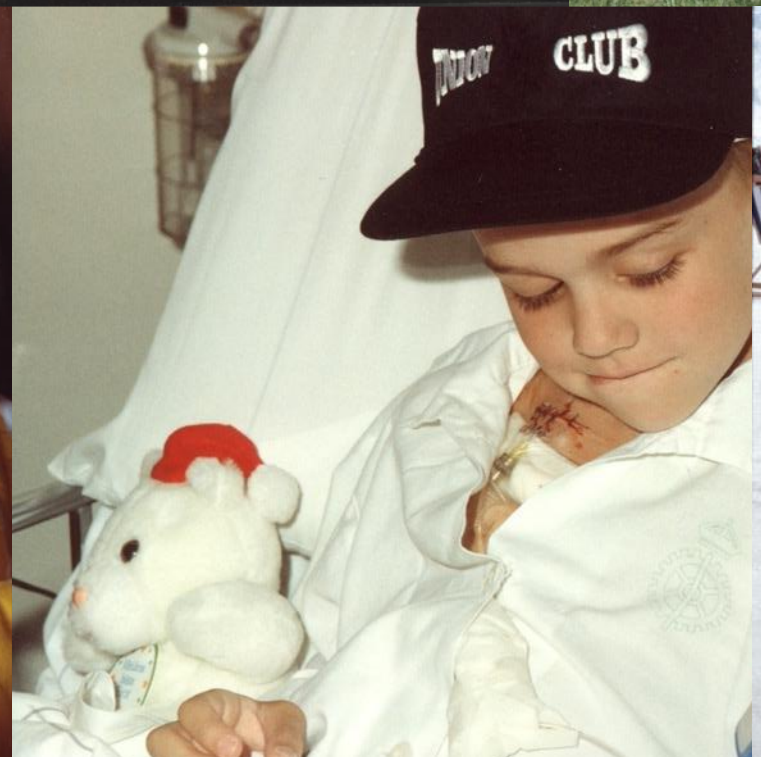
“It’s in your head”

You have to accept that you never will get a diagnosis. Learn to live with it”

“You are doing harm to your child”

“You harming your child since you take him/her to new specialists all the time”

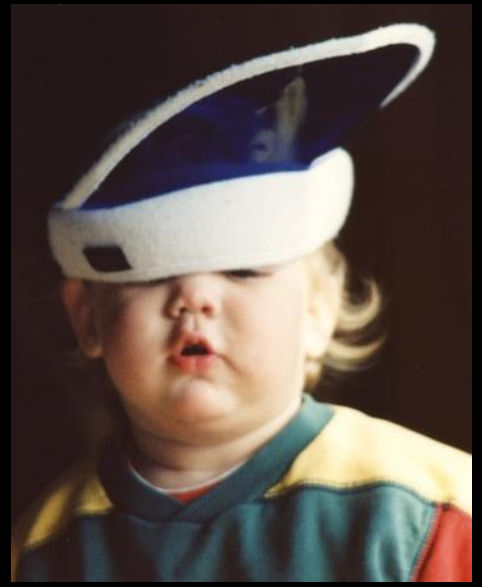
Did I do anything wrong during the pregnancy?


CLUB









Wille
1983 - 1999

Hugo
1991 - 2002

Lilla Emma
1994 - 2000

Wilhelm Foundation

350 million people worldwide living with an Undiagnosed Disease

Advancing Genetic Disease Testing

Next-generation sequencing technology is helping to drive breakthroughs in genetic disease testing by facilitating identification of disease-causing genetic variants. We recognize the significant impact of genetic and rare diseases on families worldwide, and we're developing solutions to facilitate early detection and intervention. A genetic diagnosis can help improve outcomes, promote enduring good health, and raise awareness about the importance of genetics in health care.

Genetic disorders and congenital anomalies are primary contributors of hospitalization and mortality in infants.[1] At least 39% of rare diseases have an identifiable genetic etiology.[2] For adults, 25% of sudden cardiac arrest is due to an inherited genetic condition.[3,4]

> "350 million people worldwide have an undiagnosed disease. I want each and every one of them to find an answer."
>
> — Ryan Taft PhD, Scientist, Medical Genomics Research at Illumina

Genetic Testing for Rare Diseases

2–6% of the population worldwide is affected by a rare disease.[5,6] 80% of these rare diseases have a genetic component,[7] but many patients struggle for years to receive a diagnosis. We are committed to ending these

Cardiovascular Genomics

Sudden cardiac arrest is one of the leading causes of nontraumatic mortality in the US. Cardiovascular genomics research has identified many genetic variants associated with cardiac conditions.

Take home messages

To the family:

- It’s hard but there is hope!
- New diseases are discovered all the time
- We are working to help you
- You are not alone
- You know your child best

To care givers:

- Don't try to normalize
- The undiagnosed are the Zebras – listen for them
- Don't make it more difficult for to search for a diagnosis
- Refer the patients

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Daniel, Williams syndrome

Advocacy in action for individuals travelling the diagnosis Odyssey

Gulcin Gumus, EURORDIS

It’s not my fault

The role of **p**atient **o**rganizations in supporting the undiagnosed community

Gulcin Gumus, PhD

Research and Policy Senior Manager

EURORDIS

21 March 2024

Our Mission

EURORDIS works across borders and diseases to improve the lives of people living with a rare disease

1009
Member patient organisations

74 countries (28 EU countries)

44 National Alliances of rare disease patient organisations

Founded in
1997

Outreach to over
2,500
patient groups

72 European Federations for specific rare diseases

40+
Staff members with offices in Paris, Brussels and Barcelona

Over
440
volunteers

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EURORDIS work to reduce the psychosocial impact of living with a rare disease

Contributing to make change happen:

- **Engaging and partnering** with policy makers, experts and organisations, to provide input to the **design and implementation of policies, initiatives, good practices and funding instruments**.
- Developing or taking part in relevant **European projects**.
- **Empowering its members** and the rare disease community on social and other human rights topics.
- **Contributing to or leading publications on** relevant topics.

International Joint Recommendations to Address Specific Needs of Undiagnosed Rare Disease Patients

1. Undiagnosed rare disease patients should be recognised by national authorities as **a distinct population with specific unmet needs** to enable development of personalised health and social care
2. National sustainable programmes dedicated specifically for undiagnosed diseases should be developed and supported by appropriate authorities in each country to enable rapid and equitable **access to diagnosis and social support**
3. Knowledge and information sharing should be structured and coordinated at national and international levels **to facilitate access to relevant resources for all undiagnosed patients**
4. Patients should be equally involved with other stakeholders in the governance of undiagnosed diseases programmes and international networks to adequately address the priorities of undiagnosed rare disease patients and contribute to **improved healthcare**
5. Ethical and responsible international data sharing should be promoted through existing initiatives to increase collaboration, improve diagnosis, facilitate research and **accelerate treatment of undiagnosed and rare conditions**

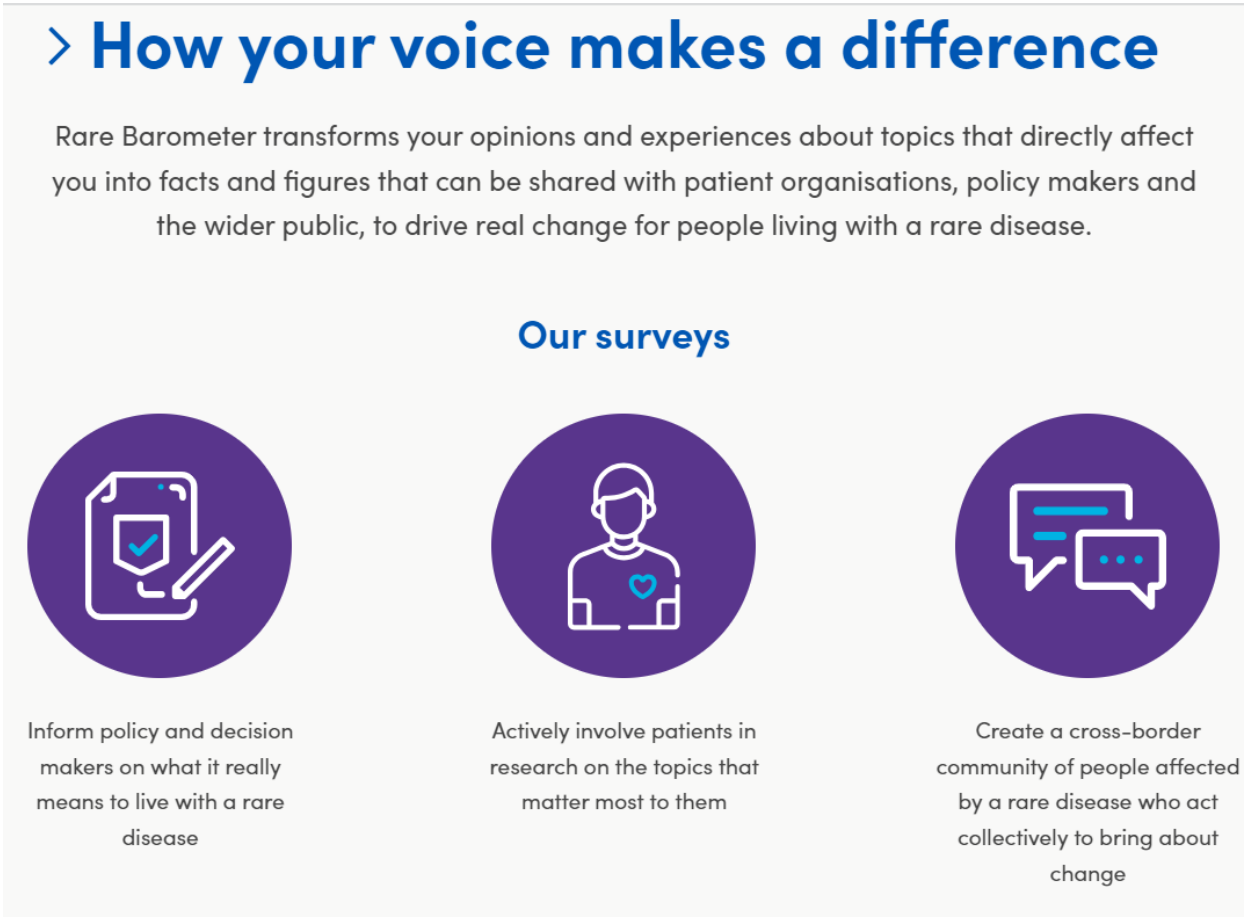
Tools: Rare Barometer Surveys

Rare Barometer

A EURORDIS INITIATIVE

Tools: Rare Barometer Surveys

A EURORDIS INITIATIVE



- 24+ languages
- PLWRD & carers
- Participate from any country in the world
- Country based analysis
- Fact sheet
- Main findings discussed in webinars

Peer reviewed articles

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Rare Barometer

A EURORDIS INITIATIVE

Rare disease patients' opinion on the future of rare diseases
June 2021
Read More →

The future of rare diseases: Leaving no one behind!
May 2021
Read More →

How has COVID-19 impacted people with rare diseases?
October 2020
Read More →

Results of the H-Care survey pilot
February 2020
Read More →

Article: Share and protect our health data
July 2019
Read More →

Share and protect our health data!
May 2019
Read More →

An insight into the participation of rare disease patients in research
November 2018
Read More →

Access to treatment: unequal care for European rare disease patients
August 2018
Read More →

Rare disease patients' participation in research
February 2018

Juggling care and daily life: The balancing act of the rare disease community

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H-Care: The Healthcare Experience of People living with a Rare Disease

Rare disease patients on average rate their healthcare experience...

on a scale from 1 to 5

> *"Often in the context of rare and complex diseases, professionals are powerless, for lack of information or lack of knowledge about an unusual disease. Despite their good will, they discover the disease at the same time as the patient discovers or develops its symptoms."*
> **Rare disease patient**

> *"My experience in different hospitals throughout my life is that only medical aspects are attended to. The psychological and emotional part is not taken into account nor is there a joint multidisciplinary treatment."*
> **Rare disease patient**

Rare disease patients seem to have a worse experience of health care than patients with chronic diseases: 70% of studies that used the same questionnaire reported a higher average score for chronic disease patients' experience of health care[1].

To ensure a better healthcare experience the top 3 areas that need to be improved are:

1

Contacting patients or carers after a visit to see how things are going

> *"No multidisciplinary follow-up is proposed as in more well-known or classic serious diseases."*
> **Rare disease patient**

> *"Since the diagnosis, about 4 years ago, the facility has not contacted me for any follow-up."*
> **Rare disease patient**

2

Encouraging patients or carers to go to a specific group or class to help them cope with the rare disease

> *"I have no psychological or social help; I do all my steps alone thanks to the support of an association. It is a shame that we are not advised to approach certain associations."*
> **Rare disease patient**

3

Helping patients and carers deal with emotions related to the patient's health status

> *"Despite the good technical competence of the facility, I feel very alone and not well supported psychologically."*
> **Rare disease patient**

> *"I lack support regarding the psychological or emotional level of the disease, the side effects of medications and how this affects my private life."*
> **Rare disease patient**

Q *Over the past 6 months, when I/the person I care for received medical care for my/his/her rare disease, I was:*

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Rare2030
Foresight in Rare Disease Policy

Rare Barometer
A EURORDIS INITIATIVE

THE FUTURE OF RARE DISEASES:
LEAVING NO ONE BEHIND!

Key findings from a survey on the opinion of people living with rare diseases on policies that may impact their lives

79% **of people living with a rare disease do not expect to be cured from their rare disease within the next 10 years, but they hope to:**

- **58%** be supported to manage the psychological or emotional aspects of the rare disease
- **53%** have their rare disease stabilised
- **49%** manage the symptoms of the rare disease even if they are still progressing
- **44%** access adapted and accessible employment as well as flexible work arrangements
- **39%** not be discriminated against due to their rare disease or due to their disabilities, in the various aspects of their daily life

ON PERSON-CENTRED CARE...

Diagnosis is a challenging step for people living with a rare disease, which often lack psychological support and accompaniment. Young Citizens think that current efforts do not sufficiently take account of aspects that go beyond the physiological side of diagnosis and therefore recommend:

+ **Bringing support to people living with a rare disease before and at the time of diagnosis**, in the form of psychological support and other initiatives aimed at accompanying patients from symptoms onset to diagnosis and beyond.

New Global Rare barometer survey

On the journey to diagnosis for people living with a rare disease

*Identifying personal and external factors influencing the process of obtaining timely and accurate diagnosis from a **patient perspective***

TARGET POPULATION

All patients living with a rare disease and their family members, including:

- Former or recovering patients (e.g. cancer survivors)
- **Undiagnosed**
- Any experience of diagnosis: difficult or easy, long or short.

WORLDWIDE

The survey is open to people living with a rare disease and their family members from **any country in the world**, and is **translated in 26 languages.**

TIMELINE

The survey run from **17 March to 15 June 2022.**

SHAPING THE ONLINE QUESTIONNAIRE

1

Literature review

Identify **indicators** already existing or still missing

2

Online panel

Refine the **diagnosis concept** and identify what is **new in the field**

61 participants + 8 individual interviews

"opinionway healthcare

3

Topic Expert Committee

Contribute to identifying **issues and indicators** to include in the questionnaire

4

Council of National Alliances

Input on **topics and indicators** to be included

Feedback on the questionnaire

5

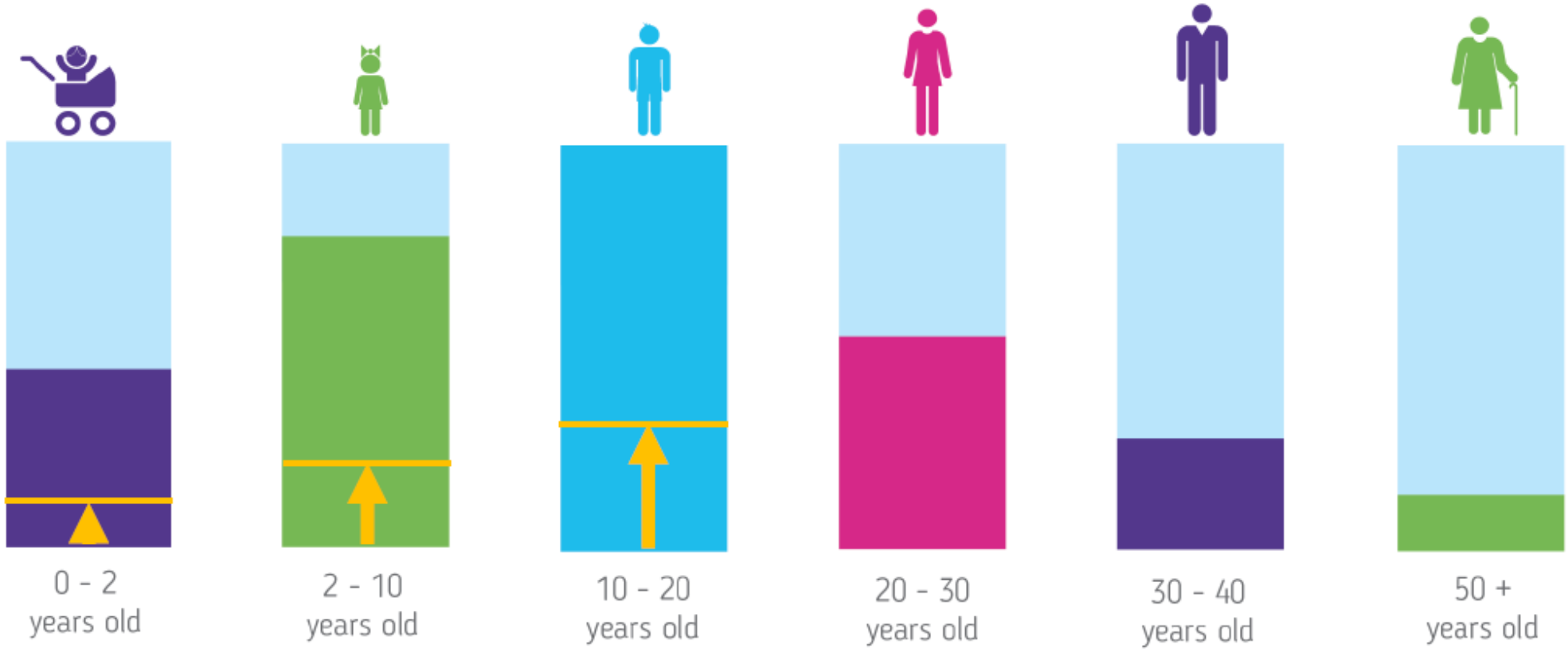
Pilot test with patients

9 participants

Translations checked **in 15 languages** by native speakers

eurordis.org/rare-on-air

THE JOURNEY IS LONGER FOR CHILDREN AND ADOLESCENTS



Average number of years between the first symptoms and the confirmed diagnosis depending on the age of the patient **when first symptoms were noticed**

- Listen to the podcast!

eurordis.org/rare-on-air

- Article submitted to the European Jo
of Human Genetics (published soon)

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Solve-RD: Solving the unso

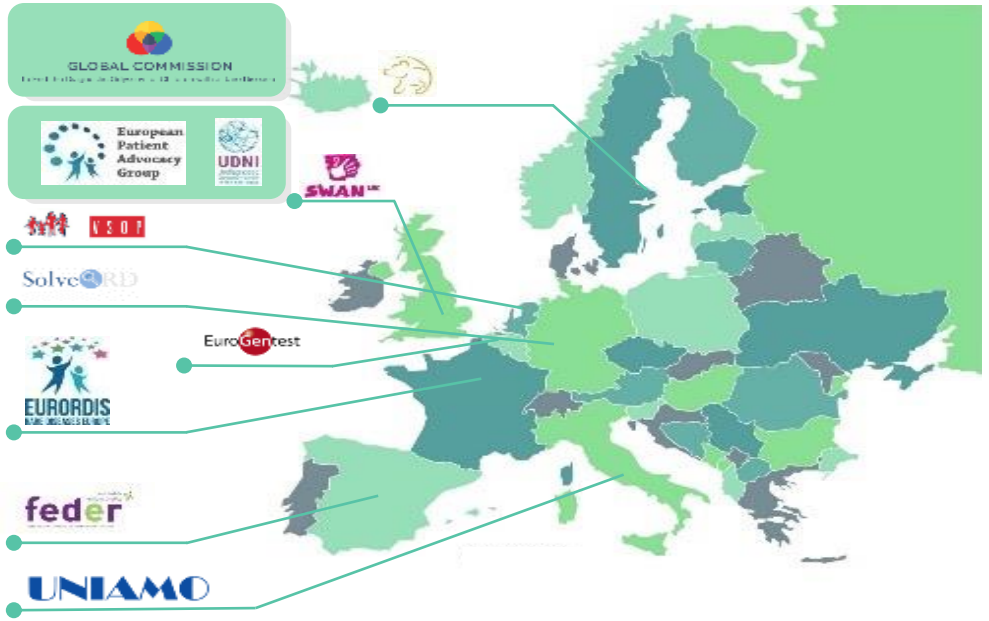
- 2018-2024
- EU funded project
- To solve large numbers of rare disease, for which a molecular cause is not known yet by sophisticated combined omics approaches

- 23 Institutions (including ERNs and UDPs)

  - Community Engagement Task Force
  - Experience Based Co-Design

Community Engagement Task Force

25 members from 19 organizations within 4 networks (UDNI, SWAN EUROPE, ePAG rep, Global Commission) and Solve-RD



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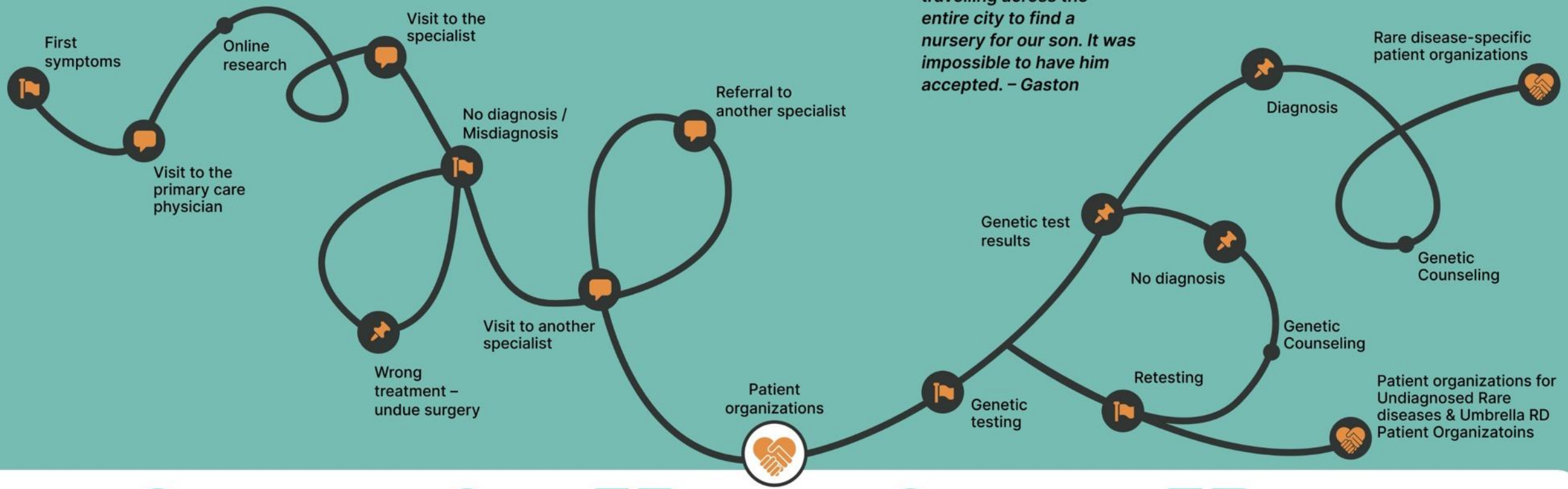

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EURORDIS

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

UNIAMO

feder

SWAN UK

UDNI

Institut Imagine @InstitutImagine · 29 Tem 2020

"Today, still more than 1/2 children come out of a genetic consultation without a diagnosis. Diagnosis is the sine qua non condition to go further in research and therefore care. There is an urgent need for new means to speed up the diagnosis." S. Lyonnet, Imagine Director

Elina Miaouli @ElinaMiaouli · 28 Tem 2020

Η "Διαγνωστική Οδύσσεια" των ασθενών με #ΣπάνιεςΠαθήσεις

Ίσως μία από τις μεγαλύτερες προκλήσεις που αντιμετωπίζουν οι σπάνιοι ασθενείς είναι η αναμονή από 5-30 χρόνια μέχρι να να λάβουν μία σωστή #Διάγνωση, με ό,τι συνεπάγεται αυτό!

#Υγεία #ΣπάνιαΣύνδρομα #SpaniosGr #Rare95

FuSCA @FuSCArgentina · 19 Tem 2020

El viaje de las familias con una #EPOF ilustrado aquí.

Desgasta? Angustia? Frustra? Claro que sí, por eso se necesita de los pacientes para que con nuestra experiencia ayudemos a que esta ODISEA no sea tal. Llevará tiempo, pero podremos, PODEMOS.

MaRIH @Filiere_MaRIH · 30 Tem 2020

L'infographie du #parcours du #patient disponible en fra
édité par @eurordis :

AchacunsonkaraT @Achacunsonkara

Tellement vrai 😢😢😢

Me gustó este errático viaje hacia el #diagnóstico aunque para el #sjögren las curvas se acentúan y se complican por el conjunto de síntomas tan diversos inesperados, y poco conocidos por muchos médicos 😜 Gracias por la visibilidad @eurordis #sjogrens #autoimmune #pacientes

neilsmith38 Similar to my journey with Kallmann syndrome. The genetics part is a dead end for my condition and most of us get dismissed as late bloomers for a long time at the start of the journey.

1d Reply

neilsmith38 Patient groups and patient contact makes all the difference

INPDR @inpdr_tweets · 16s

Rare disease patients shouldn't have to go through a diagnostic odyssey like this😔

Bringing patient data together, like we are, will improve knowledge and understanding so we can help patients get answers quicker.

SCN2A Europe @scn2aeurope · 8 Haz

Thank you for making it so graphic! Also the #SCN2A community, especially the Loss-of-Function cases, have to wait far too long!

Matt B @MattB49301653 · 23s

@LinerJoyce ve @eurordis adlı kullanıcılara yanıt olarak

slightly different in the UK though; forget genetic testing in many cases, add accusations of hypochondria from friends and family, Trusts refusing to treat patients and in many cases "what patient organisation?". Too often its a very solitary and soul destroying journey.

LIFE Worldwide @LIFEworld... · 8 Haz

Very relevant to conditions like chronic pulmonary #aspergillosis

#breathe #chronicillness #invisibleillness #spoonie

Patient Safety / Patients Rights 🇨🇦 @pttopt · 28 Tem 2020

Can relate to this journey , it takes a toll (emotionally , physically , financially) on the Patient as well as their family .

#PatentExperience #RareDisease #rare #onpoli

@gulcingumus1 @ACURARE1 ve diğer 5 kişiye yanıt olarak

O kadar gerçekçi bir sıralama ki👏👏👏 her #nadirhasta başına gelenlerin sebebini öğrenmeyi hakediyor, geçimini sağlıkla ilgili kazanan herkes de bu konuda katkı sunmaya vebaldeler, literatürleri güncelleyecek / değiştirecek çalışmalara ihtiyaç var, emek verenlere Teşekkürler🙏

Integration of ERNs into Healthcare Systems

3 years €18 million funding to improve the diagnosis, treatment a
patients with rare diseases

Develop recommendations for national patient organizations for
patients

A task force with key stakeholders

- Patient experts
- Clinicians
- Psychologists
- Policymakers
- ERN network

Take part in our task force!

Improving the pathways /Best practices

What type of support is needed? (Psychological) Where can patients find that support?

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UDNI PATIENT ENGAGEMENT GROUP

UDNI PATIENT ENGAGEMENT GROUP

Objectives:

- A platform to bring together the organizations tha

on undiagnosed

- To guarantee increased involvement and active contribution in the UDNI

22 Member organisations from all continents (Countries including US,Uruguay,Argentina,India, Ghana, China, Australia)

UDNI PATIENT ENGAGEMENT GROUP

Activities

Patient session at the 12th International UDNI Conference

Pre-conference session: “Towards International Integration”

- 50+ participants
- Undiagnosed families and patients from 23 patient organizations
- Presentations from Wilhelm Foundation, EURORDIS, NORD and SCN2A Georgia on:
  - Diagnostic Odyssey
  - Newborn Screening
  - Centers of Excellence
  - Mental health

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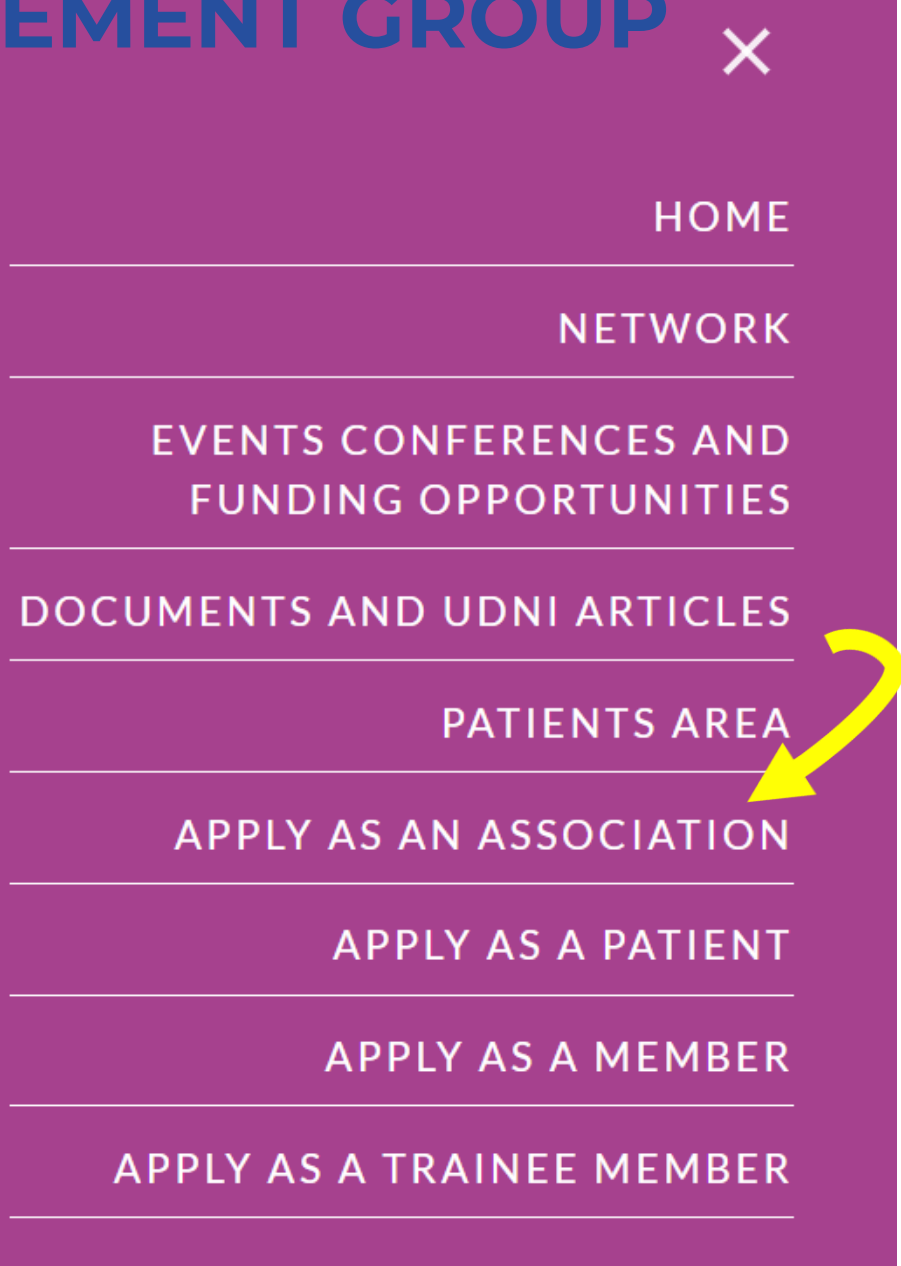
UDNI PATIENT ENGAGEMENT GROUP

- The organization is listed on the UDNI website
- Invited to join our PEG meetings
- View slides from presentations of our membership
- Register to the UDNI Conferences & information on UDNI meetings
- Email updates from the PEG on meetings, networking opportunities and other activities

https://www.udninternational.org/

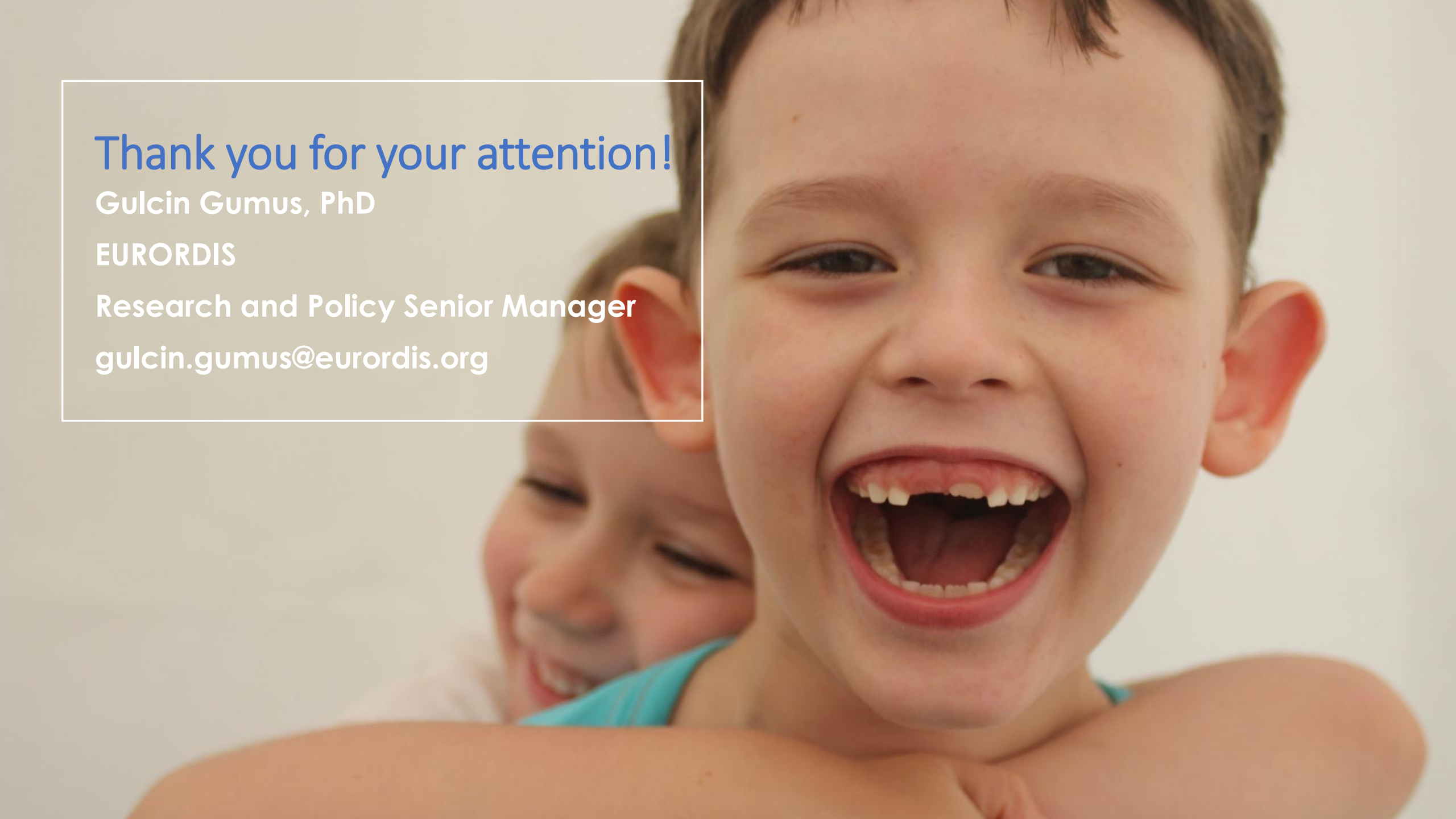


Take home messages

- Undiagnosed rare disease patients should be recognised by national authorities as a **distinct population with specific unmet needs**

- **Patient empowerment is** needed for adequate and **full involvement of patient representatives in research for undiagnosed and to integrate psychosocial support in policy topics concerning undiagnosed diseases.**

- Establishing **an active undiagnosed community** is key!



Thank you for your attention!

Gulcin Gumus, PhD

EURORDIS

Research and Policy Senior Manager

gulcin.gumus@eurordis.org

Kasper from Poland is living with Kawasaki disease

Panel Discusion

Matt Bolz-Johnson, EURORDIS

Panellists

Matt Bolz-Johnson

EURORDIS

Moderator

Helene Cederroth

WILHELM FOUNDATION

Panellist

Gulcin Gumus

EURORDIS

Panellist

Gareth Baynam

WESTERN AUSTRALIA UNIVERSITY

Panellist

Charlotte Gaasterland

FEDERATIE MEDISCH SPECIALISTEN

Panellist

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Misha, Duchenne muscular dystrophy

Q&A

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Closing Remarks

3rd Undiagnosed Day
29 April 2024
in person at Harvard.

And a streamed Undiagnosed Day update from around the globe.

A celebration to the beauty and diversity in the Undiagnosed Community

For more information visit

www.undiagnosed-day.org

Undiagnosed Day 2024

Awareness webinar

A celebration the beauty and diversity in the Undiagnosed Community

Undiagnosed Day 2024

by Wilhelm Foundation in collaboration with UDNF and UDNI

29 April

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Eddison, xeroderma pigmentosum

Thank you!

Please complete the webinar survey