

Neuronal and glial cell alterations involved in the retinal degeneration of the familial dysautonomia optic neuropathy.

Schultz A, Albertos-Arranz H, Sáez XS, Morgan J, Darland DC, Gonzalez-Duarte A, Kaufmann H, Mendoza-Santiesteban CE, Cuenca N, Lefcort F. *Glia*. 2024 Dec;72(12):2268-2294. doi: 10.1002/glia.24612. Epub 2024 Sep 3. PMID: 39228100

Metabolic Deficits in the Retina of a Familial Dysautonomia Mouse Model.

Costello SM, Schultz A, Smith D, Horan D, Chaverra M, Tripet B, George L, Bothner B, Lefcort F, Copié V. *Metabolites*. 2024 Jul 31;14(8):423. doi: 10.3390/metabo14080423. PMID: 39195519 **Free PMC article.**

ELP1, the Gene Mutated in Familial Dysautonomia, Is Required for Normal Enteric Nervous System Development and Maintenance and for Gut Epithelium Homeostasis.

Chaverra M, Cheney AM, Scheel A, Miller A, George L, Schultz A, Henningsen K, Kominsky D, Walk H, Kennedy WR, Kaufmann H, Walk S, Copié V, Lefcort F. *J Neurosci*. 2024 Sep 11;44(37):e2253232024. doi: 10.1523/JNEUROSCI.2253-23.2024. PMID: 39138000

Elp1 function in placode-derived neurons is critical for proper trigeminal ganglion development.

Hines MA, Taneyhill LA. *Dev Dyn*. 2024 Oct 9. doi: 10.1002/dvdy.749. Online ahead of print. PMID: 39381860

Age-dependent regulation of ELP1 exon 20 splicing in Familial Dysautonomia by RNA Polymerase II kinetics and chromatin structure.

Riccardi F, Romano G, Licastro D, Pagani F. *PLoS One*. 2024 Jun 3;19(6):e0298965. doi: 10.1371/journal.pone.0298965. eCollection 2024. PMID: 38829854 **Free PMC article.**

Parasympathetic neurons derived from human pluripotent stem cells model human diseases and development.

Wu HF, Saito-Diaz K, Huang CW, McAlpine JL, Seo DE, Magruder DS, Ishan M, Bergeron HC, Delaney WH, Santori FR, Krishnaswamy S, Hart GW, Chen YW, Hogan RJ, Liu HX, Ivanova NB, Zeltner N. *Cell Stem Cell*. 2024 May 2;31(5):734-753.e8. doi: 10.1016/j.stem.2024.03.011. Epub 2024 Apr 11. PMID: 38608707

Retrograde tubing as a rescue treatment for megaesophagus: a case report.

Weiss CS, Demma JA, Koplewitz B, Maayan C, Slae M. *BMJ Open Gastroenterol*. 2024 Mar 27;11(1):e001285. doi: 10.1136/bmjgast-2023-001285. PMID: 38538089 **Free PMC article.**

Linking the Extended Autonomic System with the Homeostat Theory: New Perspectives about Dysautonomias.

Goldstein DS. *J Pers Med*. 2024 Jan 22;14(1):123. doi: 10.3390/jpm14010123. PMID: 38276245 **Free PMC article.**

Transcriptome analysis in a humanized mouse model of familial dysautonomia reveals tissue-specific gene expression disruption in the peripheral nervous system.
Harripaul R, Morini E, Salani M, Logan E, Kirchner E, Bolduc J, Chekuri A, Currall B, Yadav R, Erdin S, Talkowski ME, Gao D, Slaugenhaupt S. *Sci Rep.* 2024 Jan 4;14(1):570. doi: 10.1038/s41598-023-51137-6. PMID: 38177237 **Free PMC article.**

Transcutaneous vagus nerve stimulation as a potential novel treatment for cyclic vomiting syndrome: a first case report.
Carandina A, Scatà C, Furlan L, Bellocchi C, Tobaldini E, Montano N. *Clin Auton Res.* 2024 Feb;34(1):209-212. doi: 10.1007/s10286-023-01002-3. Epub 2023 Dec 19. PMID: 38110825 **Free PMC article.**

The Relationship Between Scoliosis, Spinal Bone Density, and Truncal Muscle Strength in Familial Dysautonomia Patients.
Yovchev I, Maayan C, Simanovsky N, Foldes AJ, Brooks R, Kaplan L, Meiner Z, Cheishvili D. *Calcif Tissue Int.* 2024 Mar;114(3):222-227. doi: 10.1007/s00223-023-01164-2. Epub 2023 Nov 29. PMID: 38030713

Height, weight, and body mass index in patients with familial dysautonomia.
Cotrina ML, Morgenstein B, Perez M, Norcliffe-Kaufmann L, Palma JA, Kaufmann H. *PLoS One.* 2023 Nov 9;18(11):e0293800. doi: 10.1371/journal.pone.0293800. eCollection 2023. PMID: 37943786 **Free PMC article.**

Reduction of retinal ganglion cell death in mouse models of familial dysautonomia using AAV-mediated gene therapy and splicing modulators.
Schultz A, Cheng SY, Kirchner E, Costello S, Miettinen H, Chaverra M, King C, George L, Zhao X, Narasimhan J, Weetall M, Slaugenhaupt S, Morini E, Punzo C, Lefcort F. *Sci Rep.* 2023 Oct 30;13(1):18600. doi: 10.1038/s41598-023-45376-w. PMID: 37903840 **Free PMC article.**

Development of an oral treatment that rescues gait ataxia and retinal degeneration in a phenotypic mouse model of **familial dysautonomia**.
Morini E, Chekuri A, Logan EM, Bolduc JM, Kirchner EG, Salani M, Krauson AJ, Narasimhan J, Gabbeta V, Grover S, Dakka A, Mollin A, Jung SP, Zhao X, Zhang N, Zhang S, Arnold M, Woll MG, Naryshkin NA, Weetall M, Slaugenhaupt SA. *Am J Hum Genet.* 2023 Mar 2;110(3):531-547. doi: 10.1016/j.ajhg.2023.01.019. Epub 2023 Feb 20. PMID: 36809767