



## Rebecca Levy

Instructor, Pediatric Neurology

### CLINICAL OFFICE (PRIMARY)

- **Pediatric Neurology**

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### Bio

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#### CLINICAL FOCUS

- Neurology with Special Qualifications in Child Neurology

#### ACADEMIC APPOINTMENTS

- Instructor, Pediatric Neurology
- Member, Maternal & Child Health Research Institute (MCHR)

#### PROFESSIONAL EDUCATION

- Board Certification: Clinical Genetics and Genomics, American Board of Medical Genetics and Genomics (2023)
- Board Certification: Neurology with Special Qualifications in Child Neurology, American Board of Psychiatry and Neurology (2020)
- Fellowship: Stanford University Division of Medical Genetics (2022) CA
- Residency: Stanford University Division of Medical Genetics (2022) CA
- Residency: Stanford University Child Neurology Residency (2020) CA
- Medical Education: Columbia University College of Physicians and Surgeons (2015) NY
- PhD, Columbia University , Neurobiology
- MD, Columbia University
- BA, Yale University , Neuroscience

### Publications

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#### PUBLICATIONS

- **De Novo Variants Associated With Autosomal Recessive Conditions: Case Series and Implications for Genetic Testing and Counseling.** *American journal of medical genetics. Part A*  
Niehaus, A. D., Bonner, D. E., Carter, J., Avello, K., Jacob, N., Neu, M. B., Mendez, R., Qiao, W., Scott, S. A., Levy, R. J., Mattas, L., Schymick, J., Van Andel, et al  
2026
- **Emerging Topics in Neurogenomics: Summary From Inaugural Child Neurology Society Genetics Summit.** *Pediatric neurology*

- Sen, K., Gottlieb-Smith, R., Amin, S., Ananth, A., Bartlett, V., Bass, N., Brock, D., Conrad, E., Ceulemans, S., Davids, L., Dhamija, R., Dills, S., Goldstein, et al  
2026; 180: 59-65
- **Biallelic variants in the noncoding RNA gene RNU4-2 cause a recessive neurodevelopmental syndrome with distinct white matter changes.** *Nature genetics*  
Rius, R., Blakes, A. J., Chen, Y., De Jonghe, J., Lecoquierre, F., Dawes, R., Cogne, B., Kim, H. C., Alvi, J. R., Amblard, F., Ansari, M., Arlt, A., Austin-Tse, et al  
2026
  - **Biallelic Variants in RNU6ATAC Result in a Minor Spliceopathy Characterized by Transcriptome-Wide Minor Intron Retention Events and Short Stature with Variable Multisystem Manifestations.** *HGG advances*  
Mendez, R., Arriaga, T. M., Ma, J., Bonner, D. E., Emami, S., Levy, R. J., Alsagheir, A., Alhaddad, B., Bakur, K., Ungar, R. A., Matalon, D. R., Miller, A. M., Nguyen, et al  
2026: 100588
  - **Timothy Syndrome and CACNA1C-Related Disorder: First International Language and Management Guidelines Consensus Statement.** *Research square*  
Underwood, J. F., Timothy, K. W., Tyroll, H., Levy, R. J., Dick, I. E., Pitt, G. S., Tunbridge, E. M., Baban, A., Wilkinson, G., Hall, N. A., Brugada, G. S., Bauer, R., Abrams, et al  
2025
  - **A National Survey on the Neurogenetics Workforce: Practice Models, Evolving Challenges, and Future Directions.** *Pediatric neurology*  
Taylor, A., Levy, R. J., Dhamija, R., Sen, K.  
2025; 169: 52-58
  - **Prevalence of Sleep Concerns and Sleep Hypnotic Use in Children with Rare Neurogenetic Conditions**  
MacLean, J., Mattas, L., Levy, R.  
OXFORD UNIV PRESS INC.2025: A443
  - **From Organoids to Assembloids: Experimental Approaches to Study Human Neuropsychiatric Disorders.** *Annual review of neuroscience*  
Levy, R. J., Paşca, S. P.  
2025
  - **Human assembloid model of the ascending neural sensory pathway.** *Nature*  
Kim, J. I., Imaizumi, K., Jurjuţ, O., Kelley, K. W., Wang, D., Thete, M. V., Hudacova, Z., Amin, N. D., Levy, R. J., Scherrer, G., Paşca, S. P.  
2025
  - **DDX3X-related neurodevelopmental disorder in males - presenting a new cohort of 19 males and a literature review.** *European journal of human genetics : EJHG*  
Kennis, M. G., Rots, D., Bouman, A., Ockeloen, C. W., Boelen, C., Marcellis, C. L., de Vries, B. B., Elting, M. W., Waisfisz, Q., Suri, M., Font-Montgomery, E., Peck, D. S., Donnelly, et al  
2025
  - **Pathogenic de novo variants in PPP2R5C cause a neurodevelopmental disorder within the Houge-Janssens syndrome spectrum.** *American journal of human genetics*  
Verbinnen, I., Douzgou Houge, S., Hsieh, T., Lesmann, H., Kirchhoff, A., Genevieve, D., Brimble, E., Lenaerts, L., Haesen, D., Levy, R. J., Thevenon, J., Faivre, L., Marco, et al  
2025
  - **Expanding the phenotype and genotype spectrum of TAOK1 neurodevelopmental disorder and delineating TAOK2 neurodevelopmental disorder.** *Genetics in medicine : official journal of the American College of Medical Genetics*  
Elkhateeb, N., Crookes, R., Spiller, M., Pavinato, L., Palermo, F., Brusco, A., Parker, M., Park, S. M., Mendes, A. C., Saraiva, J. M., Hammer, T. B., Nazaryan-Petersen, L., Barakat, et al  
2024: 101348
  - **Clinical study of ferredoxin-reductase-related mitochondriopathy: Genotype-phenotype correlation and proposal of ancestry-based carrier screening in the Mexican population.** *Genetics in medicine open*  
Campbell, T., Slone, J., Metzger, H., Liu, W., Sacharow, S., Yang, A., Moosajee, M., La Morgia, C., Carelli, V., Palombo, F., Lines, M. A., Innes, A. M., Levy, et al  
2024; 2: 100841

- **Pearls & Oy-sters: Exquisite Response of Sleep-Related Hypermotor Epilepsy to a Nicotine Patch.** *Neurology*  
Nam, S., Von Stein, E. L., Meador, K. J., Levy, R. J., Gallentine, W., Li, Y.  
2024; 103 (7): e209790
- **Ocular features of NGLY1 deficiency from a prospective longitudinal cohort.** *Journal of AAPOS : the official publication of the American Association for Pediatric Ophthalmology and Strabismus*  
Frater, C. H., Ruzhnikov, M. R., Beres, S., Alcorn, D., Shue, A., Levy, R. J.  
2024: 103925
- **De novo FRMD5 Missense Variants in Patients with Childhood-Onset Ataxia, Prominent Nystagmus, and Seizures.** *Movement disorders : official journal of the Movement Disorder Society*  
Keller Sarmiento, I. J., Bustos, B. I., Blackburn, J., Hac, N. E., Ruzhnikov, M., Monroe, M., Levy, R. J., Kinsley, L., Li, M., Silani, V., Lubbe, S. J., Krainc, D., Mencacci, et al  
2024
- **Antisense oligonucleotide therapeutic approach for Timothy syndrome.** *Nature*  
Chen, X., Birey, F., Li, M. Y., Revah, O., Levy, R., Thete, M. V., Reis, N., Kaganovsky, K., Onesto, M., Sakai, N., Hudacova, Z., Hao, J., Meng, et al  
2024; 628 (8009): 818-825
- **A Cross-Sectional Study of the Neuropsychiatric Phenotype of CACNA1C-Related Disorder**  
Levy, R., Timothy, K., Underwood, J., Hall, J., Bernstein, J., Pasca, S.  
LIPPINCOTT WILLIAMS & WILKINS.2023
- **What Have Organoids and Assembloids Taught Us About the Pathophysiology of Neuropsychiatric Disorders?** *Biological psychiatry*  
Levy, R. J., Paşca, S. P.  
2022
- **A Cross-Sectional Study of the Neuropsychiatric Phenotype of CACNA1C-Related Disorder.** *Pediatric neurology*  
Levy, R. J., Timothy, K. W., Underwood, J. F., Hall, J., Bernstein, J. A., Paşca, S. P.  
2022; 138: 101-106
- **Genetic Testing to Inform Epilepsy Treatment Management From an International Study of Clinical Practice.** *JAMA neurology*  
McKnight, D., Morales, A., Hatchell, K. E., Bristow, S. L., Bonkowsky, J. L., Perry, M. S., Berg, A. T., Borlot, F., Esplin, E. D., Moretz, C., Angione, K., Rios-Pohl, L., Nussbaum, et al  
2022
- **Delineating the Epilepsy Phenotype of NGLY1 Deficiency.** *Journal of inherited metabolic disease*  
Levy, R. J., Frater, C. H., Gallentine, W. B., Phillips, J. M., Ruzhnikov, M. R.  
2022
- **Evaluation of Seizure Risk in Infants After Cardiopulmonary Bypass in the Absence of Deep Hypothermic Cardiac Arrest.** *Neurocritical care*  
Levy, R. J., Mayne, E. W., Sandoval Karamian, A. G., Iqbal, M., Purington, N., Ryan, K. R., Wusthoff, C. J.  
2021
- **Aicardi-Goutières syndrome may present with positive newborn screen for X-linked adrenoleukodystrophy.** *American journal of medical genetics. Part A*  
Tise, C. G., Morales, J. A., Lee, A. S., Velez-Bartolomei, F. n., Floyd, B. J., Levy, R. J., Cusmano-Ozog, K. P., Feigenbaum, A. S., Ruzhnikov, M. R., Lee, C. U., Enns, G. M.  
2021
- **Acute Hyperextension "Surfer's" Myelopathy in a Gymnast: Bending over Backwards for Diagnosis**  
Levy, R., Guimaraes, C., Partap, S.  
LIPPINCOTT WILLIAMS & WILKINS.2020
- **Continuous EEG for Seizure Detection in Neonates after Cardiac Bypass without Deep Hypothermic Cardiac Arrest**  
Levy, R., Karamian, A., Mayne, E., Iqbal, M., Purington, N., Ryan, K., Wusthoff, C.  
LIPPINCOTT WILLIAMS & WILKINS.2020
- **A Previously Healthy Adolescent With Acute Psychosis and Severe Hyperhidrosis.** *Pediatrics*  
Rosenblatt, T. n., Ort, K. n., Shaw, R. n., Levy, R. J., Chen, C. n., Niemi, A. n., Hoang, K. n.

2020

- **Differentiation and maturation of oligodendrocytes in human three-dimensional neural cultures** *NATURE NEUROSCIENCE*  
Marton, R. M., Miura, Y., Sloan, S. A., Li, Q., Revah, O., Levy, R. J., Huguenard, J. R., Pasca, S. P.  
2019; 22 (3): 484-+
- **Deoxycytidine and deoxythymidine treatment for thymidine kinase 2 deficiency.** *Annals of neurology*  
Lopez-Gomez, C., Levy, R. J., Sanchez-Quintero, M. J., Juanola-Falgarona, M., Barca, E., Garcia-Diaz, B., Tadesse, S., Garone, C., Hirano, M.  
2017
- **Deletion of *Rapgef6*, a candidate schizophrenia susceptibility gene, disrupts amygdala function in mice** *TRANSLATIONAL PSYCHIATRY*  
Levy, R. J., Kvajo, M., Li, Y., Tsvetkov, E., Dong, W., Yoshikawa, Y., Kataoka, T., Bolshakov, V. Y., Karayiorgou, M., Gogos, J. A.  
2015; 5
- **Long Survival in Patients With Leigh Syndrome and the m.10191T > C Mutation in MT-ND3: A Case Report and Review of the Literature** *JOURNAL OF CHILD NEUROLOGY*  
Levy, R. J., Rios, P. G., Akman, H. O., Sciacco, M., De Vivo, D. C., DiMauro, S.  
2014; 29 (10): NP105-NP110
- **Copy number variation and psychiatric disease risk.** *Methods in molecular biology (Clifton, N.J.)*  
Levy, R. J., Xu, B., Gogos, J. A., Karayiorgou, M.  
2012; 838: 97-113
- **Altered axonal targeting and short-term plasticity in the hippocampus of *Disc1* mutant mice** *PROCEEDINGS OF THE NATIONAL ACADEMY OF SCIENCES OF THE UNITED STATES OF AMERICA*  
Kvajo, M., McKellar, H., Drew, L. J., Lepagnol-Bestel, A., Xiao, L., Levy, R. J., Blazeski, R., Arguello, P. A., Lacefield, C. O., Mason, C. A., Simonneau, M., O'Donnell, J. M., MacDermott, et al  
2011; 108 (49): E1349-E1358
- **A Case of Abulia From Left Middle Cerebral Artery Stroke in an Adolescent Treated Successfully With Short Duration Olanzapine.** *Clinical neuropharmacology*  
Connor, A. T., Crawford, A. n., Levy, R. J., Schneider, L. M., Hollander, S. A., Shaw, R. J.  
; 43 (3): 86-89