

Lipodystrophy and the Metabolic Consequences of Altered Fat Deposition

November 2-3, 2006

Lister Hill Auditorium

Bethesda, MD

Meeting Summary

Dr. Phil Smith (co-director, Office of Obesity Research, NIDDK) introduced the meeting, noting that the primary objective of the workshop was to stimulate research that will translate basic cell biological, genetic and molecular findings about proteins and lipids regulating fat mass and fat deposition into potential therapies for genetic and acquired Lipodystrophies and related rare diseases. The first international workshop on Lipodystrophy co-sponsored by the NIDDK and the Office of Rare Diseases was held in March of 2001. During the 5 year period following this first meeting, tremendous progress has been made in understanding some of the previously unknown proteins and molecular pathways that are defective in rare diseases such as Berardinelli-Seip syndrome, Congenital generalized Lipodystrophy, Lawrence syndrome, Barraquer-Simons syndrome, Familial partial Lipodystrophy (Dunnigan, Kobberling and Mandibuloacral Dysplasia variety), and SHORT syndrome.

The meeting was well-attended, with 110 participants. The audience included clinicians, clinical researchers and basic scientists at all career levels from both U.S. and several foreign academic institutions, the NIH, and the private sector. This venue was a wonderful opportunity to unite researchers interested in understanding the molecular mechanisms that are disrupted in syndromes of Lipodystrophy with geneticists, clinical investigators and clinicians working with patients with these rare diseases, and to encourage young students, talented postdocs, and new faculty to investigate Lipodystrophy syndromes.

On the first morning, speakers focused on the genetic etiologies of Lipodystrophies, beginning with Abhimanyu Garg's (UT Southwestern) introductory overview of the clinical syndromes of Lipodystrophies. The morning session ended with an exciting hot topic presentation by Joycelyne Magre who described a new Berardinelli-Seip Congenital Lipodystrophy locus (BSCL3). Mutations in the BSCL3 gene, which encodes caveolin-1, were shown to be strongly associated with the syndrome. Researchers are now working to understand how mutations in caveolin-1 lead to BSCL in certain families. Following an exciting and well attended poster session, the afternoon speakers talked about five animal models with altered fat deposition. Generation and study of these models of Lipodystrophies have provided significant insights into the molecular pathways that are important for regulating the life cycle of the adipocyte and fat cell mass.

The second morning was devoted to talks about clinical studies. Phil Gorden (NIDDK) focused on the use of leptin therapy in the treatment of Lipodystrophy, and Steve Grinspoon (MGH) and Jacqueline Capeau (Univ. Pierre de Marie Curie) on treatment strategies for HIV-related Lipodystrophies. The final series of presentations focused on different aspects of the life cycle of the adipocytes in cell systems and in mice. Kaveh Ashrafi (UCSF) also spoke about how *c. elegans* can be easily manipulated and used to determine the regulatory networks controlling fat deposition that are also likely to be operating in humans.

In all, there were 22 scientific talks, 4 of which were selected from abstracts submitted by junior investigators. Twenty-six posters were presented at an active and crowded poster session. Five junior investigators who presented posters also received travel awards to help defray the costs of attending the meeting.

The abstract book for the meeting will be available on the NIDDK website at: <http://www.niddk.nih.gov/fund/other/lipodystrophy>. I have also enclosed a copy of the meeting booklet in this package. If you would like any additional copies please let me know and I will have them sent to you.

Carol