“It’s not my fault”

Webinar on the diagnosis Odyssey

Webinar, 21.03.2024
Welcome & Opening Remarks

Philipp, osteogenesis imperfecta
Diagnosis Odyssey

Delayed Diagnosis
- Often undiagnosed 5Y and over 6 visits to specialists
- Symptoms difficult to recognize and often misinterpreted

Lack of Recognition
- Mislabelling as hypochondriacs or psychosomatic
- 60% of conditions remain undiagnosed

Symptoms Challenge

SWAN
- Some individuals never receive a diagnosis

Psychological Impact
- Stress, isolation, anxiety, and depression, even after diagnosis

Undiagnosed cases
- Often undiagnosed
Psychosocial Support
## Agenda

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<td>Concha Mayo, EURORDIS</td>
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<td>Keynote Speech: Impact of the diagnostic odyssey on mental health and well-being.</td>
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<td>Gulcin Gumus, EURORDIS</td>
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Impact on Mental Wellbeing of Rare Diseases

Helene Cederroth, Wilhelm Foundation

Misha, Duchenne muscular dystrophy
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
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?
Hugo passed away 10 years old
Wille
1983 - 1999

Hugo
1991 - 2002

Lilla Emma
1994 - 2000
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 healthcare.

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

Geneve Testing for Rare Diseases

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

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.

"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
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
Advocacy in action for individuals travelling the diagnosis Odyssey

Gulcin Gumus, EURORDIS
It’s not my fault

The role of patient organizations 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

72 European Federations for specific rare diseases

1997
Founded in

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

440 volunteers

Outreach to over 2,500 patient groups
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

“Make your Voice heard!”
**Tools: Rare Barometer Surveys**

> How your voice makes a difference

Rare Barometer transforms your opinions and experiences about topics that directly affect you into facts and figures that can be shared with patient organisations, policymakers and the wider public, to drive real change for people living with a rare disease.

**Our surveys**

- Inform policy and decision makers on what it really means to live with a rare disease.
- Actively involve patients in research on the topics that matter most to them.
- Create a cross-border community of people affected by a rare disease who act collectively to bring about change.

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

**Peer reviewed articles**
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

2.5

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.

2.0

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. Not is there a multidisciplinary treatment.

2.1

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.

2.3

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

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

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

"opinionway healthcare"
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

- 0 - 2 years old
- 2 - 10 years old
- 10 - 20 years old
- 20 - 30 years old
- 30 - 40 years old
- 50 + years old
• Listen to the podcast!

eurordis.org/rare-on-air

• Article submitted to the European Journal of Human Genetics (published soon)
Solve-RD: Solving the unsolved

• 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
Patient Journey through diagnosis

“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

First symptoms
Online research
Visit to the primary care physician
Visit to the specialist
No diagnosis / Misdiagnosis
Wrong treatment – undue surgery
Referral to another specialist
Patient organizations
Genetic testing
Genetic test results
Retesting
No diagnosis
Genetic Counseling
Diagnosis
Rare disease-specific patient organizations
Patient organizations for Undiagnosed Rare diseases & Umbrella RD Patient Organizations

Information and training for patient organizations
Helplines
Information on secondary findings
Next Steps Toolkit
EUROGENTEST Guidelines
Experience Based Co-Design
RareConnect.org
ENSEMO study: Time to Diagnosis
Undiagnosed Photo Project
Protocol to support ultra-rare diagnosis
Training for professionals
SolveRD
CHEO
European Patient Advocacy Group
Eurogentest
GLOBAL COMMISSION to End the Diagnostic Delay for Children with a Rare Disease
UNIAMO
GLOBAL RARE
UNICEF
IPFD
feder
SWAN
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 Miaoulis @ElinaMiaoulis - 28 Tem 2020

Η "Διαγνωστική Οδύσσεια" των ασθενών με #ΣπάνιεςΠαθήσεις

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

#Υγεία #ΣπάνιαΣύνδρομα #SpaniosGr #Rare95

FuSCA @FuSCAArgentina - 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 frais 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

Patient groups and patient contact makes all the difference

INPDR @inpr_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.

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.

#PatientExperience #RareDisease #rare #onpoli

@gulcingumus1 @ACURARE1 ve diğer 5 kişiye yanıt olarak

O kadar gerçekçi bir sırılama ki❤️❤️❤️ her #nadirhasta başına gelenlerin sebebin bagi organneyi hakediyor, güçünü sağlakca ilgili kazanan herkes de bu konuda katki sunmaya vebildeler, literatürleri güncelleyeceğiz / değiştireceğiz çalışmalarla ihtiyaç var, emek verenlere Teşekkürler🏆
3 years €18 million funding to improve the diagnosis, treatment and care of 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

Improving the pathways /Best practices

What type of support is needed? (Psychological) Where can patients find that support?
UDNI PATIENT ENGAGEMENT GROUP

Objectives:

- A platform to bring together the organizations that focus 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)
Activities

Patient session at the 12th International UDNI Conference

UDNI PATIENT ENGAGEMENT GROUP

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
• 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
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
Misha, Duchenne muscular dystrophy
Iris, Ehlers-Danlos syndrome, chronic paroxysmal hemicrania, cluster headaches
3rd Undiagnosed Day
29 April 2024
in person at Harvard.

And a streamed
Undiagnosed Day
update from around
the globe.

Home  |  2023  |  2022  |  More

A celebration
to the beauty
and diversity
in the
Undiagnosed
Community

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 UDRN

29 April

For more
information
visit

www.undiagnosed-day.org
Thank you!

Please complete the webinar survey