

11th Annual Meeting
Dublin, Ireland
31 May 2023

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11th Annual Meeting Agenda

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

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

Dystonia Coalition: For More Info



The Dystonia Coalition: A Multicenter Network for Clinical and Translational Studies

REVIEW

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

Sources of Support



National Center
for Advancing
Translational Sciences



National Institute of
Neurological Disorders
and Stroke

- Other supporters
 - Industry*
 - Professional societies*
 - Patient advocacy groups*

Sources of Support



Special Sources of Support !



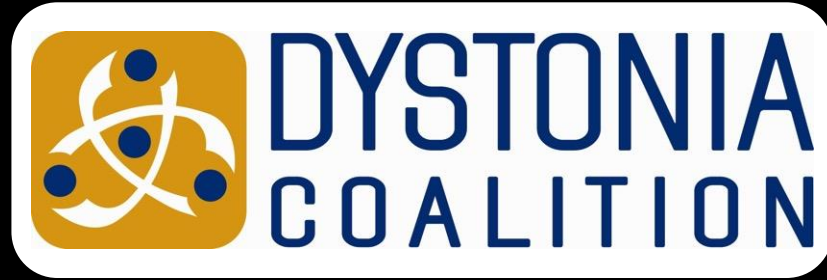
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Pilot Projects Program

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); Feinsein 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
 - 1st funding cycle: natural history, biorepository, CD scales*
 - 2nd funding cycle: more attention to BSP diagnosis and scales*
 - 3rd funding cycle: more attention to objective measures and PROs*
- What's next ?
 - diversify sources of support*
 - pros and cons of NIH-dominated funding*

