

Diagnostic delay and clinical pathway among people with undiagnosed rare diseases in Spain

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Background

- Diagnostic delay affects more than half of people with RD
- The average time to obtain a diagnosis is around 6 years
- There is very limited information about people who continue their diagnostic odyssey without a diagnosis

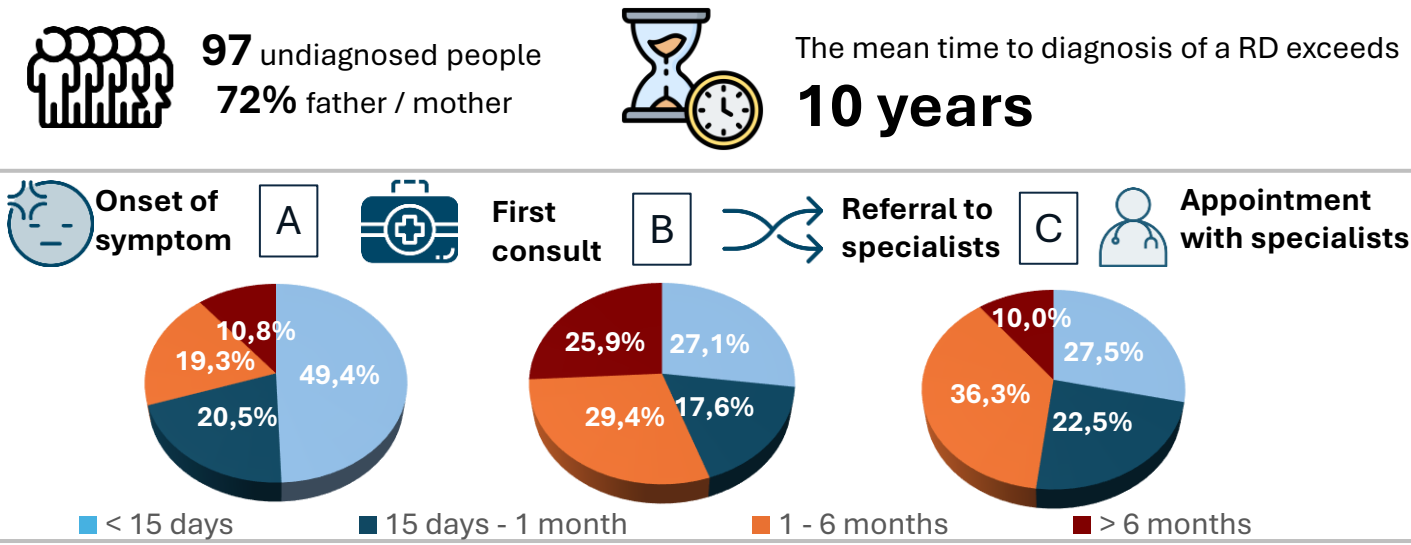
Objective

To understand the diagnostic process of undiagnosed RD patients and to identify potential risk factors that may contribute to a longer diagnostic delay

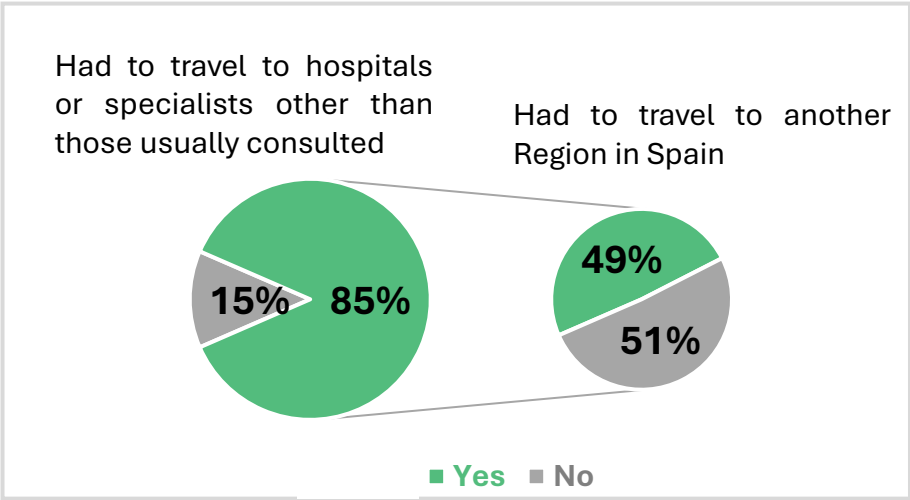
Methods

- Data were obtained from a questionnaire specifically designed for this study and distributed through the Spanish Rare Diseases Patient Registry (<https://registroraras.isciii.es/>).
- All participants are enrolled in the Undiagnosed Rare Diseases Program (SpainUDP) and had not yet obtained a diagnosis at the time of the study.
- A descriptive analysis of the diagnostic process and the time elapsed since the onset of symptoms was conducted

Results



- When symptoms onset, **57%** of people first consulted a **primary care physician**, the main entry point to the National Health System in Spain
- 28%** of people reported **having moved residence**, with **more than half doing so in search of a diagnosis**.



Conclusion

Patients with undiagnosed rare diseases face a complex diagnostic process. Identifying the factors associated with this diagnostic odyssey is necessary in order to implement appropriate public health and health policy measures.