

GBA1 meeting, June 27-29, Montreal, Canada - Final program

Day 1

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 The normal structure and function of Glucocerebrosidase – **Gregory Grabowski**

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

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

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 Lysosomal dysfunction and alpha-synuclein accumulation – **Joe Mazzuli**

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

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

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 Other mechanisms potentially involved in GBA1-associated neurodegeneration – **Stacy Henry**

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

15:00-15:30 Break

15:30-17:00 Data Blitz 1

Chair: **Ziv Gan-Or**

15:30 – 15:42 Deciphering the Immunogenomic Landscape of GBA1-Associated Parkinson's Disease –

Towfique Raj

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

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

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

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

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

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

Day 2

9:00-10:30 – Genetic considerations in GBA1-associated neurodegeneration

Chair: **Ignacio Mata**

9:10 – 9:30 Gaucher disease, an overview – **Ari Zimran**

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

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

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

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 Is SMPD1 a PD gene? – **Jia Nee Foo**

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

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

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 The phenotype of *GBA1*-associated PD – Speaker: **Kathrin BrockMann**

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

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

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 G-CAN: an overview – **Lorna Chebon-Bore**

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

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

Day 3

9:00-10:30 Data Blitz 2

Chair: **Michael Schlossmacher**

09:00 – 09:12 Lipidomic Profiling Reveals Shared Pathological Signatures in Sporadic Parkinson's Disease and *GBA* Mutation Carriers: Implications for Disease Mechanisms – **Céline Galvagnion**

09:12 – 09:24 Fluorescent activity-based probes and selective inhibitors for *GBA1* – **Aarnoud van der Spoel**

09:24 – 09:36 15-year evaluation for prodromal Parkinson features in an at-risk cohort with Gaucher disease – **Ellen Sidransky**

09:36 – 09:48 High-throughput screening for small-molecule stabilizers of misfolded glucocerebrosidase in Gaucher disease and Parkinson's disease – **Logan M. Glasstetter**

09:48 – 10:00 Restoration of lysosomal acidification rescues mitophagy along with lysosomal and mitochondrial pathophysiology in *GBA1*-mediated Parkinson's Disease – **Preethi Sheshadri**

10:00 – 10:12 GT-02287, a clinical stage allosteric GCase modulator for the treatment of *GBA1*-Parkinson's disease – **Joanne Taylor**

10:12 – 10:24 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 – **Kimberly McDowell**

10:30-11:00 Coffee break

11:00-12:20 Current status of *GBA1* clinical trials

Chair: **Anastacia Henry**

11:00-11:20 The landscape of the current pre-clinical and clinical trials on *GBA1* – **Simon Stott**

11:20-11:40 What can we learn from the MOVES-PD trial? – **Pablo Sardi**

11:40-12:00 Overview of completed and ongoing Ambroxol trials – **Stephen Mullin**

12:00-12:20 Gene therapy for *GBA1*-associated neurodegeneration – **Sebastian Boland**

12:20-14:15 Lunch + posters 26-37

14:00-15:00 Considerations in clinical trial design for *GBA1*-associated neurodegeneration

Chair: **Lorraine Kalia**

14:00-14:20 Trial ready populations and the future of clinical genetic screening – **Peter Bauer**

14:20-14:40 Mutation classification and genetic stratification in *GBA1* trials – **Ziv Gan-Or**

14:40-15:00 Other consideration for inclusion / exclusion and endpoints in *GBA1* trials – **Miguel Fonseca**

15:00-15:30 Moving forward, Awards for best trainee abstracts and concluding remarks – Ziv Gan-Or