11th Annual Meeting
Dublin, Ireland
31 May 2023

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<table>
<thead>
<tr>
<th>Time</th>
<th>Session</th>
<th>Chair</th>
</tr>
</thead>
<tbody>
<tr>
<td>8:15-8:30</td>
<td>Welcoming Remarks &amp; Introduction</td>
<td>Jinnah</td>
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<td>A brief overview of The Dystonia Coalition</td>
<td>Jinnah</td>
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<tr>
<td>8:40-8:55</td>
<td>Natural History Project</td>
<td>Perlmutter</td>
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<td>9:05-9:20</td>
<td>Biobank Project</td>
<td>Perlmutter</td>
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<td>9:30-9:45</td>
<td>Objective Measures Project</td>
<td>Peterson</td>
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<td>9:55-10:05</td>
<td>COFFEE BREAK</td>
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<td>10:05-10:20</td>
<td>Patient-Centered Outcomes Project</td>
<td>Richardson</td>
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<td>10:30-10:40</td>
<td>Pilot Project Program: A program that supports pilot projects</td>
<td>Jinnah</td>
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<td>10:50-11:00</td>
<td>Career Development: A program that supports junior investigators</td>
<td>Jinnah</td>
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<td>11:10-11:20</td>
<td>Patient Advocacy Groups (PAG)</td>
<td>Hiesheter &amp; Kuman</td>
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<td>11:30-11:45</td>
<td>Future of the DC</td>
<td>Chair: Jinnah &amp; Perlmutter</td>
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<td>Introduction</td>
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<td>Discussion / Suggestions</td>
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<td>12:00-12:30</td>
<td>LUNCH</td>
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<td>13:00-14:00</td>
<td>DC Steering &amp; Executive Committees Meet to Review Progress (Conference room)</td>
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What is the Dystonia Coalition?

- Infrastructure to advance dystonia therapeutics
  *not a “research study”*

- Support first began in 2009
  *started with 8 centers in North America*
  *now more than 40 centers in North America, Europe, Australia*

- Main goal is clinical trial readiness
  *not basic science*
Clinical Trial Readiness

- Engage all relevant stakeholders
  - patients in need of novel therapeutics
  - patient advocacy groups & experts who can advise on trials
  - industry experts to facilitate trials

- De-risk clinical trials
  - natural history studies to characterize patient subgroups
  - develop and test measurement endpoints for trials
  - collaboration rather than competition
  - identify novel potential agents
What Have We Done So Far?

- Delineate relevant target populations
  Definition and classification of all types of dystonia facilitated diagnostic criteria/guidelines: CD, BSP, LD
  Largest natural history population to date

- Develop and test clinical trial outcome measures
  Clinical rating scales: CD, BSP
  Patient-reported outcomes: CD, BSP, LD
  More objective assessments: digital video and wearable sensors
  Create biobank for biomarker discovery

- More than 200 publications!
Ongoing Major Clinical Projects

- Natural history project
  largest collection of patients, uniform data collection, centralized database

- Biobank project
  DNA, plasma for biomarker discovery

- Objective measures project
  digital video to diagnose and score severity of most common subtypes

- Patient-reported app
  most common subtypes of dystonia
The Dystonia Coalition: A Multicenter Network for Clinical and Translational Studies

Gamze Kilic-Berkmen¹, Laura J. Wright², Joel S. Perlmutter³, Cynthia Comella⁴, Mark Hallett⁵, Jan Teller⁶, Sarah Pirio Richardson⁷, David A. Peterson⁸, Carlos Cruchaga⁹, Codrin Lungu¹⁰ and H. A. Jinnah¹,¹¹*  

- Summary of main projects
- Summary of smaller pilot projects
- How to access data/materials
- How to get more involved
Sources of Support

- Other supporters
  - Industry
  - Professional societies
  - Patient advocacy groups
Sources of Support
Special Sources of Support!

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Pilot Projects Program
Pilot Projects Program: Description

- Purpose is to serve as seed funding to foster promising ideas into more competitive research projects
- Focus is clinical & translational research in dystonia
- Plan is for 1-2 grants each year
- Ideal budgets less than $50,000
- Partnerships with private patient groups or industry partners extend the reach of this program
Pilot Projects: Review Criteria

- Candidate: prior record, potential, and career goals
- Research plan: relevance to primary dystonia, impact, feasibility
- Project development: will the award lead bigger projects/grants?
- Mentor: experience, ability, track record
- Environment: local or collaborative
Pilot Projects: Review Committee

- Buz Jinnah (Chair)
- Committee members: Sophie Cho, Carlos Cruchaga, Joel Perlmutter, David Peterson, Sarah Pirio Richardson, Jan Teller
Pilot Projects Program: Progress

17 grants have been funded so far

Several co-funded by BEBRF, BSDPF, NSTA & CDN
Pilot Project Awards

LeDoux 2009 (USA); Gene sequence variants in primary dystonia
DeFazio 2009 (Italy); A diagnostic & rating scale for blepharospasm
Bhatia 2010 (UK); DYT6 as a window to mechanisms of primary dystonia
Roze 2010 (France); Cortical plasticity & cerebellum in focal dystonias
Klein 2011 (Germany); Endophenotypes in focal task-specific dystonias
Peterson 2011 (USA); Facial recognition software for blepharospasm
Houlden 2012 (UK); Neuropathology in DYT1 and DYT6 dystonia
Frucht 2012 (USA); Rating scale for musicians with dystonia
Eichenseer 2012 (USA); Item response theory for CD rating scale
Jinnah 2012 (USA); Fibroblast and iPS resource for dystonia
LeDoux 2013 (USA); Targeted sequencing in primary dystonias
Norris 2013 (USA); fMRI in spasmodic dysphonia and MTD
Lohmann 2013 (Germany); Genome sequencing in dystonia
Lohmann 2013 (Germany); Genetic causes in alcohol-responsive dystonia
Mink (2015) (USA); Rating scales for children with dystonia
Hammer 2020 (USA); Motor & somatosensory cortical responses in SD
Cruchaga 2021 (USA); A proteomic study of adult-onset focal dystonia
Corp 2022 2022 (USA); Theta-burst stimulation on symptoms of cervical dystonia
Martino 2022 (Canada); Moodscreen for cervical dystonia
Schreglmann 2023 (Germany); Genetic characterization of GBA in dystonia
Career Development Program
Career Development Program

- Main purpose is to help junior faculty develop careers in dystonia research
- Focus is clinical & translational research in dystonia
- Plan is for 1-2 awards each year
- Each award is about $50,000
Career Development Program: Progress

19 awards have been made so far

Several were co-funded by the DMRF or NSDA
Career Development Program Review Criteria

- Candidate: prior record, potential, and career goals
- Research plan: relevance to primary dystonia, impact, feasibility
- Career development: will the award really help?
- Mentor: experience, ability, track record
- Environment: local or collaborative
- Use of projects exploiting data/resources already collected by DC
Career Development Program
Review Committee

❖ Cindy Comella (Chair)
❖ Committee members rotate
Career Development Awards

Carbon-Corell (2009); Feinstein Institute, New York, USA
Zurowski (2010); Toronto Western Hosp, Canada
Espay 2010); Univ Cincinnati, Cincinnati, USA
Karimi (2011); Washington Univ, St Louis, USA
Kimberley (2011); Univ Minnesota, Minneapolis, USA
Berman (2012); Univ Colorado, Denver, USA
Wagle-Shukla (2012); Univ Florida, Gainesville, USA
Pirio Richardson (2013); Univ NM, Albuquerque, USA
Peterson (2013); Univ Calif San Diego, USA
Bologna (2013); Univ of Rome, Italy
Arkadir (2013); Hadassah Univ, Israel
Shaikh (2016); Case Western, Cleveland OH, USA
Udupa (2016); Toronto Western Hospital, Canada
Hammer (2016); Univ Wisconsin, Madison, USA
Rocchi (2020); Univ Rome, Italy
Parlakturk (2020); Duke Univ, Durham NC, USA
Scorr (2021); Emory Univ, USA
Hoffmeister (2022); Univ Minnesota, Minneapolis, MN
What’s in The Future?

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What’s in the Future?

- The NIH RDCRN funding cycle is 5 years. *We have been funded for 3 cycles.*

- Each funding cycle provides an opportunity to re-focus:
  1. **1st funding cycle:** natural history, biorepository, CD scales
  2. **2nd funding cycle:** more attention to BSP diagnosis and scales
  3. **3rd funding cycle:** more attention to objective measures and PROs

- What’s next?
  - Diversify sources of support
  - Pros and cons of NIH-dominated funding