



WORKSHOP ON RARE SYNDROMIC BODY FAT DISORDERS

What Can They Teach Us?

March 1 - 2, 2012

Lister Hill Auditorium, Bethesda, MD

Agenda

Day 1 – Thursday, March 1, 2012

- 7:30 – 8:15 a.m. **Registration**
- 8:15 – 8:30 a.m. **Welcome**
Gregory Germino, Deputy Director of NIDDK
- 8:30 – 9:15 a.m. **Obesity: Insights From Human Genetic Studies**
Sadaf Farooqi, University of Cambridge
- 9:15 – 10:00 a.m. **Genetic Lipodystrophies: Disorders of Adipose Tissue Development, Differentiation, and Death**
Abhimanyu Garg, The University of Texas Southwestern Medical Center
- 10:00 – 10:30 a.m. **Break**

Session I: Pluses and Minuses of Models Used to Investigate Rare Fat Deposition Disorders

Moderator: Abhimanyu Garg

This session will highlight several different experimental systems used by investigators to elucidate the details of the molecular mechanism leading to altered fat deposition and obesity. Speakers should explain why the system used was chosen, and detail the advantages and limitations of the system.

- 10:30 – 11:00 a.m. **Brain-Derived Neurotrophic Factor in WAGR Syndrome and Non-Syndromic Obesity**
Joan Han, National Institute of Child Health and Human Development
- 11:00 – 11:30 a.m. **Ciliopathies and Fat Deposition**
Philip Beales, University College London

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- 11:30 – 12:00 p.m. **Loss of Processed snoRNA (psnoRNA) Expression in Prader-Willi Syndrome Changes Expression of Genes Involved in Energy Metabolism**
Stefan Stamm, University of Kentucky
- 12:00 – 12:30 p.m. **Use of IPS Cells to Study Lipodystrophies and Other Diseases of Fat Deposition**
Chad Cowan, Harvard University
- 12:30 – 2:00 p.m. **Lunch and Poster Session**

Session II: Genetic Determinants of Metabolic Diseases

Moderator: Jay Shendure

This session will focus on recent technological advances in functional genomics and will address major challenges in identifying causal variants in human disease.

- 2:00 – 2:30 p.m. **Genetic Approaches for Finding Phenotypically Causal Variants in Human Disease**
Jay Shendure, University of Washington
- 2:30 – 3:00 p.m. **Searching for Genes for the Metabolically Healthy Obese (and the Metabolically Unhealthy Lean) to Identify New Physiological Pathways That Underlie Adiposity and Fat Deposition**
Ruth Loos, Mount Sinai School of Medicine
- 3:00 – 3:15 p.m. **A Novel Null Mutation in Human Hormone Sensitive Lipase (HSL) Is Associated With Lipodystrophy, Dyslipidemia, and Insulin Resistance**
Coleen M. Damcott, University of Maryland School of Medicine
- 3:15 – 3:45 p.m. **Break**

Several meeting participants with interesting patients/leads/mouse models taken from submitted abstracts will be asked to present a few background slides, which will then be followed by a discussion with the group about what could be done to move these projects forward.

- 3:45 – 5:15 p.m. **Mice Deficient of the Inflammatory Gene—FAT10, Display Markedly Reduced Fat Formation and Increased Longevity**
Allon Canaan, Yale University School of Medicine
- Characterization of New Non-Coding RNAs That Regulate Genes Involved in Fat/Energy Metabolism**
Marina Falaleeva, University of Kentucky
- A Novel Mouse Model of Combined Lipomatosis and Partial Lipodystrophy Reveals White Adipocytes Arise From Myf5+ Precursors**
David Guertin, University of Massachusetts Medical School
- XL α s Antagonizes Metabolic Control by G $_s$ α *In Vivo***
Ahmed Kablan, NIDDK

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5:15 p.m. **Adjourn**

Day 2 – Friday, March 2, 2012

7:30 – 8:15 a.m. **Registration**

Session III: Pleiotropic “Obesity Syndromes”

Moderator: Philip Beales

This session will examine rare genetic syndromes for which obesity is manifest in many but not all individuals. Speakers should explore possible mechanisms underlying the variable expression of obesity in these syndromes.

8:15 – 8:45 a.m. **Obesity and Associated Traits in Alström Syndrome**
Jürgen Naggert, The Jackson Laboratory

8:45 – 9:15 a.m. **Obesity and X-linked Mental Retardation: Mechanistic Insights Into Börjeson-Forssman-Lehmann Syndrome**
David Picketts, Ottawa Hospital Research Institute

9:15 – 9:45 a.m. **Imprinting, Brain Development, and Pleiotropy of 15q11-q13 Disorders**
Janine M. LaSalle, University of California, Davis, School of Medicine

9:45 – 10:15 a.m. **Break**

Session IV: Treatment of Rare Syndromic Body Fat Disorders

Moderator: Jack Yanovski

This session will explore approaches being taken to develop effective treatments for rare, sometimes pleiotropic “obesity syndromes.” Speakers should comment on some of the challenges encountered in trying to treat rare diseases involving multiple pathways and tissues even when the cellular defect is known.

10:15 – 10:45 a.m. **NIH Resources to Support Translational Research**
Gurmit Grewal, National Center for Advancing Translational Sciences

10:45 – 11:00 a.m. **Pharmacological Chaperones: Potential Therapeutic Approach to Treat MC4R-Linked Early Onset Obesity**
Patricia René, University of Montreal

11:00 – 11:15 a.m. **Biochemical and Molecular Characterization of the Serotonin Receptor 2C (HTR2C) in Energy Homeostasis Control**
Manli Shen, University of Kentucky

11:15 – 11:45 a.m. **Rare Mutations in the Leptin-Melanocortin Pathway: Implications for the Treatment of Obesity**
Christian Vaisse, University of California, San Francisco

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- 11:45 – 12:15 p.m. **Learning on the Job in Lipodystrophy: Going From Therapy to Pathophysiology**
Elif Oral, University of Michigan
- 12:15 – 12:45 p.m. **Identification of Allosteric Modulators of the MC4 Receptor for Treatment of
Melanocortin Obesity Syndrome**
Roger Cone, Vanderbilt University Medical Center
- 12:45 – 1:00 p.m. **Closing Remarks**
- 1:00 p.m. **Adjourn**