MeetingsPremium
MP Journal of Medical Research
Journal of poster communications · ISSN application in progress
Vol. 1 · 2026-0048
OPEN ACCESS

TYPE
Poster communication

SPECIALTY
Public Health

PRESENTED AT
12th European Conference on Rare Diseases and Orphan Products (ECRD), Brussels, 15–16 May 2024

PUBLISHED
27 October 2026

DOI
10.5281/zenodo.23695402

CORRESPONDENCE
Juan Benito-Lozano
ORCID 0000-0002-5400-9057

KEYWORDS
rare diseases · diagnostic delay · psychological impact · social impact · Spain

LICENSE
CC BY 4.0. © The authors.

POSTER COMMUNICATION · PUBLIC HEALTH

DetERminants and impact of diagnostic delay in people living with a RD in Spain

Juan Benito-Lozano[1]*, Dr Greta Arias-Merino[1], Beatriz Arconada-López[2], Mario Gómez-Martínez[1], Begoña Ruiz García[3], Verónica Alonso Ferreira[1]

[1] Institute of Rare Diseases Research (IIER), Instituto de Salud Carlos III (ISCIII)
[2] Spanish Federation of Rare Diseases (FEDER)
[3] National Reference Center for care of People with Rare Diseases and their Families (Creer), belonging to Institute for Older Persons and Social Services (Imserso)
* Corresponding author: Juan Benito-Lozano

KEY MESSAGES

1. More than half of people with rare diseases in Spain experience diagnostic delay, especially women and those with mental and behavioural disorders
2. Having to travel to see different specialists outside the home province and consulting many specialists increase the risk of diagnostic delay
3. Diagnostic delay is associated with greater psychological and social impact, increasing the need for psychological care

ABSTRACT

Introduction. According to the International Rare Diseases Research Consortium (IRDiRC), all known rare diseases (RD) should be diagnosable within a year. Therefore, to wait more than one year could be considered diagnosis delay. This project aims to know the overall figures of the diagnosis of people affected by a rare disease (RD) in Spain, the clinical determinants that increase the risk of having a delayed diagnosis and their psychological and social impact by comparing their situation with those in whom the diagnosis has arrived in one year or less.

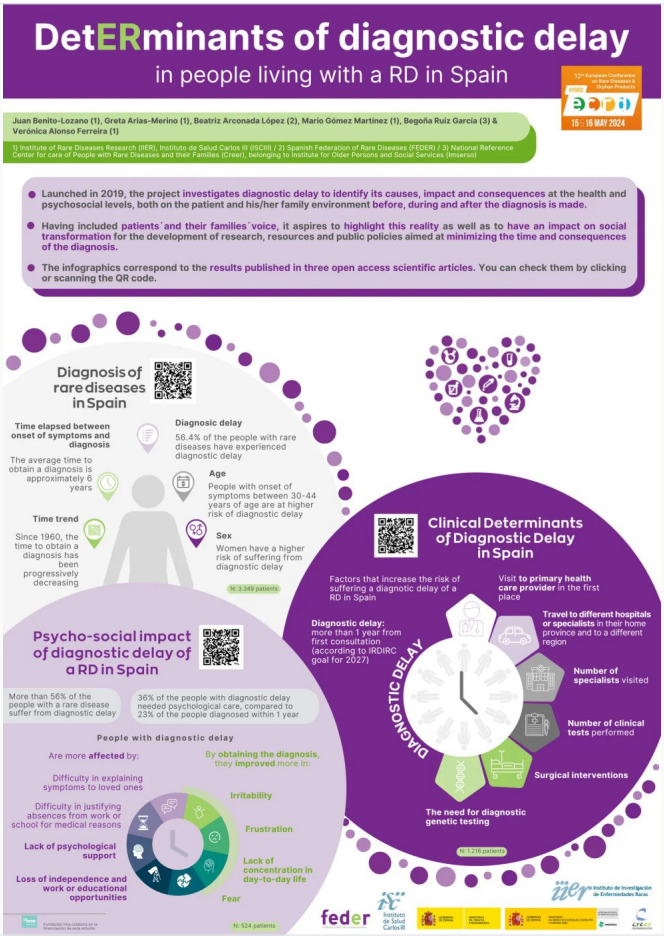
Materials and methods. A purpose-designed form from the Spanish RD Patient Registry for data-collection purposes was used. Three institutions linked to the RD collective in Spain collaborated in the study: IIER-ISCIII (research centre), FEDER (patient advocacy groups) and CREER (public administration).

Results. Main results were published in three open access scientific articles: 56.4% of people had experienced delay in diagnosis of their RDs, being higher in women (OR 1.25; 95% CI 1.07-1.45); those with symptom onset at age 30-44 years (OR 1.48; 95% CI 1.19-1.84); and in mental and behavioural disorders (OR 4.21; 95% CI 2.26-7.85). Some determinants associated with experiencing DD were having to travel to see a specialist other than that usually consulted in the patient's home province (OR 2.1; 95% CI 1.6-2.9); or visiting more than 10 specialists (OR 2.6; 95% CI 1.7-4.0). People who underwent DD experienced greater psychological and social impact than those diagnosed within the first year of disease (36.2% vs 23.2% needed psychological care; p = 0.002).

Conclusion. These findings provide insight into the determinants that have the greatest influence on delayed diagnosis. It will also contribute to develop recommendations to reduce the time required to obtain a diagnosis, as well as correct social inequalities and improve the quality of life of people who live with a RD.

Online record, HD poster
journal.meetingspremium.com/p/2026-0048

POSTER



REFERENCES

1. Benito-Lozano J, Lopez-Villalba B, Arias-Merino G, Posada de la Paz M, Alonso-Ferreira V. Diagnostic delay in Rare Diseases: data from the Spanish Rare Diseases Patient Registry. Orphanet J Rare Dis. 2022;17: 418.

2. Benito-Lozano J, Arias-Merino G, Gómez-Martínez M, Ancochea-Díaz A, Aparicio-García A, Posada de la Paz M, Alonso-Ferreira V. Diagnostic Process in Rare Diseases: Determinants Associated with Diagnostic Delay. Int J Environ Res Public Health. 2022;19(11): 6456.

3. Benito-Lozano J, Arias-Merino G, Gómez-Martínez M, Arconada-López B, Ruiz-García B, Posada de la Paz M, Alonso-Ferreira, V. Psychosocial impact at the time of a rare disease diagnosis. PLoS ONE. 2023;18(7): e0288875.

CITE THIS PUBLICATION

Benito-Lozano J, Arias-Merino G, Arconada-López B, Gómez-Martínez M, Ruiz García B, Alonso Ferreira V. DetERminants and impact of diagnostic delay in people living with a RD in Spain [Poster]. MP J Med Res. 2026;1:2026-0048. doi:10.5281/zenodo.23695402

Vancouver format · APA, BibTeX and RIS on the online record

Original poster in high definition on the online record.

SOURCE
Presented at 12th European Conference on Rare Diseases and Orphan Products (ECRD), Brussels, 15–16 May 2024. Republished with the agreement of the authors.

PROCESS
Record checked by the editorial team, approved by the corresponding author, then reviewed by the journal's editorial board.

INDEPENDENCE
Partners and advertisers take no part in scientific selection.

© The authors. Open access publication under the Creative Commons Attribution 4.0 International (CC BY 4.0) — https://creativecommons.org/licenses/by/4.0/ license. Published by MeetingsPremium LLC, 75 East 3rd Street, Sheridan, WY 82801, United States.