Selected Published Medical Articles

1. Crossnohere, N.L., Armstrong N., Fischer, R., Bridges, J.F.P. (2022). Diagnostic experiences of Duchenne families and their preferences for newborn screening: A mixed-methods study. *American journal of medical genetics*. Part C, Seminars in medical genetics,190(2):169-177. https://doi.org/10.1002/ajmg.c.31992

2. Crowe, A., McAneney H., Morrison, P.J., Cupples, M.E., McKnight, A.J. (2020). A quick reference guide for rare disease: supporting rare disease management in general practice. *The British journal of general practice: the journal of the Royal College of General Practitioners,* 70(694):260-261. https://doi.org/10.3399/bjgp20X709853

3. Deuitch, N.T., Beckman, E., Halley, M.C., Young, J.L., Reuter, C.M., Kohler, J., Bernstein, J.A., Wheeler, M.T., Undiagnosed Diseases Network, Ormond, K.E., & Tabor, H.K. (2021). "Doctors can read about it, they can know about it, but they've never lived with it": How parents use social media throughout the diagnostic odyssey. *Journal of genetic counseling.* 30(6):1707-1718. https://doi.org/10.1002/jgc4.1438

4. Elliott, E., & Zurynski Y. (2015). Rare diseases are a 'common' problem for clinicians. *Australian family physician,* 44(9):630-3. https://pubmed.ncbi.nlm.nih.gov/26488039/

5. Hulick, P.J. (2023). Next-generation DNA sequencing (NGS): Principle and clinical applications. *UpToDate*. Retrieved December 5, 2023. https://www.uptodate.com/contents/next-generation-dna-sequencing-ngs-principles-and-clinical-applications

6. Kenny, T., Bogart, K., Freedman, A., Garthwaite, C., Henley, S.M.D., et al. (2022). The importance of psychological support for parents and caregivers of children with a rare disease at diagnosis. *Rare disease orphan drugs*, 1, 7. https://dx.doi.org/10.20517/rdodj.2022.04

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9. Quaio, C.R.D.C, Obando, M.J.R., Perazzio, S.F., Dutra, A.P., Chung, C.H., et al. (2021). Exome sequencing and targeted gene panels: a simulated comparison of diagnostic yield using data from 158 patients with rare diseases. *Genetics and molecular biology*, 44(4), 20210061. https://doi.org/10.1590/1678-4685-gmb-2021-0061

10. Seaby, E.G., Pengelly, R.J., & Ennis, S. (2016). Exome sequencing explained: a practical guide to its clinical application. *Briefings in functional genomics,* 15(5), 374–384. https://doi.org/10.1093/bfgp/elv054

11. Vandeborne, L., van Overbeeke, E., Dooms, M., De Beleyr, B., & Huys, I. (2019). Information needs of physicians regarding the diagnosis of rare diseases: a questionnaire-based study in Belgium. *Orphanet journal of rare diseases,* 14(1), 99. https://doi.org/10.1186/s13023-019-1075-8

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