



## AGENDA FOR DYSTONIA COALITION'S SIXTH ANNUAL MEETING

### Day 1: Research Priorities for the Genetics of Dystonia

6-7 November 2014

St. Louis, MO. USA

8:00am Welcome Ozelius/Jinnah  
8:05am Goals for the meeting

#### Session 1: The basics and the first gene

Chair: Ozelius

8:10-8:30am Genetics for the uninitiated  
*This lecture will be a basic introduction for non-genetics people so they are familiar with basic concepts in genetics used during later discussions (dominant/recessive, risk genes, penetrance, monogenic/polygenic, importance of few vs many mutations in a gene, etc...)*  
Comments & questions  
Celeste Karch

8:35-8:55am Clinical features of dystonia and their relevance for gene discovery Joel Perlmutter  
*This lecture will be a primer for non-neurologists so they are familiar with some of the unique clinical features of the dystonias as they relate to gene discovery and pathogenesis.*  
Comments & questions

9:00-9:20am How do we find new genes? Andrew Singleton  
*This lecture will introduce the various types of gene discovery methods available, their strengths & weaknesses. (family studies, exome sequencing, GWAS, linkage, candidate genes, etc...)*  
Comments & questions

9:25-9:45am TOR1A & DYT1 dystonia Susan Bressman  
*This lecture will describe what we have learned about dystonia from the first gene identified. It is not intended as a historical review of what we learned specifically about the gene and its product, but rather how studying DYT1 dystonia may provide a model for understanding broader issues in dystonia.*  
Comments & questions

#### 9:50-10:10 BREAK

#### Session 2: Update on known genes: what is their significance for understanding dystonia?

Chair: Perlmutter

10:10-10:30am THAP1 & DYT6 dystonia Laurie Ozelius  
*This lecture will not review the discovery of the THAP1 gene in detail, but will focus instead on how learning about the THAP1 gene and its function advances our knowledge of the dystonias in general.*  
Comments & questions

10:35-10:55am New genes for isolated dystonia syndromes (CIZ1, GNAL, ANO3, TUBB4) Mark LeDoux  
*This lecture will not review the discovery of these genes in detail but will instead describe how learning about the genes and their functions advances our knowledge of the dystonias in general.*  
Comments & questions

11:00-11:20am Dopa-responsive dystonia syndromes (GCH1, TH, AADC, PTPS) Marie Vidailhet  
*This lecture will not describe the discovery of these genes in detail but will instead focus on describing how studies of this group of disorders advance our knowledge of the dystonias in general.*  
Comments & questions

**AGENDA FOR DYSTONIA COALITION'S SIXTH ANNUAL MEETING**  
**Day 1 continued: Research Priorities for the Genetics of Dystonia**

11:25-11:45am Degenerative dystonia/parkinsonism syndromes Kailash Bhatia  
*This lecture will not describe the discovery of these genes in detail, but will describe instead how this group of disorders advances our knowledge of the dystonias in general. It will not describe how the genes were found.*  
Comments & questions

11:50-12:10pm Are there shared pathways of pathogenesis? Buz Jinnah  
*This lecture will address the similarities and differences among the different dystonic disorders, and whether there are shared pathways of pathogenesis that could serve as targets for therapeutic interventions.*  
Comments & questions

**12:15-1:30pm LUNCH**

**Session 3: Challenge questions**

**Chair: Jinnah**

1:30-1:45pm How important is it to continue the search for more genes? Standaert  
*This lecture will address what has been learned about gene discovery for other disorders, such as Parkinson's disease or the spinocerebellar ataxias, and the value of continuing the search for every disease causing gene. There will be 10 minutes of discussion following.*  
Comments & questions

1:55-2:15pm Is dystonia a neurodevelopmental or neurodegenerative disorder? Dauer/Norris  
*The first speaker will take 10 minutes to present the developmental perspective and the second will take 10 minutes to present the degenerative perspective. There will be 10 minutes of discussion following.*  
Comments & questions

2:25-2:55pm What are the best experimental models for exploring pathogenesis? Klein/LeDoux /Hallett  
*The discovery of a new gene opens the door to an enormous number of potential studies including cell and animal models, and imaging or physiological studies of defined patient groups. The first speaker will address the value of cell models, the second will address the value of animal models, and the last will address human studies. Each will speak for 10 minutes and there will be 15 minutes of discussion following.*

**3:10-3:30 BREAK**

**Session 4: Research priorities**

**Chairs: Teller/Galpern**

3:30-4:10pm Panel Discussion: What are the priorities for research in the genetics of dystonia?  
*A panel of 3-5 people will lead this discussion, based on the prior lectures and personal experience. We need to include people with differing opinions on what would be most important. This discussion might include number and quality of available specimens, the importance of the associated phenotype, and what are the best ways to exploit collected materials for discovering new genes. The goal is to delineate the most important research priorities for the future. Panelists will be identified from individuals attending the meeting*

**Chairs: Hallett/Sieber**

4:15-4:55pm Panel Discussion: After identifying a new gene, what is the most important thing to do first?  
*This will be a panel of 3-5 people to lead this discussion. We need to include people with differing opinions on what would be most important. The goal is to delineate the most important research priorities for the future. Panelists will be identified from individuals attending the meeting*

5:00 pm Closing comments

**DINNER on your own**



**AGENDA FOR DYSTONIA COALITION'S SIXTH ANNUAL MEETING**

**Day 2: Dystonia Coalition Progress & Plans**

6-7 November 2014

St. Louis, MO. USA

<b>7:00am</b>	<b>BREAKFAST</b>	
<b>8:00am</b>	<b>Welcoming Remarks</b>	Ami Rosen
<b>8:05am</b>	<b>Dystonia Coalition: Where are we now &amp; where are we going?</b>	Buz Jinnah
	Progress Report	
	Thanks to Our Sponsors	
	Future Plans	
8:25am	Questions	
<b>8:35am</b>	<b>Project 1: Natural History &amp; Biorepository</b>	
	Progress Report & Next Steps	Joel Perlmutter
8:55am	Questions	
<b>9:05am</b>	<b>Project 2: Comprehensive Rating Scales for Cervical Dystonia</b>	
	Progress Report and Next Steps	Cynthia Comella
9:25am	Questions	
<b>9:35am</b>	<b>Project 3: Diagnostic &amp; Rating Scales for Spasmodic Dysphonia</b>	
	Progress Report and Next Steps	Christy Ludlow
9:55am	Questions	
<b>10:05am</b>	<b>Closing remarks</b>	Buz Jinnah

**GENERAL MEETING ADJOURNED**

**10:30am DC Steering Committee & Executive Committee Meeting**

**1:00pm STEERING COMMITTEE & EXECUTIVE COMMITTEE MEETING ADJOURNED**