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RARE DISEASE WORKSHOP SERIES
Improving the *Clinical Development Process*

Workshop #2

Clinical Evaluation of Rare Disease Treatments

June 14-15, 2011

Welcome
Day 2

Emil D. Kakkis, M.D., Ph.D.
President, Kakkis EveryLife Foundation



Workshop #2 Agenda: Day #2

Rare Disease Experiences

Day 2 – Morning

8:00 - 8:30 am Breakfast

8:30 am Brief previous day review

Emil Kakkis, M.D., Ph.D., President, Kakkis EveryLife Foundation

- Examples of development of clinical evaluation tools to assess degree of clinical benefit in neurological and rare diseases

8:50 am **Ron Crystal, M.D.**, Prof. of Genetic Medicine, Weill Cornell — Natural history data, development of LINCL clinical scale, interpretation of brain imaging scans

9:20 am **Ali Skrinar, M.A., MPH**, Senior Director, Clinical Research & Regulatory Affairs, Enobia Pharma — Development of rare disease evaluation tools

9:50 am Midmorning Break

10:10 am **P.K. Tandon, Ph.D.**, Senior Vice President, Global Biomedical Data Sciences and Informatics, Genzyme Corporation — Use of registry information to inform on clinical evaluation of rare diseases

10:40 am **Andy Blight, Ph.D.**, Chief Science Officer, Acorda Therapeutics — Successful application of patient reported outcomes



Workshop #2 Agenda: Day 2

Multi-domain assessment of rare disease

Day 2 - Afternoon

- Evaluation of clinical benefit across multiple physiological domains
 - 12:55 pm **Emil Kakkis, M.D., Ph.D.**, President, Kakkis EveryLife Foundation — The necessity for improving the accessibility to multi-domain assessments in rare disease treatment development: A proposal for moving forward
 - 1:25 pm **L.J. Wei, Ph.D.**, Professor of Biostatistics, Harvard School of Public Health — Approaches and challenges to multi-domain analysis and study design
 - 1:45 pm **James Signorovitch, Ph.D.**, Manager, Analysis Group — Evaluation of various statistical approaches for multiple domains using data from three MPS studies



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Workshop #2 Agenda: Day 2 Rare Disease Experiences Developing Endpoints and PRO's for Rare Diseases

- **11:10 am Discussion**
 - What are the key challenges observed in rare disease-specific instrument development and what practical solutions might allow for better development, utilization, and qualification of such measures?
 - What is the critical information that might be derived from similar disease states to the one being studied that might support the qualification of an evaluative tool as reasonable for a rare disease?



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Workshop #2 Agenda: Day 2

Multiple Domain Assessment in Rare Diseases: How do we develop a multi-domain strategy?

- **2:15 pm Discussion**

- How do we capture and interpret the real effect of a treatment on patients using multi-domain endpoints?
- How would we then interpret clinical data from multi-domain results and accurately make labeling claims based on the result of a multi-domain analysis?



Thanks to participants and organizers

- Tom Fleming, Ph.D., John Powers III, M.D.
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- Brigitta Miyamoto
- FDA personnel
- NIH personnel
- Academic presenters
- Industry presenters