

MAIN FINDINGS

- **25% of patients had to wait between 5 and 30 years** from early symptoms to confirmatory diagnosis of their disease.
- **40% of patients first received an erroneous diagnosis**, others received none. This led to medical interventions (including surgery and psychiatric treatments) that were based on a wrong diagnosis.
- **25% of patients had to travel to a different region** to obtain the confirmatory diagnosis, and 2% had to travel to a different country.
- **In 33% of cases**, the diagnosis was announced in unsatisfactory terms or conditions. In 12.5% of cases, it was announced in unacceptable ones.
- **The genetic nature of the disease was not communicated to the patient or family in 25% of cases.** This is paradoxical, given the genetic origin of rare diseases.
- **There was genetic counselling in only 50% of cases.**

FOR MORE INFORMATION:

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 (What we do / Eurordis Survey Programme)
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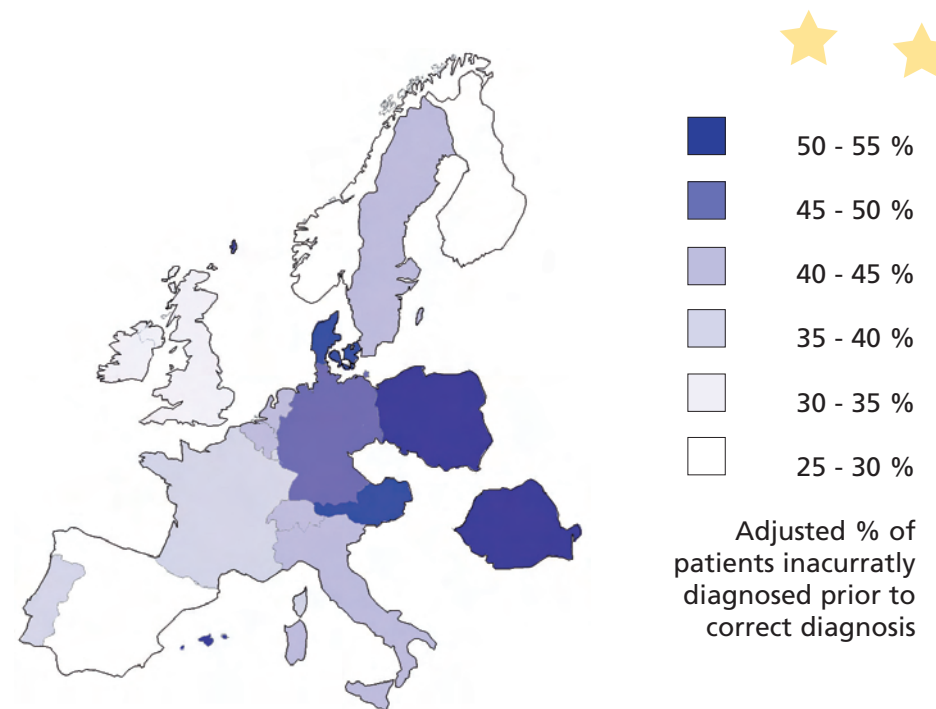


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 Rare Diseases Europe



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SURVEY OF THE DELAY IN DIAGNOSIS FOR 8 RARE DISEASES IN EUROPE ('EURORDISCARE 2')



Diseases and Countries
 Number of respondents by country

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EurordisCare is a series of studies aiming to compare access to care for rare disease patients in Europe.

Comparisons are based on data from a selection of significantly relevant rare diseases across current and future EU Member States. The approach adopted by EurordisCare is unique, because the surveys are based on a **scientific methodology to collect data from people** identified through the EU patient group network. EurordisCare provides solid, quantitative data with each of its surveys, **to help develop public health policies** further.

Objective of the EurordisCare 2 survey

Late diagnoses delay the beginning of adapted treatments and can have severe, irreversible, debilitating and life threatening consequences. Delay in diagnosis can vary greatly depending on the disease and on the country, as well as on individual factors. The survey was launched to provide evidence on the delay to diagnosis and identify its main causes.

Relatively common symptoms can hide underlying rare diseases, leading to misdiagnosis.

The methodology

In collaboration with **8 European rare disease networks** and the **INSERM** Unit of Biostatistics and Biomathematics (French National Institute for Health and Medical Research).

The questionnaire developed for the survey was translated in **12 languages** and mailed to **70 rare disease patient groups** in **17 countries**.

Each patient group then distributed the questionnaires to individual patients or their families. 18,000 questionnaires were sent out, of which **6,000** were returned completed to Eurordis.

The analysis was performed by INSERM and Eurordis; results were shared with participating patient groups and European networks (analysis per disease and per country).

Cross-disease and cross-country comparisons were presented at conferences.

Rare diseases included in the survey

Crohn's disease
Cystic fibrosis
Duchenne muscular dystrophy
Ehlers-Danlos syndrome
Marfan syndrome
Prader Willi syndrome
Tuberous sclerosis
Fragile X syndrome