

REFERENCIAS BIBLIOGRÁFICAS

- Alonso, V., Villaverde-Hueso, A., Hens, M., Morales-Piga, A., & Abaitua, I. (2011). Public health research on rare diseases. *Georgian medical news*, (193), 11-16.
- Benito-Lozano, J., López-Villalba, B., Arias-Merino, G., Posada De la Paz, M., & Alonso-Ferreira, V. (2022). Diagnostic delay in rare diseases: data from the Spanish rare diseases, patient registry. *Orphanet Journal of Rare Diseases*, 17(1), 418.
- Benito-Lozano, J., Arias-Merino, G., Gómez-Martínez, M., Arconada-López, B., Ruiz-García, B., Posada De la Paz, M., & Alonso-Ferreira, V. (2023). Psychosocial impact at the time of a rare disease diagnosis. *Plos one*, 18(7), e0288875.
- Bogart, K.R., & Dermody, S. S. (2020). Relationship of rare disorder latent clusters to anxiety and depression symptoms. *Health Psychology*, 39(4), 307.
- EURORDIS (2009). *The Voice of 12.000 Patients. Experiences and Expectations of Rare Disease Patients on Diagnosis and Care in Europe*. Eurordis: Paris, France. ISBN 9782953031812.
- EURORDIS (2017). *Juggling care and daily life: The balancing act of the rare disease community*. EURORDIS Rare Disease Europe. Recuperado de: <https://www.eurordis.org/es/publications/juggling-care-and-daily-life-the-balancing-act-of-the-rare-disease-community-infographic/>
- EURORDIS (2019). *Position paper: Achieving Holistic Person-Centred Care to Leave No One Behind*. EURORDIS Rare Disease Europe. Recuperado de: <https://www.eurordis.org/es/publications/position-paper-achieving-holistic-person-centred-care-to-leave-no-one-behind/>
- EURORDIS (2020). *How has COVID-19 impacted people with rare diseases? Rare Barometer Voices*. EURORDIS survey initiative. Recuperado de: <https://www.eurordis.org/es/publications/how-has-covid-19-impacted-people-with-rare-diseases/>
- EURORDIS (2021). *Rare 2030. Foresight in Rare Disease Policy. Recommendations from the rare 2030 foresight study*. Disponible en: https://www.infocop.es/wp-content/uploads/2024/02/Rare2030_recommendations.pdf
- EURORDIS (2023). *TIME TO ACT – Improving Rare Disease Diagnosis and solving the unsolved Rare Disease through collaboration in Europe*. EURORDIS Rare Disease Europe. Disponible en: <https://www.infocop.es/wp-content/uploads/2024/02/Solve-RD-Time-to-act-RD-diagnosis-in-Europe.pdf>
- Faccio, E., Bottecchia, M., & Rocelli, M. (2023). Caring for People with Rare Diseases: A Systematic Review of the Challenges of, and Strategies for Dealing with, COVID-19. *International Journal of Environmental Research and Public Health*, 20(19), 6863.
- Faviez C, Chen X, Garcelon N, Neuraz A, Knebelmann B, Salomon R, & col. (2020). Diagnosis support systems for rare diseases: a scoping review. *Orphanet Journal of Rare Diseases*, 15(1):94.

FEDER (2009). *Guía de Apoyo Psicológico para Enfermedades Raras*. Federación Española de Enfermedades Raras. Recuperado de: <https://www.infocoponline.es/pdf/Guia-enfermedades-raras.pdf>

FEDER (2021). *Estudio sobre situación de Necesidades Sociosanitarias de las personas con Enfermedades Raras en España. Estudio ENSERio*. Datos actualizados. Federación Española de Enfermedades Raras. Ministerio de Sanidad. Centro de Referencia Estatal de Atención a Personas con Enfermedades Raras y sus Familias. Disponible en: https://www.infocop.es/wp-content/uploads/2024/02/FINAL-ENSERio_Estudio-sobre-situacionde-Necesidades-Sociosanitarias-Personas-con-Enfermedades-Raras-en-Espana.pdf

FEDER (2023). *La mitad de las personas con enfermedades poco frecuentes ha tenido retraso en el diagnóstico*. Noticias. Federación Española de Enfermedades Raras. Recuperado de: <https://www.enfermedades-raras.org/actualidad/noticias/la-mitad-de-las-personas-con-enfermedades-poco-frecuentes-ha-tenido-retraso-en-el-diagnostico>

FEDER (2025). *¿Qué es el Día Mundial de las Enfermedades Raras?* 2025. Federación Española de Enfermedades Raras. Recuperado de: <https://www.enfermedades-raras.org/que-es-el-dia-mundial-de-las-enfermedades-raras-2025>

Giménez-Lozano, C., Páramo-Rodríguez, L., Cavero-Carbonell, C., Corpas-Burgos, F., López-Maside, A., Guardiola-Vilarroig, S., & Zurriaga, O. (2022). Rare Diseases: Needs and Impact for Patients and Families: A Cross-Sectional Study in the Valencian Region, Spain. *International Journal of Environmental Research and Public Health*, 19(16), 10366.

Hedley, V., Bolz-Johnson, M., Hernando, I., Kenward, R., Nababout, R., Romero, C., ... & Upadhyaya, S. (2023). Together4RD position statement on collaboration between European reference networks and industry. *Orphanet Journal of Rare Diseases*, 18(1), 272.

Kenny, T., & Stone, J. (2022). Psychological Support at Diagnosis of a Rare Disease. A Review of the Literature. Rare disease research partners. Recuperado de: www.infocoponline.es/pdf/220202-Literature-Review-Report.pdf

Kenny, T., Bogart, K., Freedman, A., Garthwaite, C., Henley, S. M. D., Bolz-Johnson, M., ... & Woodman, D. (2022). The importance of psychological support for parents and caregivers of children with a rare disease at diagnosis. *Rare Diseases Orphan Drugs*, 1, 7.

Low, C.E., Loke, S., Pang, G.E., Sim, B., and Yang, V.S. (2024). Psychological outcomes in patients with rare cancers: a systematic review and meta-analysis. *eClinicalMedicine*, 4, 72:102631. doi: 10.1016/j.eclim.2024.102631

Mund, M., Uhlenbusch, N., Rillig, F., Weiler-Normann, C., Herget, T., Kubisch, C., ... & Schramm, C. (2023). Psychological distress of adult patients consulting a center for rare and undiagnosed diseases: a cross-sectional study. *Orphanet Journal of Rare Diseases*, 18(1), 1-10.

Nguengang Wakap, S., Lambert, D. M., Olry, A., Rodwell, C., Gueydan, C., Lanneau, V., ... & Rath, A. (2020). Estimating cumulative point prevalence of rare diseases: analysis of the Orphanet database. *European Journal of Human Genetics*, 28(2), 165-173.

OMS (2025, 4 de febrero). Enfermedades raras: una prioridad de salud mundial para la equidad & la inclusión Proyecto de decisión propuesto por el Brasil, Chile, China, Ecuador, Egipto, España, Filipinas, Francia, la India, Iraq, Jordania, Kuwait,

Luxemburgo, Malasia, Pakistán, Palestina, Panamá, Qatar, Rumanía, Somalia y Vanuatu. Punto 6 del Orden del Día. EB156/CONF./2. Consejo Ejecutivo 156.^a reunion. Organización Mundial de la Salud. Recuperado de:
https://apps.who.int/gb/ebwha/pdf_files/EB156/B156_CONF2-sp.pdf

ORPHANET (2023). *Prevalence of rare diseases: Bibliographic data*. Orphanet Report Series (November 2023). Recuperado de:
https://www.orpha.net/pdfs/orphacom/cahiers/docs/GB/Prevalence_of_rare_diseases_by_decreasing_prevalence_or_cases.pdf

Pavisich, K., Jones, H., & Baynam, G. (2024). The Diagnostic Odyssey for Children Living with a Rare Disease-Caregiver and Patient Perspectives: A Narrative Review with Recommendations. Rare. *Science Direct* (Available online 23 February 2024). Recuperado de:
<https://www.sciencedirect.com/science/article/pii/S295000872400005X>

Riera-Mestre, A. (2022). Rare diseases in Spain: a look into the future. *Medicina clínica*, 158(6), 274-276.

Spencer-Tansley, R., Meade, N., Ali, F., Simpson, A., & Hunter, A. (2022). Mental health care for rare disease in the UK—recommendations from a quantitative survey and multi-stakeholder workshop. *BMC Health Services Research*, 22(1), 648.

The Lancet (2024). *The landscape for rare diseases in 2024*. Editorial (March, 2024), 12, Issue 3e341. The Lancet Global Health. Recuperado de:
[https://www.thelancet.com/journals/langlo/article/PIIS2214-109X\(24\)00056-1/fulltext](https://www.thelancet.com/journals/langlo/article/PIIS2214-109X(24)00056-1/fulltext)

Zhang, Z. (2023). Diagnosing rare diseases and mental well-being: a family's story. *Orphanet Journal of Rare Diseases*, 18(1), 1-4.