

# 3rd IWGGD Symposium, Trieste, Italy (03-06 May 2026)

Venue: Savoia Excelsior Palace, Riva del Mandracchio 4, 34124 Trieste, Italy

Bridging the gap: from basic research to patient care

Sunday 03/May/2026	
15:00 - 16:30	Simultaneous IWGGD Working Groups - Open to Members Only
16:30 - 18:00	Educational Session - Open to Members & Eligible Sponsors Only
19:00 - 20:00	<b>Opening Lecture</b> How it All Began: The Story of Gaucher Disease and the IWGGD <b>Keynote: Bruno Bembi (Italy)</b>
Monday 04/May/2026	
09:00 - 10:45	<b>Session 1: Patients' perspective (Part 1)</b> <b>Moderators: Maurizio Scarpa, Derralynn Hughes, Tanya Collin-Histed</b>
09:00 - 09:45	<b>Plenary Lecture</b> An Overview of the Current Situation of Rare Diseases in Africa <b>Chris Hendriksz (United Kingdom)</b>
09:45 - 10:45	<b>Case Study Presentations</b> African physicians share experiences and challenges in diagnosing and managing GD <b>Pheobe Wamalwa (Kenya), Kandi-Catherine Muze (Tanzania), Magy Abdelwahab (Egypt)</b>
10:45 - 11:15	<b>Coffee Break (Poster Viewing)</b>
11:15 - 12:30	<b>Session 1: Patients' perspective (Part 2)</b> <b>Moderators: Maurizio Scarpa, Derralynn Hughes, Tanya Collin-Histed</b>
11:15 - 11:45	IGA in Africa: Highlights of ongoing initiatives <b>Presented by Roselyn Karungari Kanja (Kenya)</b>

Gold Sponsors:



Silver Sponsors: Bronze Sponsors:



Scientific Committee: Andrea Dardis, Derralynn Hughes, Johannes Aerts, Shoshana Revel-Vilk, Predrag Rodic, Ida Schwartz, Ozlem Goker-Alpan, Jeremy Manuel, Kasper Ter-Horst, Majdolen Istiti

11:45 - 12:30	<b>Selected Oral Presentations</b>
11:45 - 12:00	Expanding Global Reach: The Impact and Evolution of the International Gaucher Alliance's Global Gaucher Connect Programme (GGCP) <b>Vesna Aleksovska (North Macedonia)</b>
12:00 - 12:15	Different diseases, different needs: Patient preferences for gene therapy and beliefs about prescribed medications vary in lysosomal storage diseases <b>Eleonore Corazolla (The Netherlands)</b>
12:15 - 12:30	Guidelines on Home Therapy in Gaucher Disease <b>Carolina Toneloto (Brazil)</b>
12:30 - 13:30	<b>Lunch (Poster Viewing)</b>
13:30 - 15:30	<b>Session 2: Laboratory – Genetics &amp; Biochemistry</b> <b>Moderators: Paula Rozenfeld, Andrea Dardis</b>
13:30 - 14:15	<b>Plenary Lecture</b> GCase genomic modifiers: an opportunity for designing therapeutics <b>Andres Klein (Chile)</b>
14:15 - 15:30	<b>Selected Oral Presentations</b>
14:15 - 14:30	Beyond Sequence Homology: Dissecting Complex Alleles and Epigenetic Landscapes at the GBA1–GBA1LP Locus via Native DNA Nanopore Long-Read Sequencing <b>Natascha Bergamin (Italy)</b>
14:30 - 14:45	Elevated soluble ACE2 in Gaucher patients - an evolutionary advantage? <b>Tova Hershkovitz (Israel)</b>
14:45 - 15:00	Glucosylated phytosterols: a new player in Gaucher disease? <b>Johannes Aerts (The Netherlands)</b>
15:00 - 15:15	Immune and Inflammatory Signatures in Gaucher Disease: Implications for Gene and Precision Therapies. <b>Margarita Ivanova (United States)</b>
15:15 - 15:30	When glucosylsphingosine does not normalize: pharmacogenetic determinants of biochemical–clinical dissociation during eliglustat therapy <b>Sonia Roca-Esteve (Spain)</b>
15:30 - 16:00	<b>Coffee Break (Poster Viewing)</b>

Gold Sponsors:



Silver Sponsors: Bronze Sponsors:



Scientific Committee: Andrea Dardis, Derralynn Hughes, Johannes Aerts, Shoshana Revel-Vilk, Predrag Rodic, Ida Schwartz, Ozlem Goker-Alpan, Jeremy Manuel, Kasper Ter-Horst, Majdolen Istiti

16:00 - 18:15	<b>Session 3: Gaucher Disease Clinical Spectrum</b> <b>Moderators: Shoshana Revel-Vilk, Francesca Carubbi</b>
16:00 - 16:45	<b>Plenary Lecture</b> Incorporating skin evaluation into the standard care of patients with Gaucher disease <b>Ayelet Ollech (Israel)</b>
16:45 - 18:15	<b>Selected Oral Presentations</b>
16:45 - 17:00	Perinatal-Lethal Gaucher Disease: Clinical, Biochemical and Pathological Insights from 15 French Cases <b>Magali Pettazzoni (France)</b>
17:00 - 17:15	Lyso-Gb1 Dynamics in Untreated Patients with Gaucher Disease <b>Shoshana Revel-Vilk (Israel)</b>
17:15 - 17:30	Gaucher Disease type 1 in Elderly Individuals: A Systematic Review <b>François Maillot (France)</b>
17:30 - 17:45	Development and Validation of an Age-Adapted Disease Severity and Burden Scoring System for Neuronopathic Gaucher Disease (nGD/GD3) <b>Leah Svarney (United States)</b>
17:45 - 18:00	Risk and Prevalence of Overweight and Obesity Among Adults with Gaucher Disease <b>Ari Zimran (Israel)</b>
18:00 - 18:15	An AI-Driven Multilayer Model of Lung Involvement in Neuronopathic Gaucher Disease (nGD): Validation in a 45-Patient Cohort and Implications for Management <b>Sara Mitchell (United States)</b>
<b>Tuesday 05/May/2026</b>	
08:30 - 10:30	<b>Session 4: Laboratory – Basic Research</b> <b>Moderators: Johannes Aerts</b>
08:30 - 09:15	<b>Johannes Aerts Basic Science Lecture 2026</b> Decoding the Brain's Lysosomes: A Cell-Type-Specific Protein Atlas and the identification of a new Lysosomal Disorder <b>Ali Ghoochani (United States)</b>
09:15 - 10:30	<b>Selected Oral Presentations</b>

Gold Sponsors:



Silver Sponsors: Bronze Sponsors:



**Scientific Committee:** Andrea Dardis, Derralynn Hughes, Johannes Aerts, Shoshana Revel-Vilk, Predrag Rodic, Ida Schwartz, Ozlem Goker-Alpan, Jeremy Manuel, Kasper Ter-Horst, Majdolen Istiti

09:15 - 09:30	Evaluation of dopaminergic neurons from isogenic iPSC lines derived from a donor with Gaucher disease and Parkinson disease reveal the role of GPNMB in GBA1-associated parkinsonism <b>Ellen Sidransky (United States)</b>
09:30 - 09:45	Mutant microglia from Parkinson's disease patients with heterozygous GBA1 mutations are key determinants of alpha-synuclein aggregation and dopamine neuron pathology <b>Ricardo A. Feldman (United States)</b>
09:45 - 10:00	Utilising machine learning for Gaucher disease: predicting structural defects due to mutation of beta-glucocerebrosidase <b>Thomas J. McCorvie (United Kingdom)</b>
10:00 - 10:15	Novel High-Resolution Exosomal Transcriptomics for Biomarker Discovery in Gaucher Disease <b>Reena V. Kartha (United States)</b>
10:15 - 10:30	Comparison of human and zebrafish glucocerebrosidase and generation of chimeric forms: importance of non-catalytic loops for catalytic activity and transglucosylation <b>Mats J. Bulterman (The Netherlands)</b>
10:30 - 11:00	<b>Coffee Break (Poster Viewing)</b>
11:00 - 13:00	<b>Session 5: Comorbidities in Gaucher Disease</b> <b>Moderators: Predrag Rodic, Ida Schwartz</b>
11:00 - 11:45	<b>Plenary Lecture</b> Developing therapeutic strategies for GBA1-PD: hints from experimental models <b>Alessio Di Fonzo (Italy)</b>
11:45 - 13:00	<b>Selected Oral Presentations</b>
11:45 - 12:00	Longitudinal follow-up of GBA1-carriers for prodromal features of Parkinson disease, Sidransky syndrome <b>Michal Becker-Cohen (Israel)</b>
12:00 - 12:15	Mechanisms Underlying Mutant LRRK2's Modifying Effect on GBA1-Associated Parkinson Disease <b>Vera Serebryany-Piavsky (Israel)</b>
12:15 - 12:30	Non-Motor and Neuropsychological Differences Across the GBA1 Spectrum <b>Ellen Sidransky (United States)</b>

Gold Sponsors:



Silver Sponsors: Bronze Sponsors:



Scientific Committee: Andrea Dardis, Derralynn Hughes, Johannes Aerts, Shoshana Revel-Vilk, Predrag Rodic, Ida Schwartz, Ozlem Goker-Alpan, Jeremy Manuel, Kasper Ter-Horst, Majdolen Istaiti

12:30 - 12:45	Not All 'Mild' Genotypes Are Equal: Divergent Skeletal Burden in c.[1226A>G] Homozygous versus c.[1226A>G]/Mild Gaucher Disease. <b>Irene Serrano-Gonzalo (Spain)</b>
12:45 - 13:00	The "Perfect Storm" of Wnt Inhibitors Drives Early Osteoporosis in Female Patients with Gaucher Disease <b>Margarita Ivanova (United States)</b>
13:00 - 14:00	<b>Lunch (Poster Viewing)</b>
14:00 - 14:45	<b>Session 6: Rapid Communication</b> <b>Moderators: Marc Berger, Reena Kartha</b>
	Expression of Mild GBA1 Mutations in A Fly Model <b>Aparna Kuppuramalingam (Israel)</b>
	Accelerating GD Diagnosis and Enabling Reliability differential detection with ASMD, thanks to MSMS analysis <b>Magali Pettazzoni (France)</b>
	The platelets lipidomic signature in Gaucher Disease <b>Giuseppe Uras (United Kingdom)</b>
	Cellular context matters: strengths and limitations of a GBA1-knockout model for functional assessment of GBA1 variants <b>Maximiliano E. Ormazabal (Italy)</b>
	Parkinson's disease and dementia with Lewy bodies in Gaucher disease: a multicenter longitudinal study <b>Marco Percetti (Italy)</b>
	Severe Secondary GM1 Ganglioside Accumulation in Gaucher Patients' Cell Lines <b>Rodolfo Tonin (Italy)</b>
14:45 - 16:45	<b>IWGGD Annual General Meeting (AGM) - Open to Members Only</b>

Gold Sponsors:



Silver Sponsors: Bronze Sponsors:



**Scientific Committee:** Andrea Dardis, Derralynn Hughes, Johannes Aerts, Shoshana Revel-Vilk, Predrag Rodic, Ida Schwartz, Ozlem Goker-Alpan, Jeremy Manuel, Kasper Ter-Horst, Majdolen Istaiti

Wednesday 06/May/2026

08:30 - 10:15	<b>Session 7: Therapies in Gaucher Disease (Part 1)</b> <b>Moderators: Ozlem Goker-Alpan, Magy Abdelwahab</b>
08:30 - 09:15	<b>Plenary Lecture</b> Genome Editing landscape: from bench to bedside <b>Andres Muro (Italy)</b>
09:15 - 11:15	<b>Selected Oral Presentations</b>
09:15 - 09:30	Therapeutic efficacy of a novel glucocerebrosidase variant in a new preclinical model of neuronopathic Gaucher disease <b>Allan Feng (United States)</b>
09:30 - 09:45	A Phase 1 First-in-Human, Single- and Multiple- Ascending Dose Study of Glucosylceramide Synthase (GCS) Inhibitor YH35995 in Healthy Adult Male Participants <b>YuKyung Kim (Republic of Korea) - Sponsored Presentation by YUHAN</b>
09:45 - 10:00	Two-year follow up of avigbagene parvec (FLT201) investigational AAV gene therapy in adults with Gaucher Disease type 1: Results from GALILEO-1 and GALILEO-2 <b>Ida Schwartz (Brazil) - Sponsored Presentation by Spur Therapeutics</b>
10:00 - 10:15	The PROCEED Study: A Phase 1/2 Dose-Escalation Investigation of Systemic AAV9-Based Gene Therapy for Peripheral Manifestations of Gaucher Disease <b>Ozlem Goker-Alpan (United States) - Sponsored Presentation by Eli Lilly and Company</b>
10:15 - 10:45	<b>Coffee Break (Poster Viewing)</b>
10:45 - 12:00	<b>Session 7: Therapies in Gaucher Disease (Part 2)</b> <b>Moderators: Ozlem Goker-Alpan, Magy Abdelwahab</b>
10:45 - 11:00	Dose Spacing in Enzyme Replacement Therapy for Stable Type 1 Gaucher Disease: A Non-Inferiority Sequential Trial Emulation from the French Gaucher Disease Registry <b>Yann Nguyen (France)</b>
11:00 - 11:15	Safety and efficacy of venglustat versus imiglucerase in patients with Gaucher Disease Type 3 (LEAP2MONO): a phase 3, randomized, double-blind multicenter trial <b>Karl Eugen Mengel (Germany) - Sponsored Presentation by Sanofi</b>
11:15 - 11:30	Chaperone and antioxidant dual action iminosugar hybrids for Gaucher Disease <b>Francesca Clemente (Italy)</b>

Gold Sponsors:



Silver Sponsors: Bronze Sponsors:



Scientific Committee: Andrea Dardis, Derralynn Hughes, Johannes Aerts, Shoshana Revel-Vilk, Predrag Rodic, Ida Schwartz, Ozlem Goker-Alpan, Jeremy Manuel, Kasper Ter-Horst, Majdolen Istiti

11:30 - 11:45	High-Dose Ambroxol for Gaucher Disease Type 3 in Children: A Prospective Exploratory Study <b>Huma Cheema (Pakistan)</b>
11:45 - 12:00	Development of a novel systemic AAV gene therapy for neuronopathic Gaucher disease <b>Ellen Sidransky (United States)</b>
12:00 - 12:15	<b>Best Oral &amp; Poster Presentation Awards</b>
12:15 - 12:30	<i>Closing Remarks</i>

Gold Sponsors:



Silver Sponsors: Bronze Sponsors:



Scientific Committee: Andrea Dardis, Derralynn Hughes, Johannes Aerts, Shoshana Revel-Vilk, Predrag Rodic, Ida Schwartz, Ozlem Goker-Alpan, Jeremy Manuel, Kasper Ter-Horst, Majdolen Istiti