THE DIAGNOSIS JOURNEY OF PEOPLE LIVING WITH RARE DISEASES

EURORDIS Membership Meeting
Workshop "Living with a rare disease in childhood"
26th-27th May 2023 – Stockholm

Jessie Dubief, Rare Barometer Senior Manager

GOALS OF THE DIAGNOSIS SURVEY

- Measuring the **time necessary to obtain a diagnosis** as a rare disease patient
- Understanding the **different steps** of the diagnosis journey
- Defining the **obstacles limiting the access** to diagnosis
- Identifying **best practices, tools, support and services** contributing to a faster diagnosis
- Identifying the **role** of new technologies

ONLINE QUESTIONNAIRE

More information: watch our webinar

17 MARCH ⇢ 15 JUNE 2022

13,304 respondents worldwide and **10,453** in Europe

27 languages

104 countries

1900+ diseases represented

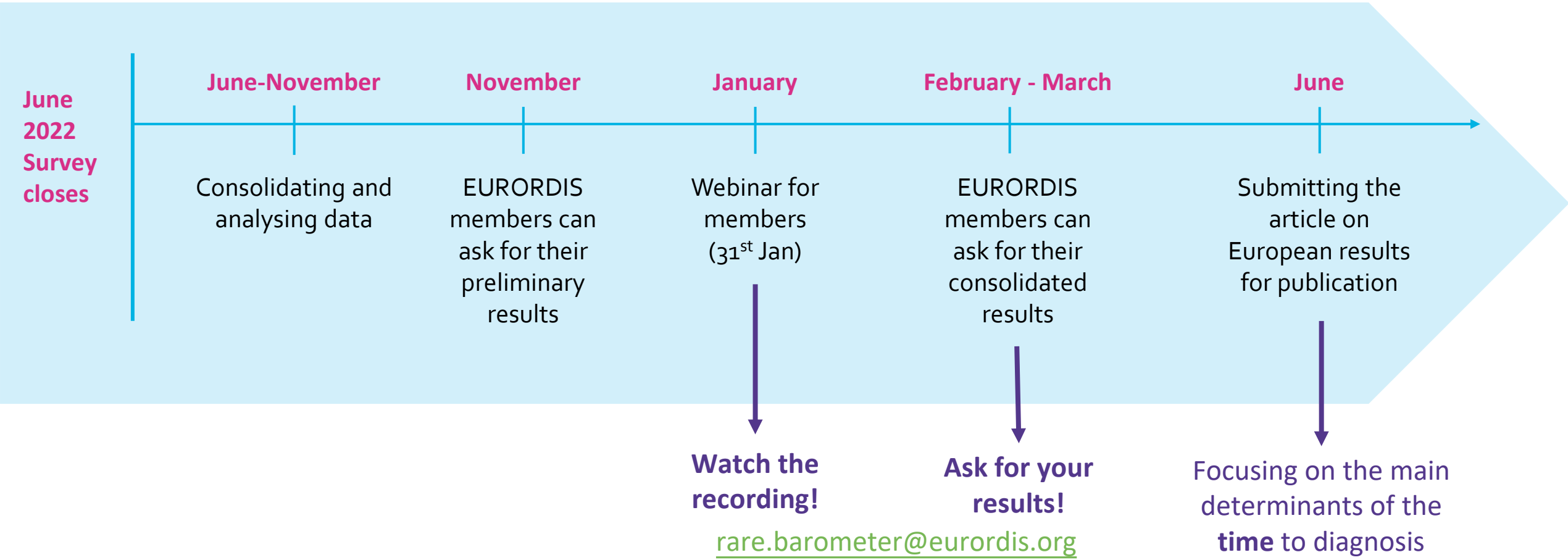
TARGET POPULATION

All patients living with a rare disease and their family members, including unsolved cases (undiagnosed)

RARE DISEASES INTERNATIONAL

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

DIAGNOSIS SURVEY TIMELINE



DIFFICULTY OF THE DIAGNOSIS

“Fortunately, I didn’t face any obstacles. The people at the hospital took care of the child properly when I arrived. In 2 days I had an MRI and in 1-2 days they told us why they suspected the syndrome. Then it took us until the genetic tests were confirmed”

Parent of a child with Leigh syndrome, Romania, diagnosis confirmed 3 months after first symptoms

“I myself went to another country at my expense to ask for help to be able to carry out the genetic test. I am unemployed and not helped by the country where I live!”

Patient living with glycogen storage disease, Luxembourg, diagnosis confirmed 25 years after first symptoms

THE AVERAGE JOURNEY TO DIAGNOSIS FOR PLWRD

1ST SYMPTOMS ARE NOTICED
(beginning of the diagnosis journey)

1ST SYMPTOMATIC TREATMENT

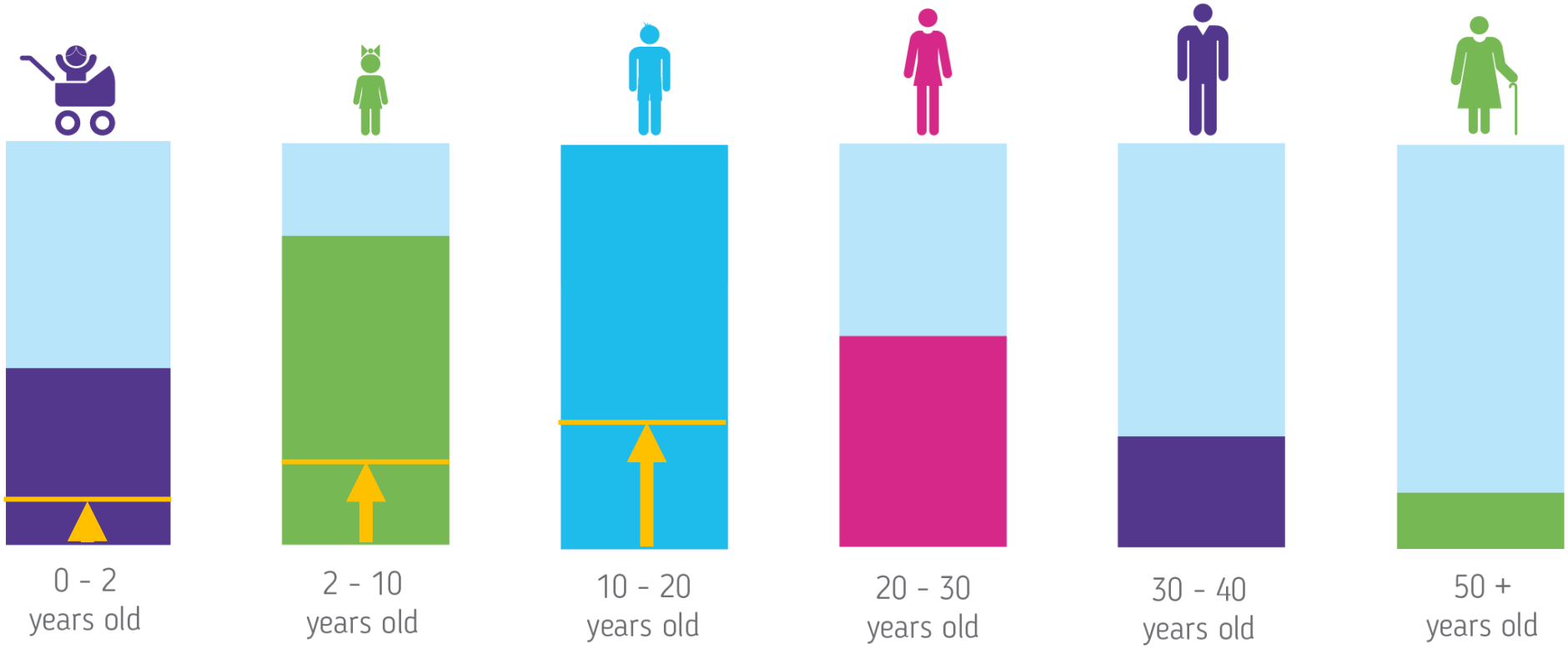
CONFIRMED DIAGNOSIS

1ST MEDICAL CONTACT

INITIAL DIAGNOSIS

REFERRAL TO A CENTRE OF EXPERTISE

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**

NEW RARE BAROMETER SURVEY

On newborn screening

SCREEN 4CARE

31 MAY → 9 JULY 2023

TARGET POPULATION

All patients living with a rare disease and their close family members

24 languages

ALL rare diseases

REGISTER FOR OUR WEBINAR!

31 May 14:00 CET

tiny.cc/webinar_NBS

Ask for your dashboard: rare.barometer@eurordis.org

EURORDIS RARE DISEASES EUROPE

Rare Barometer
A EURORDIS INITIATIVE

Questions?

Thank you

to the Rare Barometer participants, partners and corporate donors in 2022!

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