

Solve-RD Community Engagement Task Force

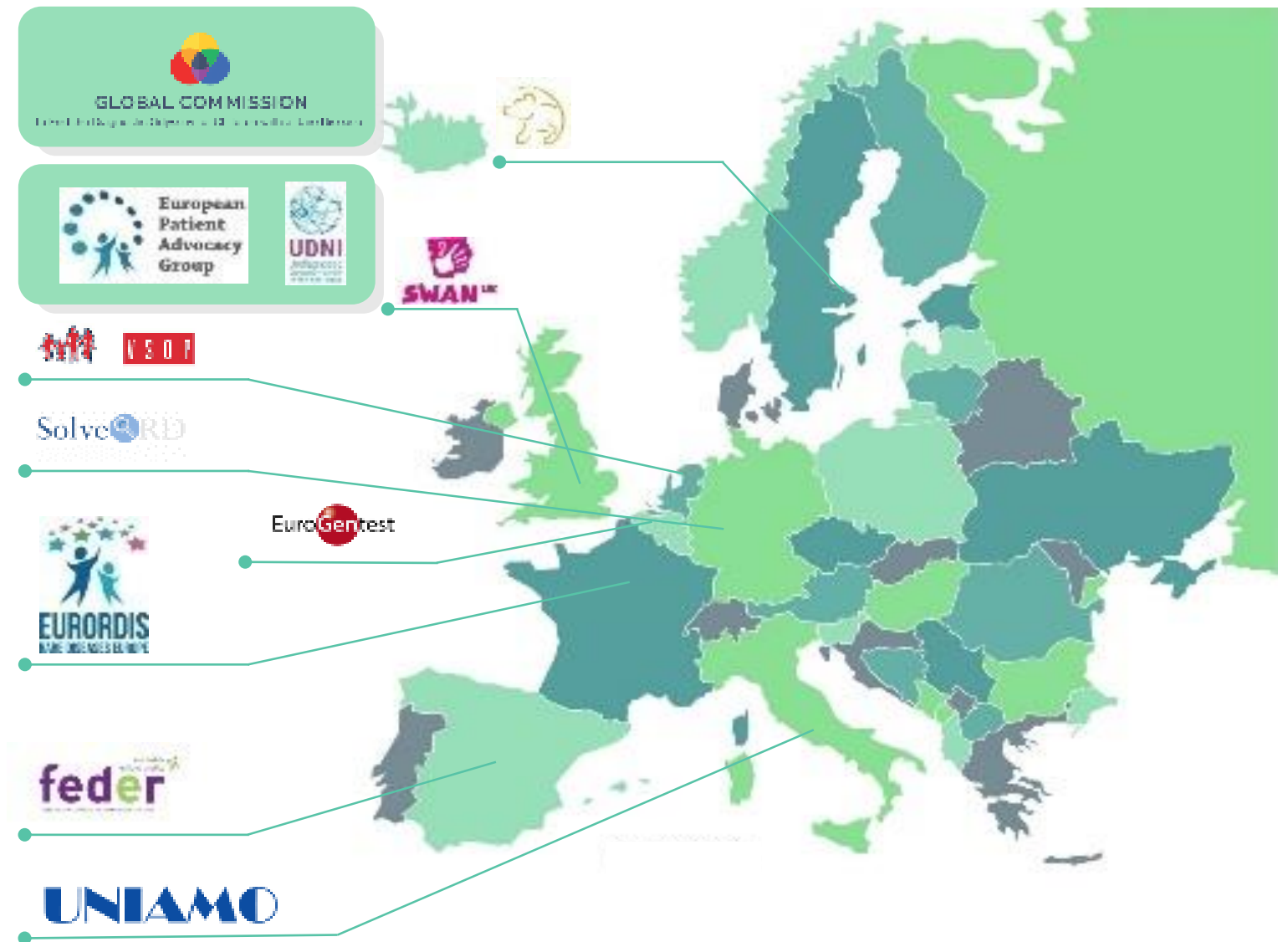
Patient Journey Through Diagnosis

Gulcin Gumus
Research & Policy Project Manager
EURORDIS

Objectives

- To support the **empowerment of undiagnosed patients** in the development and implementation of novel diagnostic and care pathways
- To serve as a **dissemination channel** toward the undiagnosed
- For any diseases still unsolved at the end of the project, to leave a legacy of a **strengthened support community** for the undiagnosed.

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



Challenges/Questions

- What does the patient journey through diagnosis look like?
- How can the results of the research project communicated to the patient?
- Guideline / toolkit following genetic counselling
- Coding of undiagnosed patients (RD-CODE)
- Undiagnosed registries

Action points for 2020

- To produce a map of existing resources for undiagnosed community
- To develop a visual infographic, of the patient journey through diagnosis

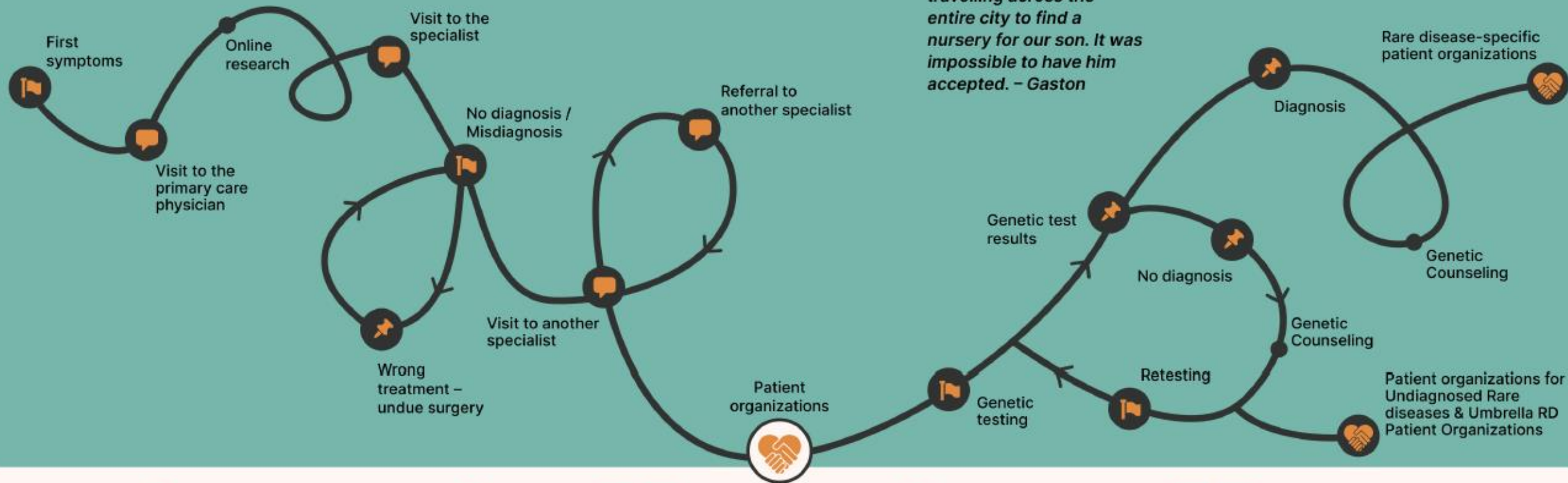
Patient Journey through diagnosis

“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



Map of Resources

Rareconnect.org

Online platform for rare disease patients, families and patient organizations to develop communities and conversations across continents and languages

Rare Diseases Helplines



Undiagnosed Photo Project:

This Wilhelm Foundation initiative aims to reach a diagnosis through sharing and it is created for patients who have been evaluated at the Undiagnosed Disease Program (UDP) and are still undiagnosed after the program.

SWAN-UK

Information to patients and parents
Help create new undiagnosed organizations

MD4RD

Training of professionals

EURORDIS.ORG



[hidden]
30 days ago [Undiagnosed diseases](#)
originally written in English

EN

Do my symptoms sound familiar? Undiagnosed, suspected malabsorption? EDS? Something else?

Hi! You can call me Mae. I'm 22 (F) and have been experiencing a constellation of symptoms that have progressively gotten worse throughout my life. Unfortunately, I had a dismissive family doctor as a child, and now, my doctors honestly have no idea what's going on: they referred me to a geneticist, however, this has been postponed due to



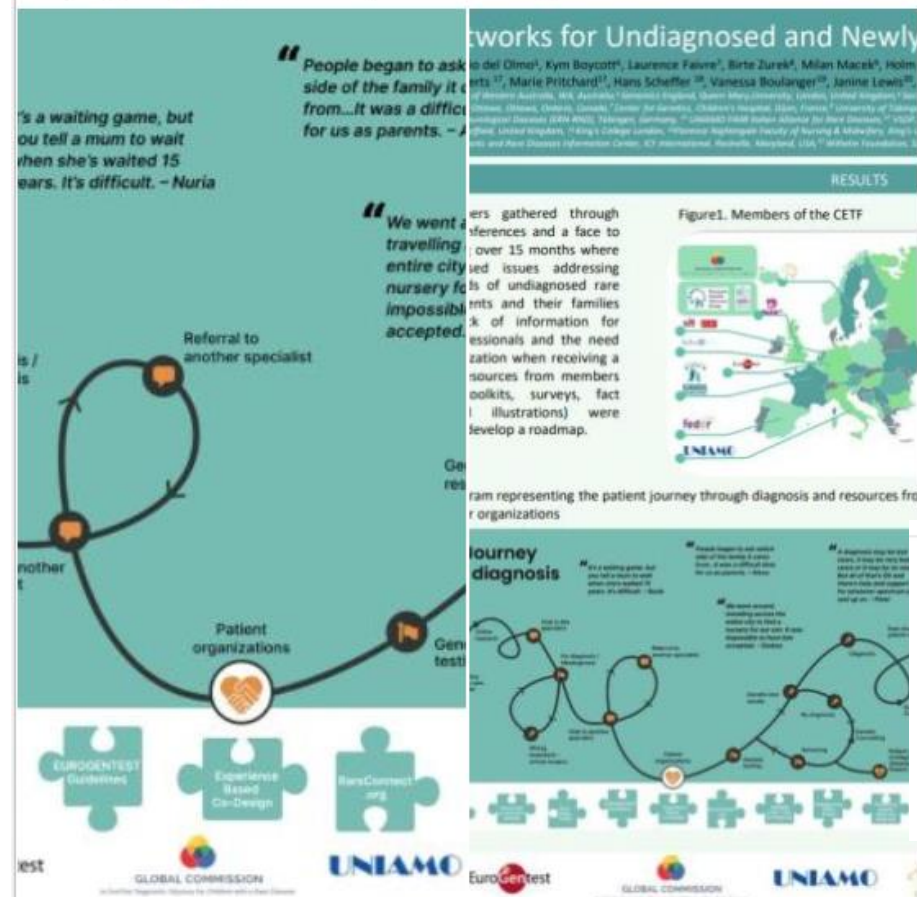


EURODIS

5,065 followers

2d • ④

Take a look at this new poster & infographic published at the European Society of Human Genetics conference this weekend. It illustrates the staggering diagnostic odyssey many people affected by rare diseases experience and was crea ...see more



eurordis • Follow



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 & patient contact makes all the difference



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



@wmpdr_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.

#inpdr #npc #asmd #niemannpick

Next Steps

- Make it interactive
- Translate
- Spread the word

Next Steps for 2021

Guidelines for creating an undiagnosed patient organization

Key messages

- Establishing an active undiagnosed community is extremely important.
- Patient organizations should be involved in earlier steps of the journey through diagnosis.
- Education of professionals on rare & ultra-rare diseases is essential
- Patients as equal partners drive forward active collaborations among the different stakeholders.