

# Modeling Parkinson's Disease in a Petri Dish



## Birgitt Schuele

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Associate Professor  
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## CURRENT RESEARCH

### Discovering novel genetic variants in Parkinson's disease for the development of new therapies

The clinical symptoms of Parkinson's disease are caused by neuronal cell loss in small areas of the brainstem, which leads to symptoms such as uncontrollable tremor, stiffness, slowness of movements and gait problems among others. This common progressive neurodegenerative disorder has currently no curative treatment and its cause is still elusive.

Dr. Birgitt Schuele, Director of Gene Discovery and Stem Cell Modeling at the Parkinson's Institute and Clinical Center developed an innovative model that turns skin cells into stem cells and further coaxes them to become dopaminergic neurons similar to the ones that die in the brains of Parkinson's patients thereby creating Parkinson's disease in a culture dish. This enables her to explore the underlying cause of Parkinson's disease. Using this model, she explores newly discovered genes and variants, identifying novel targets with a goal of discovering new curative therapies. This research lays the foundation for personalized and precision medicine, as well as advancements in regenerative medicine for Parkinson's disease.

At the Clinical Center of the Parkinson's Institute, about 10 percent of patients report a positive family history of the disease. Relatives of these patients often want to know, "What's my genetic risk of developing Parkinson's disease?" Such questions led to the development of Dr. Schuele's gene discovery program, in which DNA samples are collected from patients with a positive family history of Parkinson's disease and their family members. Once a genetic cause is discovered, which is in only 20-30 percent of the families with Parkinson's disease, Dr. Schuele's team unravels how these gene mutations lead to pathological...

## AFFILIATION



Parkinson's Institute and Clinical Center

## AWARDS

- Research Fellowship for "Mechanisms of Imprinting in selected genes in Prader-Willi-/Angelman syndrome and myoclonus-dystonia," 2003
- Dean's Postdoctoral Fellowship for "Knock-down strategy of Pwcr1 small nucleolar RNAs with RNAi to generate models for Prader-Willi syndrome," 2004
- Dean's Postdoctoral Fellowship for "Evaluation of the Imprinting status of DLX5 and its alteration by Mecp2 deficiency," 2005
- Research Fellowship, 2008
- Groundbreaking Award, Parkinson's Story, 2013

## RESEARCH AREAS

Life Science, Genomics / Congenital, Neurological / Cognitive, Regenerative Medicine

## FUNDING REQUEST

Your contributions can help in several ways. \$2.5K helps to create a "genetic profile" from a patient. To help Dr. Schuele's team define how defects in genes cause Parkinson's, you can fund the study of patient stem cells in a petri dish. \$10K allow generating and characterizing stem cells from one patient. \$20K helps to genetically study an entire family with familial Parkinson's. Overall cost for Dr. Schuele's program is \$400K/year. Play a role in exploring novel ways to solve Parkinson's!

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