



Birgitt Schuele

Associate Professor (Research) of Pathology and, by courtesy, of Neurology and Neurological Sciences

Bio

BIO

Birgitt Schüle, MD, is an Associate Professor in the Department of Pathology, and by courtesy, Neurology at Stanford University School of Medicine. Her research focuses on medical genetics and stem cell modeling to uncover disease mechanisms and pathways involved in neurodegeneration in Parkinson's disease and related disorders. She is dedicated to developing novel therapeutic strategies that contribute to the advancement of precision medicine.

Dr. Schüle obtained her medical training from the Georg-August University Göttingen and Medical University Lübeck, Germany, between 1993 and 2001. She earned her doctoral degree in medicine (Dr. med.) in neurophysiology from the Georg-August University Göttingen in 2001. During her neurology internship from 2001 to 2002 at the Medical University of Lübeck under the guidance of Prof. Christine Klein. Subsequently, she pursued a postdoctoral fellowship in human genetics with Prof. Uta Francke at Stanford University School of Medicine from 2003 to 2005. From 2005 to 2019, Dr. Schüle demonstrated leadership in spearheading critical clinical research programs and establishing essential biospecimen repositories for neurogenetics, translational stem cell research, and brain donation at the Parkinson's Institute and Clinical Center.

Since joining Stanford in 2019, she has served as Associate Core Leader for Neuropathology and the Research and Education Component (REC) within the Stanford Alzheimer's Disease Research Center (ADRC). Her contributions to the ADRC include establishing a Stem Cell Program that supports a human induced pluripotent stem cell and post-mortem leptomeninges tissue bank. These resources are shared with repositories at the National Institutes of Health (NIH) and others, facilitating collaborative research and advancing understanding of neurodegenerative diseases.

Her global health work focuses on improving equitable access to neurogenetic diagnosis and care for patients and families with spinocerebellar ataxia type 10 (SCA10) in Peru, Brazil, and Mexico. Through long-standing collaborations in Latin America, she has helped build a program that integrates patient recruitment, return of results, genetic counseling, and access to rare disease care networks, serving as a model for how genomics can expand access and strengthen care for patients with rare disease.

ACADEMIC APPOINTMENTS

- Associate Professor (Research), Pathology
- Associate Professor (Research) (By courtesy), Adult Neurology
- Member, Bio-X
- Member, SPARK at Stanford
- Member, Wu Tsai Neurosciences Institute

ADMINISTRATIVE APPOINTMENTS

- Investigator, Center for Definitive and Curative Medicine, Stanford University School of Medicine, (2023- present)

- Member, PREP Committee, Stanford ADRC, (2021- present)
- Mentor, Pathways to Neurosciences Program, (2021- present)
- Member, Clin Gen, Clinical Domain Working Groups, Parkinson's Disease Gene Curation Expert Panel, (2020- present)
- Member, Diversity and Inclusion Committee, Department Pathology, (2020-2023)

PROFESSIONAL EDUCATION

- Dr. med., Georg-August University Goettingen, Germany , Neurophysiology (2001)
- MD, Medical University Luebeck, Germany , Human Medicine (2001)

COMMUNITY AND INTERNATIONAL WORK

- Clinico-genetic studies in spinocerebellar ataxia 10 (SCA-10) in Mexico, Mexico, Peru, Brazil

LINKS

- Schuele lab website: <https://theschuelelab.weebly.com/>

Research & Scholarship

CURRENT RESEARCH AND SCHOLARLY INTERESTS

The Schuele lab focuses on neurogenetics, human stem cell modeling, and gene therapy approaches to uncover disease mechanisms and pathways involved in neurodegeneration in Parkinson's, Alzheimer's disease and related disorders.

Teaching

STANFORD ADVISEES

Doctoral Dissertation Reader (AC)

Jack Marciano

GRADUATE AND FELLOWSHIP PROGRAM AFFILIATIONS

- Human Genetics and Genetic Counseling (Masters Program)
- Neurosciences (Phd Program)

Publications

PUBLICATIONS

- **Prevention of transgene silencing during human pluripotent stem cell differentiation.** *Cell stem cell*
Uenaka, T., Napole, A., Saha, A. D., Sun, D., Singavarapu, A., Calzada, E., Chen, J., Erlebach, L., McQuade, A., Ramos, D. M., Rigamonti, A., Salazar, L., Samelson, et al
2026
- **Functional connectivity alterations in spinocerebellar ataxia type 10: insights from gray matter atrophy.** *Brain imaging and behavior*
Padron-Rivera, G., Ramirez-Garcia, G., Chirino-Perez, A., Romero-Molina, A. O., Ochoa-Morales, A., Garcia-Gomar, M. G., Ramirez-Garcia, M. A., Rodriguez-Mendoza, O., Torres-Vences, D. L., Schule, B., Pasaye-Alcaraz, E. H., Hernandez-Castillo, C. R., Fernandez-Ruiz, et al
2026; 20 (1): 3
- **Novel ATXN10 Repeat Motif Patterns in Peruvian Families Modify Disease Onset.** *Neurology. Genetics*
Sedov, K., Manrique-Enciso, C., Yang, M. J., Araujo-Aliaga, I., Dolzhenko, E., Kalla, S., Kingan, S. B., Sarapura-Castro, E., Valdivia, A. R., Illanes-Manrique, M. Z., Cornejo-Olivas, M., Schule, B.
2025; 11 (6): e200316
- **Heartbeat signature for predicting motor and non-motor involvement among nonparkinsonian LRRK2 G2019S mutation carriers.** *Clinical autonomic research : official journal of the Clinical Autonomic Research Society*

Carricarte Naranjo, C., Marras, C., Visanji, N. P., Cornforth, D. J., Sanchez-Rodriguez, L., Schule, B., Goldman, S. M., Estevez, M., Stein, P. K., Jelinek, H. F., Lang, A. E., Machado, A.
2025

- **Novel Intermediate ATXN10 Alleles in the Healthy Peruvian Population: A Matter of Indigenous American Ethnic Origin.** *Cerebellum (London, England)*
Milla-Neyra, K., Araujo-Aliaga, I., Manrique-Enciso, C., Sarapura-Castro, E., Illanes-Manrique, M., Veliz-Otani, D., Saldarriaga-Mayo, A., Medina-Colque, A., Rios-Pinto, J., Cornejo-Herrera, I., Rivera-Valdivia, A., F Mata, I., Loesch, et al
2025; 24 (2): 44
- **Alpha-Synuclein Gene Alterations Modulate Tyrosine Hydroxylase in Human iPSC-Derived Neurons in a Parkinson's Disease Animal Model.** *Life (Basel, Switzerland)*
Bernal-Conde, L. D., Pena-Martinez, V., Morato-Torres, C. A., Ramos-Acevedo, R., Arias-Carrion, O., Padilla-Godinez, F. J., Delgado-Gonzalez, A., Palomero-Rivero, M., Collazo-Navarrete, O., Soto-Rojas, L. O., Gomez-Chavarin, M., Schule, B., Guerra-Crespo, et al
2024; 14 (6)
- **Identification and validation of N-acetylputrescine in combination with non-canonical clinical features as a Parkinson's disease biomarker panel.** *Scientific reports*
Peng, K., Klotz, A., Guven, A., Kapadnis, U., Ravipaty, S., Tolstikov, V., Vemulapalli, V., Rodrigues, L. O., Li, H., Kellogg, M. D., Kausar, F., Rees, L., Sarangarajan, et al
2024; 14 (1): 10036
- **Inactive S. aureus Cas9 downregulates alpha-synuclein and reduces mtDNA damage and oxidative stress levels in human stem cell model of Parkinson's disease.** *Scientific reports*
Sastre, D., Zafar, F., Torres, C. A., Piper, D., Kirik, D., Sanders, L. H., Qi, L. S., Schüle, B.
2023; 13 (1): 17796
- **Automated 384-well SYBR Green Expression Array for Optimization of Human Induced Pluripotent Stem Cell Differentiation** *BIO-PROTOCOL*
Chen, M. Y., Heinrich, L., Zafar, F., Sedov, K., Schule, B.
2023; 13 (11)
- **Automated 384-well SYBR Green Expression Array for Optimization of Human Induced Pluripotent Stem Cell Differentiation.** *Bio-protocol*
Chen, M. Y., Heinrich, L., Zafar, F., Sedov, K., Schüle, B.
2023; 13 (11): e4689
- **Nuclease-Dead Cas9-Mediated Downregulation of alpha-Synuclein as a Disease-Modulatory Therapeutic Strategy for Parkinson's Disease**
Schuele, B., Torres, C., Zafar, F., Hermesky, D., Vazquez, J., Piper, D., Sastre, D., Qi, L., Sanders, L., Kirik, D.
CELL PRESS.2023: 339
- **Embryoid Body Cells from Human Embryonic Stem Cells Overexpressing Dopaminergic Transcription Factors Survive and Initiate Neurogenesis via Neural Rosettes in the Substantia Nigra.** *Brain sciences*
Ramos-Acevedo, R., Morato-Torres, C. A., Padilla-Godinez, F. J., Bernal-Conde, L. D., Palomero-Rivero, M., Zafar, F., Collazo-Navarrete, O., Soto-Rojas, L. O., Schule, B., Guerra-Crespo, M.
2023; 13 (2)
- **Nuclease-dead S. aureus Cas9 downregulates alpha-synuclein and reduces mtDNA damage and oxidative stress levels in patient-derived stem cell model of Parkinson's disease.** *bioRxiv : the preprint server for biology*
Sastre, D., Zafar, F., Torres, C. A., Piper, D., Kirik, D., Sanders, L. H., Qi, S., Schule, B.
2023
- **A reference human induced pluripotent stem cell line for large-scale collaborative studies.** *Cell stem cell*
Pantazis, C. B., Yang, A., Lara, E., McDonough, J. A., Blauwendraat, C., Peng, L., Oguro, H., Kanaujiya, J., Zou, J., Sebesta, D., Pratt, G., Cross, E., Blockwick, et al
2022; 29 (12): 1685-1702.e22
- **Early embryonic lethality in complex I associated p.L104P Nubpl mutant mice.** *Orphanet journal of rare diseases*
Cheng, C., Cleak, J., Weiss, L., Cater, H., Stewart, M., Wells, S., Columbres, R. C., Shmara, A., Morato Torres, C. A., Zafar, F., Schule, B., Neumann, J., Hatchwell, et al
2022; 17 (1): 386
- **ATTCT and ATTCC repeat expansions in the ATXN10 gene affect disease penetrance of spinocerebellar ataxia type 10.** *HGG advances*

- Morato Torres, C. A., Zafar, F., Tsai, Y., Vazquez, J. P., Gallagher, M. D., McLaughlin, I., Hong, K., Lai, J., Lee, J., Chirino-Perez, A., Romero-Molina, A. O., Torres, F., Fernandez-Ruiz, et al
2022; 3 (4): 100137
- **Gene Expression Analysis in Stem Cell-derived Cortical Neuronal Cultures Using Multi-well SYBR Green Quantitative PCR Arrays *BIO-PROTOCOL***
Srinivasaraghavan, V., Zafar, F., Schule, B.
2022; 12 (14)
 - **Multiplex imaging of human induced pluripotent stem cell-derived neurons with CO-Detection by indEXing (CODEX) technology. *Journal of neuroscience methods***
Heinrich, L., Zafar, F., Morato Torres, C. A., Singh, J., Khan, A., Chen, M. Y., Hempel, C., Nikulina, N., Mulholland, J., Braubach, O., Schule, B.
2022: 109653
 - **Effect of LRRK2 protein and activity on stimulated cytokines in human monocytes and macrophages. *NPJ Parkinson's disease***
Ahmadi Rastegar, D., Hughes, L. P., Perera, G., Keshiya, S., Zhong, S., Gao, J., Halliday, G. M., Schule, B., Dzamko, N.
2022; 8 (1): 34
 - **Isogenic human SNCA gene dosage induced pluripotent stem cells to model Parkinson's disease. *Stem cell research***
Zafar, F., Nallur Srinivasaraghavan, V., Yang Chen, M., Alejandra Morato Torres, C., Schule, B.
2022; 60: 102733
 - **Phenotypic Heterogeneity among GBA p.R202X Carriers in Lewy Body Spectrum Disorders. *Biomedicines***
Napolioni, V., Fredericks, C. A., Kim, Y., Channappa, D., Khan, R. R., Kim, L. H., Zafar, F., Couthouis, J., Davidzon, G. A., Mormino, E. C., Gitler, A. D., Montine, T. J., Schule, et al
1800; 10 (1)
 - **The commercial genetic testing landscape for Parkinson's disease. *Parkinsonism & related disorders***
Cook, L., Schulze, J., Verbrugge, J., Beck, J. C., Marder, K. S., Saunders-Pullman, R., Klein, C., Naito, A., Alcalay, R. N., ClinGen Parkinson's Disease Gene Curation Expert Panel and the MDS Task Force for Recommendations for Genetic Testing in Parkinson's Disease Clinical Genome Resource (ClinGen) Parkinson's Disease Gene Curation Expert Panel Authors, Movement Society Disorder (MDS) Task Force on Recommendations for Clinical Genetic Testing in Parkinson's Disease Authors, Brice, A., Kumeh, A., West, et al
2021
 - **Embryoid Body Formation from Mouse and Human Pluripotent Stem Cells for Transplantation to Study Brain Microenvironment and Cellular Differentiation. *Methods in molecular biology (Clifton, N.J.)***
Guerra-Crespo, M., Collazo-Navarrete, O., Ramos-Acevedo, R., Morato-Torres, C. A., Schule, B.
2021
 - **Short-term deceleration capacity of heart rate: a sensitive marker of cardiac autonomic dysfunction in idiopathic Parkinson's disease. *Clinical autonomic research : official journal of the Clinical Autonomic Research Society***
Carricarte Naranjo, C., Marras, C., Visanji, N. P., Cornforth, D. J., Sanchez-Rodriguez, L., Schule, B., Goldman, S. M., Estevez, M., Stein, P. K., Lang, A. E., Jelinek, H. F., Machado, A.
2021
 - **Genome-wide association studies of LRRK2 modifiers of Parkinson's disease. *Annals of neurology***
Lai, D., Alipanahi, B., Fontanillas, P., Schwantes-An, T., Aasly, J., Alcalay, R. N., Beecham, G. W., Berg, D., Bressman, S., Brice, A., Brockman, K., Clark, L., Cookson, et al
2021
 - **Genetic and Environmental Factors Influence the Pleomorphy of LRRK2 Parkinsonism. *International journal of molecular sciences***
Chittoor-Vinod, V. G., Nichols, R. J., Schule, B.
2021; 22 (3)
 - **The Role of Alpha-Synuclein and Other Parkinson's Genes in Neurodevelopmental and Neurodegenerative Disorders. *International journal of molecular sciences***
Morato Torres, C. A., Wassouf, Z., Zafar, F., Sastre, D., Outeiro, T. F., Schule, B.
2020; 21 (16)
 - **Establishing polygenic risk score reporting standards and a polygenic score catalog to improve validation, interpretation and reproducibility**

Kraft, P., Wand, H., Lambert, S. A., Tamburro, C., MacArthur, J., Iacocca, M. A., Sillari, C., O'Sullivan, J., Brockman, D., Schuele, B., Venner, E., Kullo, I. J., Rowley, et al
WILEY.2020: 496–97

- **Report of the Phenotype of a Patient with Roberts Syndrome and a Rare ESCO2 Variant.** *Journal of pediatric genetics*
da Costa Almeida, C. B., Welter, A. T., Abech, G. D., Brandão, G. R., Flores, J. A., Schüle, B. n., Francke, U. n., Fiegenbaum, M. n., Zen, P. R., Rosa, R. F.
2020; 9 (1): 58–62
- **Pulse-Field capillary electrophoresis of repeat-primed PCR amplicons for analysis of large repeats in Spinocerebellar Ataxia Type 10.** *PLoS one*
Hashem, V., Tiwari, A., Bewick, B., Teive, H. A., Moscovich, M., Schuele, B., Bushara, K., Bower, M., Rasmussen, A., Tsai, Y., Clark, T., McFarland, K., Ashizawa, et al
2020; 15 (3): e0228789
- **Loss-of-Function NUBPL Mutation May Link Parkinson's Disease to Recessive Complex I Deficiency.** *Frontiers in neurology*
Eis, P. S., Huang, N., Langston, J. W., Hatchwell, E., Schule, B.
2020; 11: 555961
- **Nonsteroidal Anti-Inflammatory Use and LRRK2 Parkinson's Disease Penetrance.** *Movement disorders : official journal of the Movement Disorder Society*
San Luciano, M. n., Tanner, C. M., Meng, C. n., Marras, C. n., Goldman, S. M., Lang, A. E., Tolosa, E. n., Schüle, B. n., Langston, J. W., Brice, A. n., Corvol, J. C., Goldwurm, S. n., Klein, et al
2020
- **Heart rate variability biomarkers of leucine-rich repeat kinase 2-associated Parkinson's disease**
Naranjo, C., Marras, C., Visanji, N. P., Cornforth, D. J., Sanchez-Rodriguez, L., Schuele, B., Goldman, S. M., Estevez, M., Stein, P. K., Lang, A. E., Machado, A., Jelinek, H. F., IEEE
IEEE.2020
- **Exosome/Microvesicle Content is Altered in LRRK2 Mutant iPSC-derived Neural Cells.** *The Journal of comparative neurology*
Candelario, K. M., Balaj, L., Zheng, T., Skog, J., Scheffler, B., Breakefield, X., Schule, B., Steindler, D. A.
2019
- **Large expert-curated database for benchmarking document similarity detection in biomedical literature search DATABASE-THE JOURNAL OF BIOLOGICAL DATABASES AND CURATION**
Brown, P., Tan, A., El-Esawi, M. A., Liehr, T., Blanck, O., Gladue, D. P., Almeida, G. M. F., Cernava, T., Sorzano, C. O., Yeung, A. W. K., Engel, M. S., Chandrasekaran, A., Muth, et al
2019
- **Increased markers of cardiac vagal activity in leucine-rich repeat kinase 2-associated Parkinson's disease.** *Clinical autonomic research : official journal of the Clinical Autonomic Research Society*
Carricarte Naranjo, C., Marras, C., Visanji, N. P., Cornforth, D. J., Sanchez-Rodriguez, L., Schule, B., Goldman, S. M., Estevez, M., Stein, P. K., Lang, A. E., Jelinek, H. F., Machado, A.
2019
- **LRRK2 modifies alpha-syn pathology and spread in mouse models and human neurons** *ACTA NEUROPATHOLOGICA*
Bieri, G., Brahic, M., Bousset, L., Couthouis, J., Kramer, N. J., Ma, R., Nakayama, L., Monbureau, M., Defensor, E., Schuele, B., Shamloo, M., Melki, R., Gitler, et al
2019; 137 (6): 961–80
- **Alpha-Synuclein Physiology and Pathology: A Perspective on Cellular Structures and Organelles.** *Frontiers in neuroscience*
Bernal-Conde, L. D., Ramos-Acevedo, R., Reyes-Hernandez, M. A., Balbuena-Olvera, A. J., Morales-Moreno, I. D., Arguero-Sanchez, R., Schule, B., Guerra-Crespo, M.
2019; 13: 1399
- **LRRK2-mediated Rab10 phosphorylation in immune cells from Parkinson's disease patients.** *Movement disorders : official journal of the Movement Disorder Society*
Atashrazm, F., Hammond, D., Perera, G., Bolliger, M. F., Matar, E., Halliday, G. M., Schule, B., Lewis, S. J., Nichols, R. J., Dzamko, N.
2018
- **A pathway for Parkinson's Disease LRRK2 kinase to block primary cilia and Sonic hedgehog signaling in the brain.** *eLife*

Dhekne, H. S., Yanatori, I., Gomez, R. C., Tonelli, F., Diez, F., Schule, B., Steger, M., Alessi, D. R., Pfeffer, S. R.

2018; 7

- **Genetic fine-mapping of the lowan SNCA gene triplication in a patient with Parkinson's disease** *NPJ PARKINSONS DISEASE*
Zafar, F., Valappil, R., Kim, S., Johansen, K. K., Chang, A. S., Tetrud, J. W., Eis, P. S., Hatchwell, E., Langston, J., Dickson, D. W., Schule, B.
2018; 4: 18
- **Clustering of Motor and Nonmotor Traits in Leucine-Rich Repeat Kinase 2 G2019S Parkinson's Disease Nonparkinsonian Relatives: A Multicenter Family Study** *MOVEMENT DISORDERS*
Mestre, T. A., Pont-Sunyer, C., Kausar, F., Visanji, N. P., Ghate, T., Connolly, B. S., Gasca-Salas, C., Kern, D. S., Jain, J., Slow, E. J., Faust-Socher, A., Kasten, M., Wadia, et al
2018; 33 (6): 960–65
- **Advancing Stem Cell Models of Alpha-Synuclein Gene Regulation in Neurodegenerative Disease** *FRONTIERS IN NEUROSCIENCE*
Piper, D. A., Sastre, D., Schuele, B.
2018; 12: 199
- **LRRK2 p. Ile1371Val Mutation in a Case with Neuropathologically Confirmed Multi-System Atrophy** *JOURNAL OF PARKINSONS DISEASE*
Lee, K., Khanh-Dung Nguyen, Sun, C., Liu, M., Zafar, F., Saetern, J., Flierl, A., Tetrud, J. W., Langston, J., Dickson, D., Schule, B.
2018; 8 (1): 93–100
- **LRRK2 G2019S-induced mitochondrial DNA damage is LRRK2 kinase dependent and inhibition restores mtDNA integrity in Parkinson's disease** *HUMAN MOLECULAR GENETICS*
Howlett, E. H., Jensen, N., Belmonte, F., Zafar, F., Hu, X., Kluss, J., Schule, B., Kaufman, B. A., Greenamyre, J. T., Sanders, L. H.
2017; 26 (22): 4340–51
- **Penetrance Estimate of LRRK2 p.G2019S Mutation in Individuals of Non-Ashkenazi Jewish Ancestry** *MOVEMENT DISORDERS*
Lee, A. J., Wang, Y., Alcalay, R. N., Mejia-Santana, H., Saunders-Pullman, R., Bressman, S., Corvol, J., Brice, A., Lesage, S., Mangone, G., Tolosa, E., Pont-Sunyer, C., Vilas, et al
2017; 32 (10): 1432–38
- **Parkinson's disease associated with pure ATXN10 repeat expansion** *NPJ PARKINSONS DISEASE*
Schule, B., McFarland, K. N., Lee, K., Tsai, Y., Khanh-Dung Nguyen, Sun, C., Liu, M., Byrne, C., Gopi, R., Huang, N., Langston, J., Clark, T., Jimenez Gil, F., et al
2017; 3: 27
- **beta(2)-Adrenoreceptor is a regulator of the alpha-synuclein gene driving risk of Parkinson's disease** *SCIENCE*
Mittal, S., Bjornevik, K., Im, D., Flierl, A., Dong, X., Locascio, J. J., Abo, K. M., Long, E., Jin, M., Xu, B., Xiang, Y. K., Rochet, J., Engeland, et al
2017; 357 (6354): 891–97
- **Heart Rate Variability in leucine-rich repeat kinase 2-Associated Parkinson's Disease** *MOVEMENT DISORDERS*
Visanji, N. P., Bhudhikanok, G. S., Mestre, T. A., Ghate, T., Udupa, K., AlDakheel, A., Connolly, B. S., Gasca-Salas, C., Kern, D. S., Jain, J., Slow, E. J., Faust-Socher, A., Kim, et al
2017; 32 (4): 610–14
- **Inflammatory profile discriminates clinical subtypes in LRRK2-associated Parkinson's disease** *EUROPEAN JOURNAL OF NEUROLOGY*
Brockmann, K., Schulte, C., Schneiderhan-Marra, N., Apel, A., Pont-Sunyer, C., Vilas, D., Ruiz-Martinez, J., Langkamp, M., Corvol, J., Cormier, F., Knorpp, T., Joos, T. O., Bernard, et al
2017; 24 (2): 427–E6
- **Derivation of Leptomeninges Explant Cultures from Postmortem Human Brain Donors** *JOVE-JOURNAL OF VISUALIZED EXPERIMENTS*
Lee, K., Saetern, O., Nguyen, A., Rodriguez, L., Schule, B.
2017
- **Functional Impairment in Miro Degradation and Mitophagy Is a Shared Feature in Familial and Sporadic Parkinson's Disease** *CELL STEM CELL*
Hsieh, C., Shaltouki, A., Gonzalez, A. E., Da Cruz, A. B., Burbulla, L. F., St Lawrence, E., Schule, B., Krainc, D., Palmer, T. D., Wang, X.
2016; 19 (6): 709-724
- **Inflammatory profile in LRRK2-associated prodromal and clinical PD** *JOURNAL OF NEUROINFLAMMATION*

- Brockmann, K., Apel, A., Schulte, C., Schneiderhan-Marra, N., Pont-Sunyer, C., Vilas, D., Ruiz-Martinez, J., Langkamp, M., Corvol, J., Cormier, F., Knorrp, T., Joos, T. O., Gasser, et al
2016; 13: 122
- **IS PARKIN PARKINSONISM A CANCER PREDISPOSITION SYNDROME? *NEUROLOGY-GENETICS***
Schule, B., Byrne, C., Rees, L., Langston, J.
2015; 1 (4): e31
 - **Multisystem Lewy body disease and the other parkinsonian disorders *NATURE GENETICS***
Langston, J., Schuele, B., Rees, L., Nichols, R., Barlow, C.
2015; 47 (12): 1378–84
 - **Elevated alpha-synuclein caused by SNCA gene triplication impairs neuronal differentiation and maturation in Parkinson's patient-derived induced pluripotent stem cells *CELL DEATH & DISEASE***
Oliveira, L. A., Falomir-Lockhart, L. J., Botelho, M. G., Lin, K., Wales, P., Koch, J. C., Gerhardt, E., Taschenberger, H., Outeiro, T. F., Lingor, P., Schuele, B., Arndt-Jovin, D. J., Jovin, et al
2015; 6: e1994
 - **Comparative Genomic Hybridization Solves a 14-Year-Old PARKIN Mystery *ANNALS OF NEUROLOGY***
Schuele, B., Hatchwell, E., Eis, P. S., Langston, J.
2015; 78 (4): 663–64
 - **Clinical Correlations With Lewy Body Pathology in LRRK2-Related Parkinson Disease *JAMA NEUROLOGY***
Kalia, L. V., Lang, A. E., Hazrati, L., Fujioka, S., Wszolek, Z. K., Dickson, D. W., Ross, O. A., Van Deerlin, V. M., Trojanowski, J. Q., Hurtig, H. I., Alcalay, R. N., Marder, K. S., Clark, et al
2015; 72 (1): 100–105
 - **Higher Vulnerability and Stress Sensitivity of Neuronal Precursor Cells Carrying an Alpha-Synuclein Gene Triplication *PLOS ONE***
Flierl, A., Oliveira, L. M. A., Falomir-Lockhart, L. J., Mak, S. K., Hesley, J., Soldner, F., Arndt-Jovin, D. J., Jaenisch, R., Langston, J., Jovin, T. M., Schuele, B.
2014; 9 (11): e112413
 - **Systems-Based Analyses of Brain Regions Functionally Impacted in Parkinson's Disease Reveals Underlying Causal Mechanisms *PLOS ONE***
Riley, B. E., Gardai, S. J., Emig-Agius, D., Bessarabova, M., Ivliev, A. E., Schuele, B., Alexander, J., Wallace, W., Halliday, G. M., Langston, J., Braxton, S., Yednock, T., Shaler, et al
2014; 9 (8): e102909
 - **Michael J. Fox Foundation LRRK2 Consortium: geographical differences in returning genetic research data to study participants *GENETICS IN MEDICINE***
Alcalay, R. N., Aasly, J., Berg, D., Bressman, S., Brice, A., Brockmann, K., Chan, P., Clark, L., Cormier, F., Corvol, J., Durr, A., Facheris, M., Farrer, et al
2014; 16 (8): 644–45
 - **DICE, an efficient system for iterative genomic editing in human pluripotent stem cells. *Nucleic acids research***
Zhu, F., Gamboa, M., Farruggio, A. P., Hippenmeyer, S., Tasic, B., Schüle, B., Chen-Tsai, Y., Calos, M. P.
2014; 42 (5)
 - **LRRK2 mutations cause mitochondrial DNA damage in iPSC-derived neural cells from Parkinson's disease patients: Reversal by gene correction *NEUROBIOLOGY OF DISEASE***
Sanders, L. H., Laganieri, J., Cooper, O., Mak, S. K., Vu, B., Huang, Y., Paschon, D. E., Vangipuram, M., Sundararajan, R., Urov, F. D., Langston, J., Gregory, P. D., Zhang, et al
2014; 62: 381–86
 - **Discovery of functional non-coding conserved regions in the alpha-synuclein gene locus. *F1000Research***
Sterling, L., Walter, M., Ting, D., Schule, B.
2014; 3: 259
 - **Identification and Rescue of alpha-Synuclein Toxicity in Parkinson Patient-Derived Neurons *SCIENCE***
Chung, C., Khurana, V., Auluck, P. K., Tardiff, D. F., Mazzulli, J. R., Soldner, F., Bar, V., Lou, Y., Frey, Y., Cho, S., Mungenast, A. E., Muffat, J., Mitalipova, et al

2013; 342 (6161): 983–87

- **Elevated Alpha-Synuclein Impairs Innate Immune Cell Function and Provides a Potential Peripheral Biomarker for Parkinson's Disease** *PLOS ONE*
Gardai, S. J., Mao, W., Schuele, B., Babcock, M., Schoebel, S., Lorenzana, C., Alexander, J., Kim, S., Glick, H., Hilton, K., Fitzgerald, J., Buttini, M., Chiou, et al
2013; 8 (8): e71634
- **Skin Punch Biopsy Explant Culture for Derivation of Primary Human Fibroblasts** *JOVE-JOURNAL OF VISUALIZED EXPERIMENTS*
Vangipuram, M., Ting, D., Kim, S., Diaz, R., Schuele, B.
2013: e3779
- **Clinical correlations with lewy body pathology in LRRK2-related Parkinson's disease**
Kalia, L., Lang, A., Hazrati, L., Fujioka, S., Wszolek, Z., Dickson, D., Ross, O., Van Deerlin, V., Trojanowski, J., Hurtig, H., Alcalay, R., Gaig, C., Tolosa, et al
WILEY-BLACKWELL.2013: S405
- **Small molecules greatly improve conversion of human-induced pluripotent stem cells to the neuronal lineage.** *Stem cells international*
Mak, S. K., Huang, Y. A., Iranmanesh, S., Vangipuram, M., Sundararajan, R., Nguyen, L., Langston, J. W., Schüle, B.
2012; 2012: 140427-?
- **SNCA Triplication Parkinson's Patient's iPSC-derived DA Neurons Accumulate alpha-Synuclein and Are Susceptible to Oxidative Stress** *PLOS ONE*
Byers, B., Cord, B., Ha Nam Nguyen, H. N., Schuele, B., Fenno, L., Lee, P. C., Deisseroth, K., Langston, J. W., Pera, R. R., Palmer, T. D.
2011; 6 (11)
- **Call for participation in the neurogenetics consortium within the Human Variome Project** *NEUROGENETICS*
Haworth, A., Bertram, L., Carrera, P., Elson, J. L., Braastad, C. D., Cox, D. W., Cruts, M., den Dunnen, J. T., Farrer, M. J., Fink, J. K., Hamed, S. A., Houlden, H., Johnson, et al
2011; 12 (3): 169–73
- **Phenotype in parkinsonian and nonparkinsonian LRRK2 G2019S mutation carriers** *NEUROLOGY*
Marras, C., Schuele, B., Munhoz, R. P., Rogaeva, E., Langston, J. W., Kasten, M., Meaney, C., Klein, C., Wadia, P. M., Lim, S., Chuang, R. I., Zadikof, C., Steeves, et al
2011; 77 (4): 325-333
- **LRRK2 Mutant iPSC-Derived DA Neurons Demonstrate Increased Susceptibility to Oxidative Stress** *CELL STEM CELL*
Ha Nam Nguyen, N. N., Byers, B., Cord, B., Shcheglovitov, A., Byrne, J., Gujar, P., Kee, K., Schuele, B., Dolmetsch, R. E., Langston, W., Palmer, T. D., Pera, R. R.
2011; 8 (3): 267-280
- **Mitochondrial Dysfunction in Skin Fibroblasts from a Parkinson's Disease Patient with an alpha-Synuclein Triplication** *JOURNAL OF PARKINSONS DISEASE*
Mak, S. K., Tewari, D., Tetrud, J. W., Langston, J., Schuele, B.
2011; 1 (2): 175–83
- **Alpha-synuclein-glucocerebrosidase interactions in pharmacological Gaucher models: A biological link between Gaucher disease and parkinsonism** *NEUROTOXICOLOGY*
Manning-Bog, A. B., Schuele, B., Langston, J.
2009; 30 (6): 1127–32
- **Can cellular models revolutionize drug discovery in Parkinson's disease?** *BIOCHIMICA ET BIOPHYSICA ACTA-MOLECULAR BASIS OF DISEASE*
Schuele, B., Pera, R., Langston, J.
2009; 1792 (11): 1043–51
- **Severe congenital encephalopathy caused by MECP2 null mutations in males: central hypoxia and reduced neuronal dendritic structure** *CLINICAL GENETICS*
Schule, B., Armstrong, D. D., Vogel, H., Oviedo, A., Francke, U.
2008; 74 (2): 116-126

- **Pure akinesia as initial presentation of PSP: A clinicopathological study** *PARKINSONISM & RELATED DISORDERS*
Facheris, M., Maniak, S., Scaravilli, F., Schuele, B., Klein, C., Pramstaller, P.
2008; 14 (6): 517–19
- **Phenotypic spectrum and sex effects in eleven myoclonus-dystonia families with epsilon-sarcoglycan mutations** *MOVEMENT DISORDERS*
Raymond, D., Saunders-Pullman, R., Aguiar, P., Schule, B., Kock, N., Friedman, J., Harris, J., Ford, B., Frucht, S., Heiman, G. A., Jennings, D., Doherty, D., Brin, et al
2008; 23 (4): 588–92
- **DLX5 and DLX6 expression is biallelic and not modulated by MeCP2 deficiency** *AMERICAN JOURNAL OF HUMAN GENETICS*
Schuele, B., Li, H. H., Fisch-Kohl, C., Purmann, C., Francke, U.
2007; 81 (3): 492-506
- **Phenotypic variation in a large Swedish pedigree due to SNCA duplication and triplication** *NEUROLOGY*
Fuchs, J., Nilsson, C., Kachergus, J., Munz, M., Larsson, E., Schuele, B., Langston, J. W., Middleton, F. A., Ross, O. A., Hulihan, M., Gasser, T., Farrer, M. J.
2007; 68 (12): 916–22
- **Parkin gene variations and parkinsonism: Association does not imply causation** *ANNALS OF NEUROLOGY*
Langston, J., Tanner, C. M., Schule, B.
2007; 61 (1): 4–6
- **Novel features in a patient homozygous for the L347P mutation in the PINK1 gene** *PARKINSONISM & RELATED DISORDERS*
Doostzadeh, J., Tetrud, J. W., Allen-Auerbach, M., Langston, J. W., Schuele, B.
2007; 13 (6): 359–61
- **Intrafamilial phenotypic and genetic heterogeneity of dystonia** *JOURNAL OF THE NEUROLOGICAL SCIENCES*
Kostic, V. S., Svetel, M., Kabakci, K., Ristic, A., Petrovic, I., Schuele, B., Kock, N., Djarmati, A., Romac, S., Klein, C.
2006; 250 (1-2): 92–96
- **Inactivating mutations in ESCO2 cause SC phocomelia and Roberts syndrome: No phenotype-genotype correlation** *AMERICAN JOURNAL OF HUMAN GENETICS*
Schule, B., Oviedo, A., Johnston, K., Pai, S., Francke, U.
2005; 77 (6): 1117-1128
- **PINK1, Parkin, and DJ-1 mutations in Italian patients with early-onset parkinsonism** *EUROPEAN JOURNAL OF HUMAN GENETICS*
Klein, C., Djarmati, A., Hedrich, K., Schafer, N., Scaglione, C., Marchese, R., Kock, N., Schule, B., Hiller, A., Lohnau, T., Winkler, S., Wiegers, K., Hering, et al
2005; 13 (9): 1086–93
- **Molecular breakpoint cloning and gene expression studies of a novel translocation t(4;15)(q27;q11.2) associated with Prader-Willi syndrome** *BMC MEDICAL GENETICS*
Schule, B., Albalwi, M., Northrop, E., Francis, D. I., Rowell, M., Slater, H. R., Gardner, R. J., Francke, U.
2005; 6
- **Genetic heterogeneity in ten families with myoclonus-dystonia** *JOURNAL OF NEUROLOGY NEUROSURGERY AND PSYCHIATRY*
Schule, B., Kock, N., Svetel, M., Dragasevic, N., Hedrich, K., Aguiar, P. D., Liu, L., Kabakci, K., Garrels, J., Meyer, E. M., Berisavac, I., Schwinger, E., Kramer, et al
2004; 75 (8): 1181-1185
- **Parkin gene alterations in hepatocellular carcinoma** *GENES CHROMOSOMES & CANCER*
Wang, F., Denison, S., Lai, J. P., Philips, L. A., Montoya, D., Kock, N., Schule, B., Klein, C., Shridhar, Roberts, L. R., Smith, D. I.
2004; 40 (2): 85–96
- **Myoclonus-dystonia: Detection of novel, recurrent, and de novo SGCE mutations** *NEUROLOGY*
Hedrich, K., Meyer, E. M., Schule, B., Kock, N., Aguiar, P. D., Wiegers, K., Koelman, J. H., Garrels, J., Durr, R., Liu, L., Schwinger, E., Ozelius, L. J., Landwehrmeyer, et al
2004; 62 (7): 1229–31
- **Clinical and genetic features of myoclonus-dystonia in 3 cases: A video presentation** *MOVEMENT DISORDERS*

- Kock, N., Kasten, M., Schule, B., Hedrich, K., Wiegers, K., Kabakci, K., Hagenah, J., Pramstaller, P. P., Nitschke, M. F., Munchau, A., Sperner, J., Klein, C.
2004; 19 (2): 231–34
- **Alterations in the common fragile site gene Parkin in ovarian and other cancers** *ONCOGENE*
Denison, Wang, F., Becker, N. A., Schule, B., Kock, N., Phillips, L. A., Klein, C., Smith, D. I.
2003; 22 (51): 8370–78
 - **Hereditary myoclonus-dystonia associated with epilepsy** *NEUROLOGY*
Foncke, E. M., Klein, C., Koelman, J. H., Kramer, P. L., Schilling, K., Muller, B., Garrels, J., Aguiar, P. D., Liu, L., de Froe, A., Speelman, J. D., Ozelius, L. J., Tijssen, et al
2003; 60 (12): 1988–90
 - **Evidence that paternal expression of the epsilon-Sarcoglycan gene accounts for reduced penetrance in myoclonus-dystonia** *AMERICAN JOURNAL OF HUMAN GENETICS*
Muller, B., Hedrich, K., Kock, N., Dragasevic, N., Svetel, M., Garrels, J., Landt, O., Nitschke, M., Pramstaller, P. P., Reik, W., Schwinger, E., Sperner, J., Ozelius, et al
2002; 71 (6): 1303–11
 - **epsilon-sarcoglycan mutations found in combination with other dystonia gene mutations** *ANNALS OF NEUROLOGY*
Klein, C., Liu, L., Doheny, D., Kock, N., Muller, B., Aguiar, P. D., Leung, J., de Leon, D., Bressman, S. B., Silverman, J., Smith, C., Danisi, F., Morrison, et al
2002; 52 (5): 675–79
 - **Phenotypic features of myoclonus-dystonia in three kindreds** *NEUROLOGY*
Doheny, D. O., Morrison, C. E., Smith, C. J., Walker, R. H., Abbasi, S., Muller, B., Garrels, J., Liu, L., Aguiar, P. D., Schilling, K., Kramer, P., de Leon, D., Raymond, et al
2002; 59 (8): 1187–96
 - **Dopamine transmission in DYT1 dystonia: A biochemical and autoradiographical study** *NEUROLOGY*
Augood, S. J., Hollingsworth, Z., Albers, D. S., Yang, L., LEUNG, J. C., Muller, B., Klein, C., Breakefield, X. O., Standaert, D. G.
2002; 59 (3): 445–448
 - **Role of SC42 mutations in early- and late-onset dopa-responsive parkinsonism** *ANNALS OF NEUROLOGY*
Kock, N., Muller, B., Vieregge, P., Pramstaller, P. P., Marder, K., Abbruzzese, G., Martinelli, P., Lang, A. E., Jacobs, H., Hagenah, J., Harris, J., Meija-Santana, H., Fahn, et al
2002; 52 (2): 257–58
 - **The parkin gene is not involved in late-onset Parkinson's disease** *NEUROLOGY*
Kann, M., Hedrich, K., Vieregge, P., Jacobs, H., Muller, B., Kock, N., Schwinger, E., Klein, C., Marder, K., Harris, J., Meija-Santana, H., Bressman, S., Ozelius, et al
2002; 58 (5): 835
 - **Slice culture of the olfactory bulb of *Xenopus laevis* tadpoles** *CHEMICAL SENSES*
Scheidweiler, U., Nezlín, L., Rabba, J., Muller, B., Schild, D.
2001; 26 (4): 399–407
 - **Noradrenergic modulation of calcium currents and synaptic transmission in the olfactory bulb of *Xenopus laevis* tadpoles** *EUROPEAN JOURNAL OF NEUROSCIENCE*
Czesnik, D., Nezlín, L., Rabba, J., Muller, B., Schild, D.
2001; 13 (6): 1093–1100