

# Thursday, June 27<sup>th</sup>

**8:00 - 9:00**    **Conference Registration**
**9:00 - 9:15**    **Welcome and opening remarks – Ziv Gan-Or, Guy Rouleau**
**9:15 - 10:30**    **GBA1 and the lysosomal glycosphingolipid metabolism pathway**
**Chair: Ellen Sidransky**
**9:15 – 9:35**    **Gregory Grabowski** – The normal structure and function of Glucocerebrosidase

**9:35 – 9:55**    **Peter Lansbury** – The lysosomal glycosphingolipid metabolism pathway: an overview

**9:55 – 10:15**    **Roy Alcalay** – Glucocerebrosidase relevant biomarkers

**10:15 – 10:30**    Discussion / questions

**10:30 - 11:00**    **Coffee break**
**11:00 - 12:15**    **Mechanisms in GBA1-associated neurodegeneration 1**
**Chair: Edward Fon**
**11:00 – 11:25**    **Joe Mazzuli** – Lysosomal dysfunction and alpha-synuclein accumulation

**11:25 – 11:50**    **Irina Alecu** – GBA1 and the mitochondria

**11:50 – 12:15**    **Malu Tansey** – GBA1 and the immune system

**12:15 - 14:00**    **Lunch + posters 1-13**
**14:00 - 15:00**    **Mechanisms in GBA1-associated neurodegeneration 2**
**Chair: Thomas Durcan**
**14:00 – 14:30**    **Stacy Henry** – Other mechanisms potentially involved in GBA1-associated neurodegeneration

**14:30 – 15:00**    **Ziv Gan-Or, Ellen Sidransky, Joe Mazzuli, Stacy Henry** – Loss- vs. Gain-of-function mechanisms – a panel

**15:00 - 15:30**    **Break**
**15:30 - 17:00**    **Data Blitz 1**
**Chair: Ziv Gan-Or**
**15:30 – 15:42**    **Towfique Raj** – Deciphering the Immunogenomic Landscape of GBA1-Associated Parkinson's Disease

**15:42 – 15:54**    **Peter Bauer** – The role of the 'African GBA1 allele' of rs3115534 in Parkinson's Disease

**15:54 – 16:06**    **Ricardo A. Feldman** – Acid ceramidase plays a key role in the pathogenic cascade leading to neurodegeneration in Gaucher and GBA1-associated Parkinson's disease

**16:06 – 16:18**    **Nathaniel Safren** – A Genome Wide CRISPR Interference Screen Reveals Commander Genes as Modifiers of Glucocerebrosidase Activity and Lysosomal Function

**16:18 – 16:30**    **Jiang-An Yin** – A Genome-Wide Landscape of Genetic Modifiers of GCase Deficiency Revealed by An Arrayed CRISPR Activation Screen

**16:30 – 16:42**    **Oliver B. Davis** – Exacerbated lysosomal and mitochondrial dysfunction is associated with a common Parkinson's Disease risk allele of GBA1

**16:42 – 16:54**    **Casey Young** – Sex-specific disruptions in lipid metabolism in GBA1-Parkinson's Disease

# Friday, June 28<sup>th</sup>

**9:00 - 10:30 Genetic considerations in *GBA1*-associated neurodegeneration**
**Chair: Ignacio Mata**
**9:10 - 9:30 Ari Zimran** – Gaucher disease, an overview

**9:30 - 9:50 Sonja Scholz** – *GBA1* frequencies in PD and dementia across different populations

**9:50 - 10:10 Oluwadamilola Ojo** – The importance of the intronic *GBA1* variant identified in people of African origin

**10:10 - 10:30 Ziv Gan-Or** – Classification and penetrance of *GBA1* variants, and the potential role of modifiers

**10:30 - 11:00 Coffee break**
**11:00 - 12:15 The role of other players in the lysosomal glycosphingolipid metabolism pathway in neurodegeneration**
**Chair: Peter Lansbury**
**11:00 - 11:25 Jia Nee Foo** – Is SMPD1 a PD gene?

**11:25 - 11:50 Konstantin Senkevich** – The role of other genes in the lysosomal glycosphingolipid metabolism pathway in PD

**11:50 - 12:15 Steffany Bennett** – The lysosomal glycosphingolipid metabolic network in PD

**12:15-14:00 Lunch + posters 14-25**
**14:00-15:30 Characteristics of individuals with *GBA1*-associated neurodegeneration**
**Chair: Roy Alcalay**
**14:00 - 14:30 Kathrin BrockMann** – The phenotype of *GBA1*-associated PD

**14:30 - 15:00 Ronald Postuma** – *GBA1* and REM-sleep behavior disorder

**15:00 - 15:30 Lorraine Kalia** – Pathology in *GBA1*-associated PD/dementia

**15:30-16:00 Break**
**16:00-17:00 Collaborative efforts to advance *GBA1* research and treatment**
**Chair: Ziv Gan-Or**
**16:00 - 16:15 Lorna Chebon-Bore** – G-CAN: an overview

**16:15 - 16:30 Kajsa Atterling Brolin & Andrew Singleton** – GP2: an overview

**16:30 - 17:00 Panel Discussion: Accelerating *GBA1* discoveries and treatment through collaboration – Panelists: Shalini Padmanabhan, Angelica Asis, Roy Alcalay, Simon Stott, Kajsa Atterling Brolin, Aviva Rosenberg**



# Saturday, June 29<sup>th</sup>

**9:00 - 10:30 Data Blitz 2**
**Chair: Rachel Saunders-Pullmant**

- 09:00 – 09:12** **Céline Galvagnion** – Lipidomic Profiling Reveals Shared Pathological Signatures in Sporadic Parkinson's Disease and GBA Mutation Carriers: Implications for Disease Mechanisms
- 09:12 – 09:24** **Aarnoud van der Spoel** – Fluorescent activity-based probes and selective inhibitors for *GBA1*
- 09:24 – 09:36** **Ellen Sidransky** – 15-year evaluation for prodromal Parkinson features in an at-risk cohort with Gaucher disease
- 09:36 – 09:48** **Logan M. Glasstetter** – High-throughput screening for small-molecule stabilizers of misfolded glucocerebrosidase in Gaucher disease and Parkinson's disease
- 09:48 – 10:00** **Preethi Sheshadri** – Restoration of lysosomal acidification rescues mitophagy along with lysosomal and mitochondrial pathophysiology in *GBA1*-mediated Parkinson's Disease
- 10:00 – 10:12** **Joanne Taylor** – GT-02287, a clinical stage allosteric GCase modulator for the treatment of *GBA1*-Parkinson's disease
- 10:12 – 10:24** **Kimberly McDowell** – Systemic AAV gene therapy with CNS-targeted engineered capsids achieves significant GCase activity increases in the primate brain to support the potential treatment of PD-*GBA1*

**10:30 - 11:00 Coffee break**
**11:00-12:20 Current status of *GBA1* clinical trials**
**Chair: Anastacia Henry**

- 11:00-11:20** **Simon Stott** – The landscape of the current pre-clinical and clinical trials on *GBA1*
- 11:20-11:40** **Pablo Sardi** – What can we learn from the MOVES-PD trial?
- 11:40-12:00** **Stephen Mullin** – Overview of completed and ongoing Ambroxol trials
- 12:00-12:20** **Sebastian Boland** – Gene therapy for *GBA1*-associated neurodegeneration

**12:20-14:15 Lunch + posters 26-38**
**14:15 – 15:15 Considerations in clinical trial design for *GBA1*-associated neurodegeneration**
**Chair: Lorraine Kalia**

- 14:00-14:20** **Peter Bauer** – Trial ready populations and the future of clinical genetic screening
- 14:20-14:40** **Ziv Gan-Or** – Mutation classification and genetic stratification in *GBA1* trials
- 14:40-15:00** **Miguel Fonseca** – Other consideration for inclusion / exclusion and endpoints in *GBA1* trials

**15:15- 15:45 Moving forward, Awards for best trainee abstracts and concluding remarks – Ziv Gan-Or**