

REFERENCIAS BIBLIOGRÁFICAS

- Alonso, V., Villaverde-Hueso, A., Hens, M., Morales-Piga, A., y 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., y 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., y Alonso-Ferreira, V. (2023). Psychosocial impact at the time of a rare disease diagnosis. *Plos one*, 18(7), e0288875.
- Bogart, K.R., y 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, y 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>

Giménez-Lozano, C., Páramo-Rodríguez, L., Cavero-Carbonell, C., Corpas-Burgos, F., López-Maside, A., Guardiola-Vilarroig, S., y 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., ... y Upadhyaya, S. (2023). Together4RD position statement on collaboration between European reference networks and industry. *Orphanet Journal of Rare Diseases*, 18(1), 272.

Kenny, T., y 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., ... y 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.

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., ... y Rath, A. (2020). Estimating cumulative point prevalence of rare diseases: analysis of the Orphanet database. *European Journal of Human Genetics*, 28(2), 165-173.

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., y 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., y 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.

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