



Natural History Project

**Joel Perlmutter
Jo Wright**

Washington University in St. Louis

Sponsored by the NINDS and ORDR-NCATS at NIH, PAG, Industry, Professional Societies, and/or other sources.

Natural History Project



Dr. Joel Perlmutter

Goal: Collect clinical and exam data to better understand:

- phenotypic spectrum of all dystonias
- how symptoms change over time
- how other family members are affected
- researchers to use data for research

Natural History Recruitment Goals

(isolated dystonia): 200 cases/year

New cases

Focal
Multifocal
Segmental
Generalized
Hemidystonia

Must have started in:

Face (Craniofacial/Blepharospasm/Oromandibular)

Larynx (Laryngeal)

Limbs (Limb dystonia)

Follow-up cases

Focal
Multifocal
Segmental
Generalized
Hemidystonia

Must have started in:

Neck (Cervical Dystonia)

Face (Craniofacial/Blepharospasm/Oromandibular)

Larynx (Laryngeal)

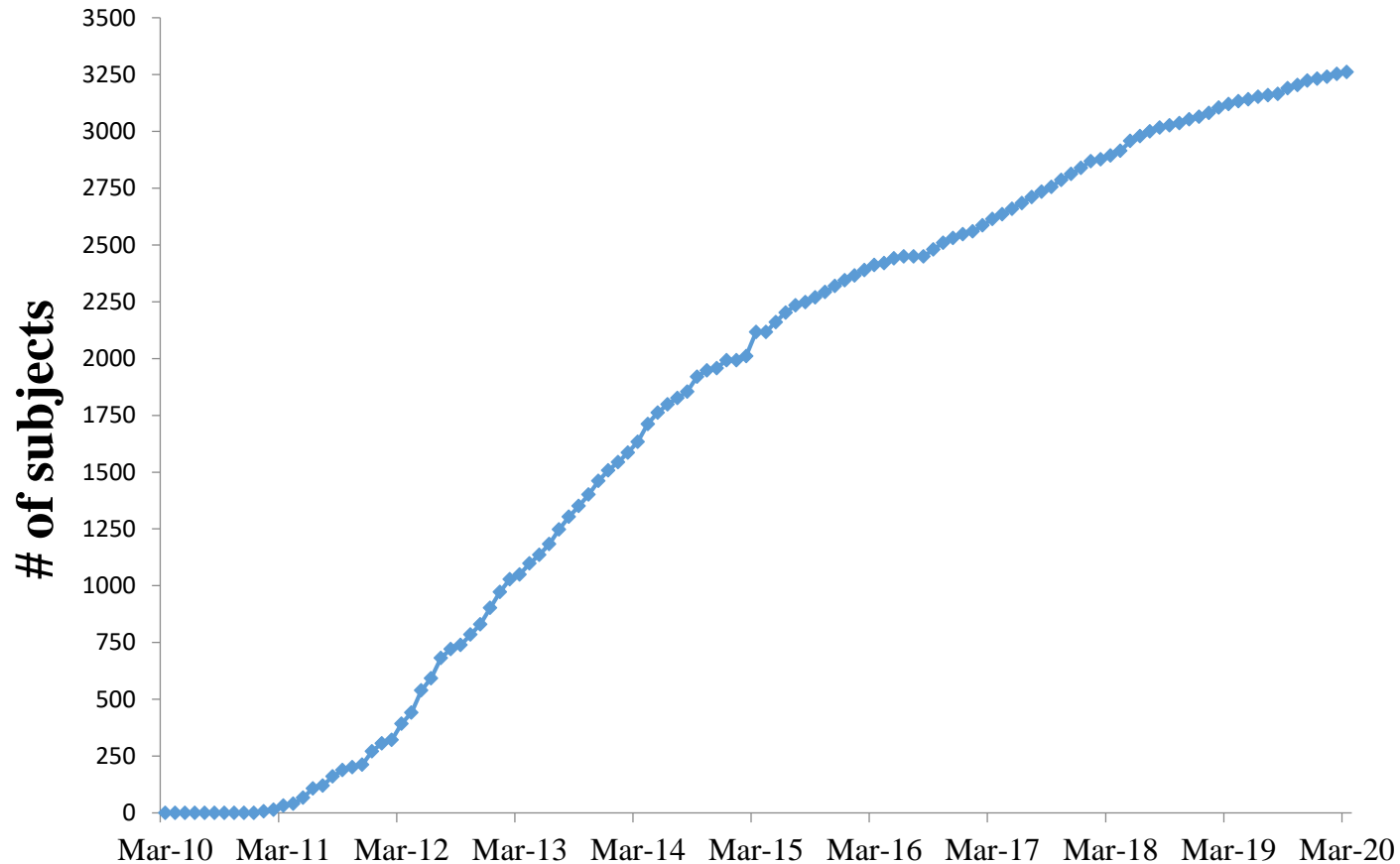
Limbs (Limb dystonia)

Natural History: To Date

- The Natural History Study began recruitment in 2010
 - Natural History/Biorepository, *2010*
 - Natural History Early Stage/Late Stage, *2016*
 - Natural History Project, *2019*
- Minimal changes to data collection and video protocols
- No change for sample Coriell collection
- Follow-up visits, determined by a sliding scale since 2016

If onset of symptoms was	Subsequent follow-up visit should be
0 to < 3 years ago	1 year from last visit
3 to < 5 years ago	2 years from last visit
5 to < 7 years ago	3 years from last visit
At least 7 years ago	4 years from last visit

Natural History Enrollment: 2010-2020



42 Clinical Sites recruited 3262 Cases, 1101 follow-ups as of 5/31/2020

Distribution of Isolated Dystonia

Types of Isolated Dystonias enrolled	
Type	Amount
Focal Dystonia	2349
Generalized Dystonia	147
Hemidystonia	14
Multifocal Dystonia	219
Segmental Dystonia	709

Distribution of Focal Dystonia Subtypes

All subjects categorized by site affected		
Body Site	Affected at onset of illness	Affected at time of study
Foot	91	181
Hand	453	786
Jaw	145	302
Larynx	359	607
Lower Face	234	657
Neck	1900	2397
Pelvis	11	46
Shoulder	154	690
Tongue	43	133
Trunk	51	218
Upper Arm	141	337
Upper Face	591	866
Upper Leg	43	91

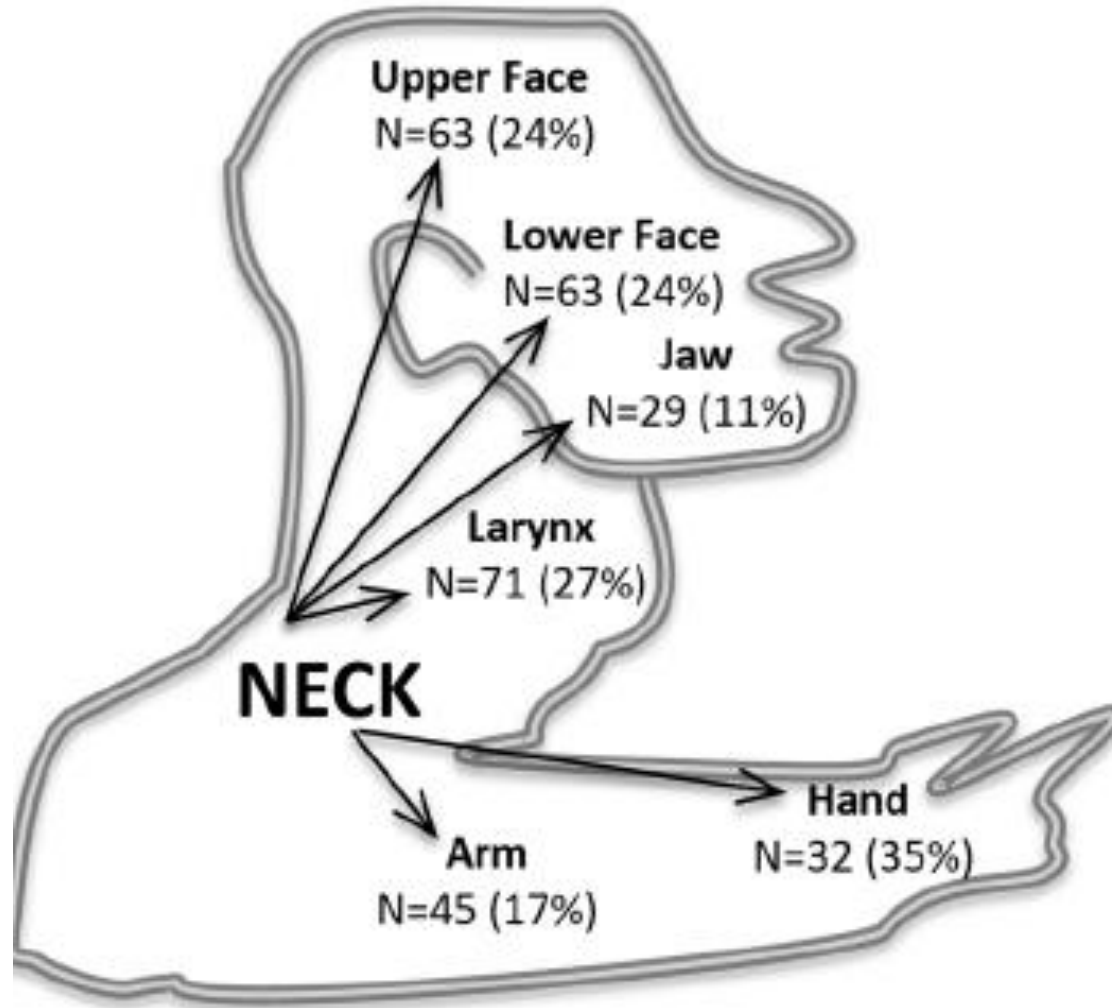
Phenomenology and Classification of Dystonia: A Consensus Update

Alberto Albanese, MD,^{1,2*} Kailash Bhatia, MD, FRCP,³ Susan B. Bressman, MD,⁴ Mahlon R. DeLong, MD,⁵ Stanley Fahn, MD,⁶
Victor S.C. Fung, PhD, FRACP,⁷ Mark Hallett, MD,⁸ Joseph Jankovic, MD,⁹ Hyder A. Jinnah, PhD,¹⁰ Christine Klein, MD,¹¹
Anthony E. Lang, MD,¹² Jonathan W. Mink, MD, PhD,¹³ Jan K. Teller, PhD¹⁴

Movement Disorders 2013

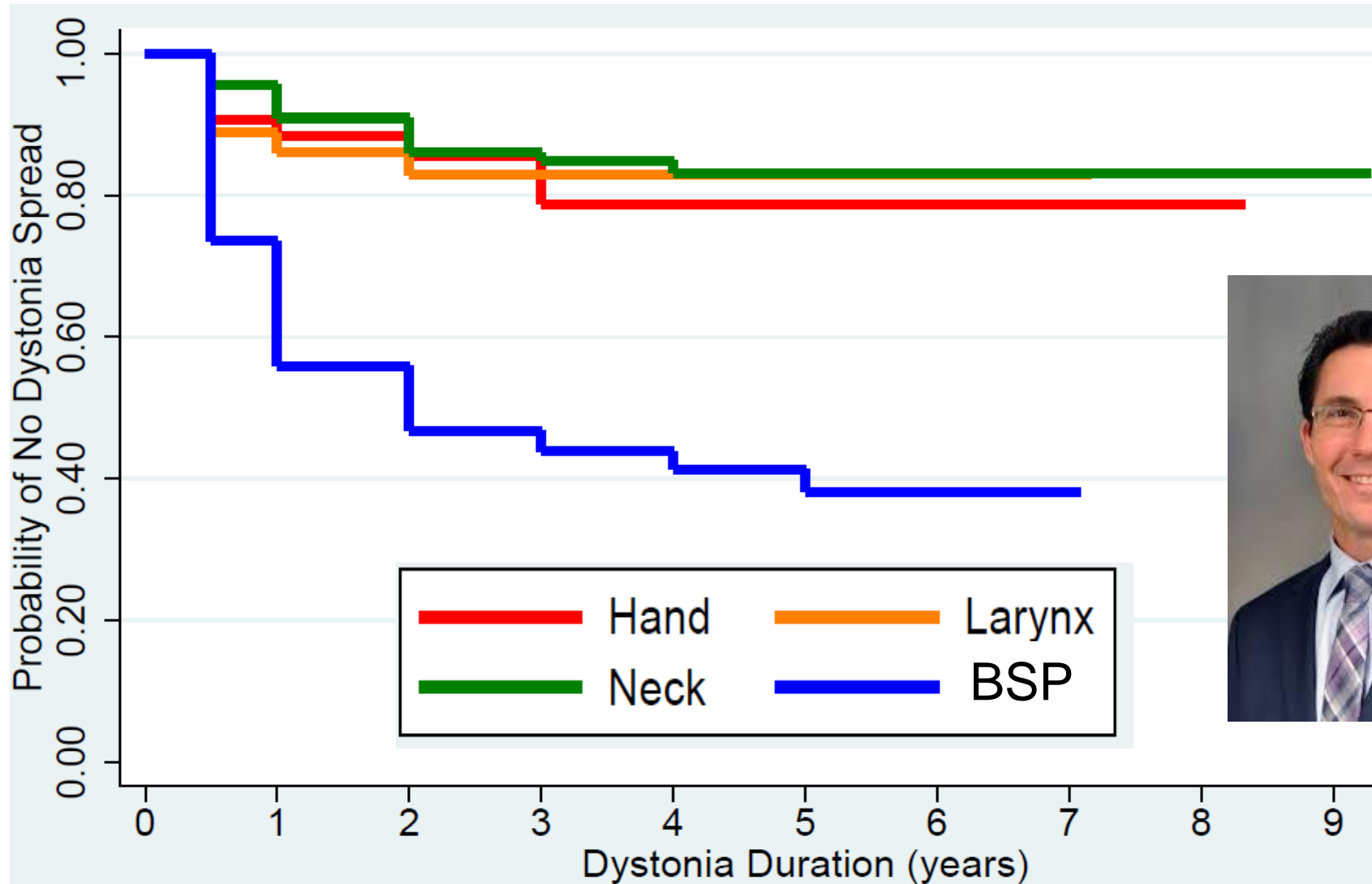
Spread of Cervical Dystonia

FNO-SS (N=264, 17.9%)



Norris SA et al, Mov Disord 2017

DC: Natural History



Berman BD et al, JNNP 2020

Cervical dystonia severity tools

- **Organizing center**
Cynthia Comella, MD
- **Revise current TWSTRS**
include non-motor elements
validate
training tape



Comella C et al, Mov Dis Clin Pract, 2015
Comella C et al, Mov Disord, 2016

Natural History Available Data -1

Completed by Neurologist

- Global Dystonia Rating Scale (GDRS)
- Burke-Fahn Marsden Dystonia Scale (BFM)

Completed by participant

- Short Form Health Survey 36 (SF-36)
- Beck Depression Inventory II (BDI-II)
- Hospital Anxiety and Depression Scale (HADS)
- Liebowitz Social Anxiety Scale (LSAS)
- Patient Health Questionnaire 9 (PHQ-9)

Natural History Available Data -2

Completed by Coordinator

➤ DCP Data Collection Form

demographic, medical history, medications, exam, family history

➤ Neurological Exam Video

over 3,000 videos available

➤ Biospecimens

over 3,000 DNA samples available

Data Accessibility - Who

- Enrolling Sites
 - Access to site data and videos
- New Research Projects
 - IRB approval
 - Data Access Agreement form
 - Executive Committee approval
 - Data
 - Videos (restricted access)
 - Biospecimens
- REDCap Data Interface (Pentaho)
Publically available non-PHI data

Data Accessibility - Where

- Washington University in St. Louis
 - REDCap
 - Video Repository
 - REDCap Data Interface (Pentaho)
 - Publically available de-identified data
- NINDS Coriell Institute for Medical Research, *New Jersey*
 - DNA samples
 - De-identified data
- Data Management and Coordinating Center (DMCC), *Cincinnati Children's Hospital Medical Center*
 - De-identified data

Data Accessibility – Video Repository

Restricted access



Washington University in St. Louis • School of Medicine



DYSTONIA
COALITION

Search by site

Search by diagnosis

Search by project

Search by Comments

Site Search

Data Sharing/Access

Public Access: REDCap Data Interface (Pentaho)

http://clinportalquery.wustl.edu/pentaho/Home?userid=DYSTONIA_OPENI&password=DYSTONIA

(Firefox or Chrome, not IE)

Full Data or Video Access

- Application
- Data access agreement
 - no secondary release*
 - return results/acknowledgment*
- IRB approval
- Executive Committee approval
- Only project specific data release
- Video: only streaming, not downloading

Investigator Responsibilities

- Use data for proposed studies only
- No secondary release
- No attempt to identify participants
- Adequate staff training
- Share findings with Dystonia Coalition and publish data
- Acknowledge source

Data Utilization

- Use data for proposed studies only
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- No attempt to identify participants
- Adequate staff training
- Share findings with Dystonia Coalition and publish data
- Acknowledge source

Q & A

Natural History (NH) Project

Project Management Center: Washington University in St. Louis

Project Leader: Joel Perlmutter, MD (perlmutterjoel@wustl.edu)

Data Manager: Jo Wright (laurajwright@wustl.edu)