

Stanford



Birgitt Schuele

Associate Professor (Research) of Pathology

Bio

BIO

Birgitt Schüle, MD, is an Associate Professor in the Department of Pathology, Stanford University School of Medicine. Her research focuses on medical genetics and stem cell modeling to unlock disease mechanisms and pathways leading to neurodegeneration in Parkinson's disease and related disorders, and to develop new therapeutic strategies to advance precision medicine.

She received her medical training from the Georg-August University Göttingen and Medical University Lübeck, Germany (1993 - 2001) and completed doctoral degree in medicine (Dr. med.) in neurophysiology at the Georg-August University Göttingen (2001). During her neurology internship from 2001 to 2002 at Medical University of Lübeck with Prof. Christine Klein, Dr. Schüle studied genes for inherited forms of Parkinson's disease and dystonia. From 2003 to 2005, she completed a postdoctoral fellowship in human genetics with Prof. Uta Francke at Stanford University School of Medicine. From 2005-2019, Dr. Schüle led key clinical research programs and biospecimen repositories for neurogenetics, translational stem cell and brain donation at the Parkinson's Institute and Clinical Center.

Dr. Schuele is the associate core leader of the Neuropathology Core with the Stanford Alzheimer Research Center (ADRC) and core leader of the Analytics Core for the Pacific Udall Center. She supports the centers with genetic characterization, biobanking, and building a human induced pluripotent stem cell and post-mortem leptomeninges tissue bank shared with the data and tissue repositories at NIH.

ACADEMIC APPOINTMENTS

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

ADMINISTRATIVE APPOINTMENTS

- 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- present)

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, Guadalajara & Mexico City, Mexico

LINKS

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

Teaching

STANFORD ADVISEES

Postdoctoral Faculty Sponsor

Laurin Heinrich

Publications

PUBLICATIONS

- **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.
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- **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., Schuele, 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.
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- **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., Schuele, 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., Schuele, 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. F., Cernava, T., Sorzano, C. O., Yeung, A. 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., Schuele, 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., Schuele, 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.
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- **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 Iowan 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
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- **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., Knorrp, T., Joos, T. O., Bernard, et al
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- **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 Mito 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.
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- **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
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- **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
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- **Higher Vulnerability and Stress Sensitivity of Neuronal Precursor Cells Carrying an Alpha-Synuclein Gene Triplication** *PLOS ONE*
Flierl, A., Oliveira, L. 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.
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- **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
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- **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.
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- **LRRK2 mutations cause mitochondrial DNA damage in iPSC-derived neural cells from Parkinson's disease patients: Reversal by gene correction** *NEUROBIOLOGY OF DISEASE*
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- **Discovery of functional non-coding conserved regions in the alpha-synuclein gene locus.** *F1000Research*
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- **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., Baru, V., Lou, Y., Freyzon, Y., Cho, S., Mungenast, A. E., Muffat, J., Mitalipova, et al
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- **Elevated Alpha-Synuclein Impairs Innate Immune Cell Function and Provides a Potential Peripheral Biomarker for Parkinson's Disease** *PLOS ONE*
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- **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.
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- **Clinical correlations with lewy body pathology in LRRK2-related Parkinson's disease**
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- **SNCA Triplication Parkinson's Patient's iPSC-derived DA Neurons Accumulate alpha-Synuclein and Are Susceptible to Oxidative Stress** *PLOS ONE*
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2011; 6 (11)
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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.
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- **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., Tetrad, J. W., Langston, J., Schuele, B.
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- **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.
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- **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*
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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*
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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.
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- **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.
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