

Argyriou, Catherine, et al. "AAV-Mediated PEX1 Gene Augmentation Improves Visual Function in the PEX1-Gly844Asp Mouse Model for Mild Zellweger Spectrum Disorder." *Molecular Therapy - Methods & Clinical Development*, vol.23, Dec. 2021, pp. 225–40. DOI.org (Crossref), <https://doi.org/10.1016/j.omtm.2021.09.002>.

Berendse, K., Klouwer, F. C. C., Koot, B. G. P., Kemper, E. M., Ferdinandusse, S., Koelfat, K. V. K., Lenicek, M., Schaap, F. G., Waterham, H. R., Vaz, F. M., Engelen, M., Jansen, P. L. M., Wanders, R. J. A., & Poll-The, B. T. (2016). Cholic acid therapy in Zellweger spectrum disorders. *Journal of Inherited Metabolic Disease*, 39(6), 859–868. <https://doi.org/10.1007/s10545-016-9962-9>

De Biase, I., Tortorelli, S., Kratz, L., J. Steinberg, S., Cusmano-Ozog, K., & Braverman, N. (2020). Laboratory diagnosis of disorders of peroxisomal biogenesis and function: A technical standard of the American College of Medical Genetics and Genomics (ACMG). *Genetics in Medicine*, 22(4), 686–697. <https://doi.org/10.1038/s41436-019-0713-9>

Berendse, Kevin, et al. "High Prevalence of Primary Adrenal Insufficiency in Zellweger Spectrum Disorders." *Orphanet Journal of Rare Diseases*, vol. 9, Sep. 2014, p. 133. PubMed Central, <https://doi.org/10.1186/s13023-014-0133-5>.

Bose, M., Cuthbertson, D. D., Fraser, M. A., Rouillet, J.-B., Gibson, K. M., Schules, D. R., Gawron, K. M., Gamble, M. B., Sacra, K. M., Lopez, M. J., & Rizzo, W. B. (2020). Zellweger spectrum disorder: A cross-sectional study of symptom prevalence using input from family caregivers. *Molecular Genetics and Metabolism Reports*, 25, 100694. <https://doi.org/10.1016/j.ymgmr.2020.100694>

Braverman, N. E., Raymond, G. V., Rizzo, W. B., Moser, A. B., Wilkinson, M. E., Stone, E. M., Steinberg, S. J., Wangler, M. F., Rush, E. T., Hacia, J. G., & Bose, M. (2016). Peroxisome biogenesis disorders in the Zellweger spectrum: An overview of current diagnosis, clinical manifestations, and treatment guidelines. *Molecular Genetics and Metabolism*, 117(3), 313–321. <https://doi.org/10.1016/j.ymgme.2015.12.009>

Ferdinandusse, Sacha, et al. "The Important Role of Biochemical and Functional Studies in the Diagnostics of Peroxisomal Disorders." *Journal of Inherited Metabolic Disease*, vol. 39, no. 4, Jul. 2016, pp. 531–43. DOI.org (Crossref), <https://doi.org/10.1007/s10545-016-9922-4>.

Klouwer, F. C. C., Berendse, K., Ferdinandusse, S., Wanders, R. J. A., Engelen, M., & Poll-The, B.T. (2015). Zellweger spectrum disorders: Clinical overview and management approach. *Orphanet Journal of Rare Diseases*, 10(1), 151. <https://doi.org/10.1186/s13023-015-0368-9>

Martinez, M. (1992). Abnormal profiles of polyunsaturated fatty acids in the brain, liver, kidney and retina of patients with peroxisomal disorders. *Brain Research*, 583(1–2), 171–182. [https://doi.org/10.1016/s0006-8993\(10\)80021-6](https://doi.org/10.1016/s0006-8993(10)80021-6)

Paker, A. M., Sunness, J. S., Brereton, N. H., Speedie, L. J., Albanna, L., Dharmaraj, S., Moser, A. B., Jones, R. O., & Raymond, G. V. (2010). Docosahexaenoic acid therapy in peroxisomal diseases. *Neurology*, 75(9), 826–830. <https://doi.org/10.1212/WNL.0b013e3181f07061>

Steinberg, S. J., Raymond, G. V., Braverman, N. E., & Moser, A. B. (1993). Zellweger Spectrum Disorder. In M. P. Adam, J. Feldman, G. M. Mirzaa, R. A. Pagon, S. E. Wallace, & A. Amemiya (Eds.), *GeneReviews*®. University of Washington, Seattle. <http://www.ncbi.nlm.nih.gov/books/NBK1448/>

Steinberg, S. J., Dodt, G., Raymond, G. V., Braverman, N. E., Moser, A. B., & Moser, H. W. (2006). Peroxisome biogenesis disorders. *Biochimica Et Biophysica Acta*, 1763(12), 1733–1748. <https://doi.org/10.1016/j.bbamcr.2006.09.010>

Volmrich, A.M., Cuénant, L.M., Forghani, I., Hsieh, S.L., Shapiro, L.T. (2022). ABCD1 Gene Mutations: Mechanisms and Management of Adrenomyeloneuropathy. *Appl Clin Genet.*, 15, 111-123. <https://www.tandfonline.com/doi/full/10.2147/TACG.S359479>

Wanders, R. J. A., & Waterham, H. R. (2006). Peroxisomal disorders: The single peroxisomal enzyme deficiencies. *Biochimica et Biophysica Acta (BBA) - Molecular Cell Research*, 1763(12), 1707–1720. <https://doi.org/10.1016/j.bbamcr.2006.08.010>

Waterham, H. R., Ferdinandusse, S., & Wanders, R. J. A. (2016). Human disorders of peroxisome metabolism and biogenesis. *Biochimica et Biophysica Acta (BBA) - Molecular Cell Research*, 1863(5), 922–933. <https://doi.org/10.1016/j.bbamcr.2015.11.015>

Waterham, H. R., & Ebberink, M. S. (2012). Genetics and molecular basis of human peroxisome biogenesis disorders. *Biochimica et Biophysica Acta (BBA) - Molecular Basis of Disease*, 1822(9), 1430–1441. <https://doi.org/10.1016/j.bbadis.2012.04.006>