

WEBINAR – 30 January, 2:30-4:30pm CET

Results of the Rare Barometer Diagnosis Survey: use them in your advocacy work and your actions!

Accurate and timely diagnosis is critical to end the diagnostic odyssey experienced by people living with rare diseases and ensure their access to appropriate health and social care. Early diagnosis allows to provide supportive holistic care as early as possible, can delay or reduce the apparition of symptoms and impairments, and defer the apparition of co-morbidities. Policy goals set for the diagnosis of rare diseases range from six months ([Rare 2030 foresight study](#)) to one year ([IRDiRC](#)) of coming to medical attention.

The landmark Rare Barometer survey on the diagnosis odyssey of people living with a rare disease show that the average diagnosis time is close to 5 years in Europe. This study is the largest conducted to date to measure diagnostic delays for rare diseases and their main determinants, as it encompassed responses from over 13,300 people from 104 countries worldwide, living with more than 1,900 rare diseases.

The results of this survey have been widely communicated: its European results were published in the *European Journal of Human Genetics* (<https://www.nature.com/articles/s41431-024-01604-z>), a factsheet presents key results in 13 languages and dashboards provide detailed results in 27 languages (tiny.cc/RB_diag). Yet, the rare disease community still have questions on how to understand and use some of those results.

In this webinar, the Rare Barometer team will answer the questions addressed to them since the publication of the article by going in depth into its methodology and its results. They will also present how all their publications (article, factsheets and dashboards) have been used and could be used to support the advocacy work and the actions of the rare disease community at large: patient representatives and organisations, healthcare professionals, researchers or companies.

Registration link: https://us02web.zoom.us/meeting/register/ZnMbx4MVQL6GP7qk1o_c0w

14:30-14:35: Welcome and introduction

14:35-14:45: Presentation of the Rare Barometer study on the diagnostic odyssey of people living with a rare disease

Aim, methods, international pilot.

14:45-15:30: Results presented in the peer-reviewed article

<https://www.nature.com/articles/s41431-024-01604-z>

(i) Aims and methods; The diagnosis odyssey: a (ii) long and (iii) difficult journey; (iv) Improving the diagnosis odyssey: main determinants; (v) Consequences of diagnostic delays; (vi) Taking actions.

15:35-15:42: Results presented in the factsheets

tiny.cc/RB_diag

Methods, descriptive statistics, main determinants.

15:42-15:50: Results presented in the dashboards

tiny.cc/RB_diag

Organisation, where to find the relevant information, how to use it.

15:50-16:30: Examples of uses of the results

How EURORDIS and the rare disease community (patient organisations, European Reference Networks, companies) have used and will use the results: concrete examples, tips and tricks.