

# 2025 International PWS Conference

## Clinical and Scientific Program Agenda



### DAY ONE | Wednesday, June 25, 2025

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| 6:30 AM                    | <b>Breakfast - Food Trucks</b><br><i>Conference Center Pavilion; Breakfast not included with conference registration</i>   |
| 7:00 AM - 6:00 PM          | <b>Conference Check-In / Help Desk Hours</b><br><i>Conference Center Lobby (first floor)</i>   |
| 8:30 AM - 8:45 AM          | <b>Introduction &amp; Welcome</b> (Main Ballroom - Ironwood Doors Entrance)<br>Dan Driscoll, Chair, Clinical and Scientific Advisory Board, IPWSO & Theresa Strong, Director of Research Programs, Foundation for Prader-Willi Research, USA                     |
| <b>8:45 AM - 10:15 AM</b>  | <b>I. Opening Session Day 1</b> (Main Ballroom - Ironwood Doors Entrance)<br>Moderator: Theresa Strong, Foundation for Prader-Willi Research, USA  |
| 8:45 AM - 9:30 AM          | I-1. Invited Speaker: <b>Obesity, Causes and Treatment: The End of the Beginning</b> - Jeffrey M Friedman, Howard Hughes Medical Institute, USA  |
| 9:30 AM - 10:15 AM         | I-2. Invited Speaker: <b>Epigenetic Activation of the PWS Locus as a Potential Therapeutic Approach</b> - Marnie Blewitt, The Walter and Eliza Hall Institute of Medical Research and Department of Medical Biology, University of Melbourne, Australia          |
| 10:15 AM - 10:45 AM        | <b>BREAK</b>   |
| <b>10:45 AM - 11:45 AM</b> | <b>II. Genetics &amp; Epigenetics</b> (Main Ballroom - Ironwood Doors Entrance)<br>Moderator: Jim Resnick, University of Florida, College of Medicine, USA   |
| 10:45 AM - 11:00 AM        | II-1. <b>Convergence of Transcriptional and Molecular Signatures in Prader-Willi Syndrome</b> - Derek JC Tai, Center for Genomic Medicine, Massachusetts General Hospital, Harvard Medical School, USA   |
| 11:00 AM - 11:15 AM        | II-2. <b>Changes in the Transcriptomic Landscape of the Hypothalamus in Prader-Willi Syndrome</b> - Gina Yosten, St Louis University School of Medicine, USA   |
| 11:15 AM - 11:30 AM        | II-3. <b>Gene Dysregulation in Isogenic Models of Prader-Willi Syndrome</b> - Gordon Carmichael, University of Connecticut Health Center, USA  |
| 11:30 AM - 11:45 AM        | II-4. <b>Transcription Profiling of SNORD109 and SNORD116 in the Human Hypothalamus and Cerebellum identifies Novel Human-Specific Expression Patterns</b> - Lara Lechner, Charité, Universitätsmedizin Berlin, Germany  |
| 11:45 AM - 12:45 PM        | <b>Networking Lunch</b><br><i>Canyon Ballroom</i>  |
| <b>12:45 PM - 1:45 PM</b>  | <b>III. Medical</b> (Main Ballroom - Ironwood Doors Entrance)<br>Moderator: Ann Scheimann, Johns Hopkins Hospital, USA   |
| 12:45 PM - 1:00 PM         | III-1. <b>Is there a role for GLP-1 Receptor Agonists in Prader-Willi Syndrome?</b> - Ann Manzardo, University of Kansas Medical Center, USA   |
| 1:00 PM - 1:15 PM          | III-2. <b>Efficacy and Tolerance of SGLT2 Inhibitors in Diabetic Patients with Prader-Willi Syndrome: Non-Insulin Diabetes Treatment</b> - Christine Poitou, Service de diabétoogie et nutrition, Hôpital Pitié-Salpêtrière, Centre de Référence Pradort, France |
| 1:15 PM - 1:30 PM          | III-3. <b>An AI Program to Determine the Nutritional Phase of PWS</b> - Dan Driscoll, University of Florida College of Medicine, USA   |
| 1:30 PM - 1:45 PM          | III-4. <b>The Clinical Presentation of Prader-Willi in the Inpatient and Emergency Settings: Insights from Real-World Evidence</b> - James Luccarelli, Massachusetts General Hospital and Harvard Medical School, USA  |
| 1:45 PM - 1:55 PM          | <b>BREAK</b>   |

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| <b>1:55 PM - 2:40 PM</b> | <b>IV. Oral/Poster Lightning Talks Session 1</b> ( <i>Main Ballroom - Ironwood Doors Entrance</i> )<br>Moderator: Dan Driscoll, University of Florida College of Medicine, USA   |
| 1:55 PM - 2:00 PM        | IV-1. <b>Functional Near-Infrared Spectroscopy of Skin Picking Behavior in Prader-Willi Syndrome</b> - Scott Hall, Stanford University School of Medicine, USA   |
| 2:00 PM - 2:05 PM        | IV-2. <b>The Heart of SNORD116: Investigating the Role of SNORD116 in Cardiomyocyte Development</b> - James Smith, University of East Anglia, United Kingdom   |
| 2:05 PM - 2:10 PM        | IV-3. <b>Reduction in Hyperphagia in ARD-101 Phase 2 Clinical Trial Informs Phase 3 HERO Trial in PWS</b> - Manasi Jaiman, Aardvark Therapeutics, USA  |
| 2:10 PM - 2:15 PM        | IV-4. <b>Assessing Osteoarticular Challenges in Prader-Willi Syndrome: Insights into postural stability, spinal deformities, and functional mobility</b> - Jorgelina Stegmann, Fundación Spine, Argentina                      |
| 2:15 PM - 2:20 PM        | IV-5. <b>N-Acetylcysteine Treatment for Skin-Picking in Children and Young Adults with PWS: A randomized placebo-controlled cross-over trial</b> - Demi Trueba-Timmermans, Erasmus MC Sophia, The Netherlands                  |
| 2:20 PM - 2:25 PM        | IV-6. <b>Circulating Levels of Ghrelin in Patients with a Rare Neurodevelopmental Disorder Associated with Hyperphagia, and/or Overweight, and/or Obesity – The HOGGRID study</b> - Gwenaëlle Diene, CHU de Toulouse, France   |
| 2:25 PM - 2:30 PM        | IV-7. <b>The Burden of Anxiousness and Distress Behaviors in Prader-Willi Syndrome</b> - Deborah Hoffman, Acadia Pharmaceuticals Inc, USA  |
| 2:30 PM - 2:35 PM        | IV-8. <b>Mining Biological Networks at Scale: A Path to New Therapeutic Targets in Prader-Willi Syndrome</b> - Rohit Singh, Duke University, USA   |
| 2:35 PM - 2:40 PM        | IV-9. <b>Study Design and Ethical Return of Results for a Fully Remote Genome Sequencing Study in Individuals with Prader-Willi Syndrome</b> - Caroline Vrana-Diaz, Foundation for Prader-Willi Research, USA                  |
| 2:40 PM - 3:00 PM        | <b>BREAK</b>   |
| <b>3:00 PM - 4:00 PM</b> | <b>V. Oral/Poster Lightning Talks Session 2</b> ( <i>Main Ballroom - Ironwood Doors Entrance</i> )<br>Moderator: Theresa Strong, Foundation for Prader-Willi Research, USA   |
| 3:00 PM - 3:05 PM        | V-1. <b>Delivery of Genes and Gene Editors using Programmable Milk Exosomes</b> - Janos Zemleni, University of Nebraska-Lincoln, USA   |
| 3:05 PM - 3:10 PM        | V-2. <b>Learning from People Living with PWS Across the World About How to Best Support their Flourishing</b> - Kate Woodcock, University of Birmingham, United Kingdom  |
| 3:10 PM - 3:15 PM        | V-3. <b>Role of Mitochondrial Function in the Oxidative Stress Profile of Children with Prader-Willi Syndrome</b> - Alvaro Carrasco-Garcia, Erasmus University Medical Center, The Netherlands                                 |
| 3:15 PM - 3:20 PM        | V-4. <b>Novel Insights into the Function of MAGEL2 in Fed and Fasted States</b> - Klementina Fon Tacer, Texas Tech University, School of Veterinary Medicine, USA  |
| 3:20 PM - 3:25 PM        | V-5. <b>Sensory Information Processing, Intelligence, and Behavior in Prader-Willi Syndrome</b> - Anja Roubos, Centre of Excellence for Neuropsychiatry, Vincent Van Gogh, Institute for Psychiatry, The Netherlands           |
| 3:25 PM - 3:30 PM        | V-6. <b>Long-Term Outcomes of Prader-Willi Syndrome Association USA Funding Awards</b> - Ann Scheimann, Johns Hopkins Hospital, USA  |
| 3:30 PM - 3:35 PM        | V-7. <b>The Dutch experience with Weight-Loss Drugs in Adults with Prader-Willi Syndrome</b> - Jacqueline Goos, Erasmus MC University Medical Center, The Netherlands  |
| 3:35 PM - 3:40 PM        | V-8. <b>Respiratory and Sleep Disorders in Children with Prader Willi Syndrome: prevalence and characterization</b> - Sofia Suco, Hospital de Niños icardo Gutierrez - Cedic, Buenos Aires, Argentina                          |
| 3:40 PM - 3:45 PM        | V-9. <b>Eating behavior and Hyperphagia in Adults with Obesity and Neurodevelopmental Disorders: Insights from Prader-Willi, Bardet-Biedl and other genetic syndromes</b> - Emilie Guillon, Pitié-Salpêtrière Hospital, France |
| 3:45 PM - 3:50 PM        | V-10. <b>Hypercapnia is Common in Lean and Obese Adults with Prader Willi Syndrome</b> - Kristen Davidse, Erasmus University Medical Centre, The Netherlands   |
| 3:50 PM - 3:55 PM        | V-11. <b>International Survey of Clinicians' Perspectives Toward Growth Hormone Therapy and Sleep Evaluation in Infants with Prader-Willi Syndrome</b> - Emily Paprocki, Children's Mercy Kansas City, USA                     |
| 4:00 PM - 6:00 PM        | <b>VI. Poster Presentation Session</b> ( <i>Arizona E, H - Eucalyptus &amp; Honeysuckle - Honeysuckle Door Entrance</i> )<br>Light refreshments will be served.  |

## DAY TWO | Thursday, June 26, 2025

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| 6:30 AM                    | <b>Breakfast - Food Trucks</b><br><i>Conference Center Pavilion; Breakfast not included with conference registration</i>  |
| 7:00 AM - 8:00 PM          | <b>Conference Check-In / Help Desk Hours</b><br><i>Conference Center Lobby (first floor)</i>  |
| <b>8:15 AM - 10:00 AM</b>  | <b>VII. Opening Session Day 2 (Main Ballroom - Ironwood Doors Entrance)</b><br><b>Neuroscience, Cognition, and Mental Health</b><br>Moderator: Janice Forster, Pittsburg Partnership, USA   |
| 8:15 AM - 9:00 AM          | VII-1. Invited Speaker: <b>Maintaining Well-Being, Improving Mental Health, and Managing Challenging Behaviors: Outcomes from the IPWSO Mental Health Initiative</b> - Anthony J Holland, Professor Emeritus, University of Cambridge, United Kingdom   |
| 9:00 AM - 9:15 AM          | VII-2. <b>How Oxytocin Acts and Restores the Suckling Behavior in Magel2 Deficient Newborn Mouse: consequences on Prader-Willi and Schaaf-Yang Syndrome</b> - Françoise Muscatelli, Institut de Neurobiologie de la Méditerranée, France  |
| 9:15 AM - 9:30 AM          | VII-3. <b>Morphological and Transcriptional Alterations of Microglia in the Magel2 KO Mouse Model of Prader-Willi Syndrome</b> - Ferdinand Althammer, Institute of Human Genetics, Heidelberg University Hospital, Germany  |
| 9:30 AM - 9:45 AM          | VII-4. <b>Physiological, Behavioral, and Neurodevelopmental Consequences of the Combined Loss of Necdin and Magel2</b> - Sebastien Bouret, INSERM UMR-S 1172 Lille Neuroendocrinology, France   |
| 9:45 AM - 10:00 AM         | VII-5. <b>Investigating Mechanisms of Psychosis in Prader-Willi Syndrome using Magnetic Resonance Spectroscopy</b> - Suzannah Lester, University of Cambridge, United Kingdom   |
| 10:00 AM - 10:15 AM        | <b>BREAK</b>  |
| <b>10:15 AM - 11:00 AM</b> | <b>VIII. Moderator: Charlotte Höybye, Karolinska Institute, Sweden (Main Ballroom - Ironwood Doors Entrance)</b><br>Invited Speaker: <b>Mapping Brain Circuitry Involved in Hyperphagia in Prader-Willi syndrome to Identify Novel Treatment Targets</b> - Laura Holsen, Associate Professor of Psychiatry, Harvard Medical School, USA |
| <b>11:00 AM - 12:00 PM</b> | <b>IX. Clinical / Clinical Trials (Main Ballroom - Ironwood Doors Entrance)</b><br>Moderator: Charlotte Höybye, Karolinska Institute, Sweden  |
| 11:00 AM - 11:15 AM        | IX-1. <b>Long-term Efficacy Results of Diazoxide Choline Extended-Release (DCCR) Tablets in Participants with Prader-Willi Syndrome from the Completed C601 (DESTINY PWS) and C602 Open Label Extension (OLE) Studies</b> - Evelien Gevers, Queen Mary University of London, Barts and the London Medical School, United Kingdom        |
| 11:15 AM - 11:30 AM        | IX-2. <b>A Randomized, Double-Blind, Controlled Trial of Bright Light Therapy on All-Cause Excessive Daytime Sleepiness in Prader-Willi Syndrome</b> - Hasan Mustafic, Maimonides Medical Center, USA   |
| 11:30 AM - 11:45 AM        | IX-3. <b>My Hunger Questionnaire (My-HQ): Progress in Developing a Self-Report Measure of Hyperphagia for Individuals with Prader-Willi Syndrome</b> - Elisabeth Dykens, Vanderbilt University, USA   |
| 11:45 AM - 12:00 PM        | IX-4. <b>The James Henson PWS Goal Inventory: An Adaptation of Goal Attainment Scaling for Prader-Willi Syndrome</b> - Maria Picone, TREND Community, USA   |
| 12:00 PM - 1:00 PM         | <b>Networking Lunch</b><br><i>Canyon Ballroom</i>   |
| <b>1:00 PM - 2:30 PM</b>   | <b>X. Clinical Care (Concurrent Session) (Main Ballroom - Ironwood Doors Entrance)</b><br>Moderator: Constanze Lämmer, Section of Paediatric Endocrinology and Diabetology, Dept. of Paediatrics and Adolescent Medicine, KJF Klinikum Josefinum, Augsburg, Germany   |
| 1:00 PM - 1:15 PM          | X-1. <b>Serious Medical Events and PWS-associated behaviors in PATH for PWS: A Non-Interventional, Observational, Natural History Study of Prader-Willi Syndrome</b> - Lisa Matesevac, Foundation for Prader-Willi Research, USA  |

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| 1:15 PM - 1:30 PM        | X-2. <b>Utility of Baseline Polysomnograms Prior to Growth Hormone Initiation in Young Infants with PWS</b><br>- Sani Roy, Cook Children's Medical Center, USA   |
| 1:30 PM - 1:45 PM        | X-3. <b>The Effects of Growth Hormone Treatment on Muscle Strength in Adults with Prader-Willi Syndrome</b> - Jannek Baan, Erasmus Medical Centre, The Netherlands   |
| 1:45 PM - 2:00 PM        | X-4. <b>Testosterone Replacement Therapy in Adolescents and Young Adults with Prader-Willi Syndrome: Efficacy and Effects on Behavior</b> - Gerthe Kerhof, Dutch Reference Centre for PWS, The Netherlands             |
| 2:00 PM - 2:15 PM        | X-5. <b>Developmental Characterization of Phenotypic Behaviors in PWS</b> - Janice Forster, Pittsburgh Partnership, USA  |
| 2:15 PM - 2:30 PM        | X-6. <b>The Lived Experiences of Teens and Adults Who Grew Up with a Sibling with Prader-Willi Syndrome</b> - Lauren Schwartz, Foundation for Prader-Willi Research, USA   |
| <b>1:00 PM - 2:30 PM</b> | <b>XI. Molecular (Concurrent Session) (Acacia)</b><br>Moderator: Jim Resnick, University of Florida, College of Medicine, USA  |
| 1:00 PM - 1:15 PM        | XI-1. <b>Light and Sex Modify Snord116 Genotype Effects on Metabolism, Behavior, and Imprinted Gene Networks following Circadian Entrainment</b> - Estefania Azevedo, MUSC, USA  |
| 1:15 PM - 1:30 PM        | XI-2. <b>In Silico Drug Repurposing Using Human Brain Omics Data Reveals Targetable Pathways for Prader-Willi Syndrome</b> - Olivia Veatch, University of Kansas Medical Center, USA                                   |
| 1:30 PM - 1:45 PM        | XI-3. <b>A Cholinergic Hypothesis Driving Hyperphagia and Cognitive Deficits in Prader-Willi Syndrome</b> - Estefania Azevedo, MUSC, USA   |
| 1:45 PM - 2:00 PM        | XI-4. <b>The Similarities between Congenital Myasthenic Syndrome-22 and Prader-Willi Syndrome: Activating the PREPL Pathway as a Potential Therapeutic Approach for PWS</b> - Yenthe Monnens, KU Leuven, Belgium       |
| 2:00 PM - 2:15 PM        | XI-5. <b>Does SNORD116 Play a Role in Ribosome Biogenesis?</b> - Amanda Whipple, Harvard University, USA   |
| 2:15 PM - 2:30 PM        | XI-6. <b>An innovative delivery of CRISPR/dCas9 epigenome editing-based therapy for Prader-Willi Syndrome</b> - Xiaona Lu, Yale University School of Medicine, USA   |
| 2:30 PM - 3:00 PM        | <b>Break and Poster Viewing</b><br><i>Main Ballroom and Arizona E, H - Eucalyptus &amp; Honeysuckle</i>  |
| <b>3:00 PM - 4:00 PM</b> | <b>XII. Closing Session (Main Ballroom - Ironwood Doors Entrance)</b><br>Moderator: Tony Holland, University of Cambridge, UK  |
| 3:00 PM - 3:15 PM        | XII-1. <b>Hypothalamic-Related Neural Activity at Rest in Prader-Willi Syndrome</b> - Sanaz Hosseini, University of Cambridge, United Kingdom  |
| 3:15 PM - 3:30 PM        | XII-2. <b>Oxytocin System Dysregulation is a Key Feature in Magel2 Pmut Rats and Associated with Changes in Protein Secretion Pathways</b> - Tim Schubert, Institute of Human Genetics, Heidelberg University, Germany |
| 3:30 PM - 3:45 PM        | XII-3. <b>High Throughput Screening to Identify Drugs that Ameliorate Secretory Granule Phenotypes in PWS iPSC-Derived Neurons</b> - Anne Bang, Sanford Burnham Prbys Medical Discovery Institute, USA                 |
| 3:45 PM - 4:00 PM        | XII-4. <b>Current Psychopharmacological Prescribing Practices in Prader-Willi Syndrome - Findings from the PATH for PWS Study</b> - Deepan Singh, Maimonides Medical Center, USA                                       |
| <b>4:00 PM - 4:30 PM</b> | <b>XIII. Close (Main Ballroom - Ironwood Doors Entrance)</b>   |
| 4:00 PM - 4:15 PM        | XIII-1. <b>Outstanding Young Investigator Presentation</b><br>Chair: Ann Scheimann, Clinical and Scientific Advisory Board, PWSA   USA   |
| 4:15 PM - 4:30 PM        | XIII-2. <b>Final Comments</b>  |