INTAKE FORM

Your patient wishes to participate in the Dystonia Brain Collective and donate their brain. The Collective is comprised of dystonia patient advocacy organization working in partnership with the Harvard Brain Tissue Resource Center (HBTRC) at McLean Hospital. The HBTRC can be contacted at 1.800.BRAIN BANK or at www.brainbank.mclean.org

HBTRC will contact you following tissue donation. Your completing this form will help us to provide important information to researchers seeking dystonia tissue for their work.

PLEASE KEEP FORM AS PART OF PATIENT MEDICAL RECORD.

Thank you for your assistance.

PART I: PARTICIPANT DETAILS (Can be completed by participant)

Subject Name: Last Name: _______________ First Name: _______________ Middle Initial: _____

Date of Birth: Month: _____ Day: ______ Year: _______ **Age in years:** _____

Gender: ☐ Female ☐ Male

Race: Check all that apply
- ☐ American Indian/Alaska Native
- ☐ Asian
- ☐ Black/African American
- ☐ Native Hawaiian/ Other Pacific Islander
- ☐ Not Reported
- ☐ Unknown
- ☐ White/Caucasian
- ☐ Other ______________________

Ethnicity: Check all that apply
- ☐ Hispanic or Latino
- ☐ Not Hispanic or Latino
- ☐ Not Reported
- ☐ Unknown
- ☐ Ashkenazi Jewish
- ☐ Mennonite
- ☐ Other ______________________

Participant address: Street: ____

City: ______________________________ State/province: ______________________________

Country: ______________________________ Postal code: ______________________________

Phone number: ______________________________ Email: ______________________________

All of the rest of the form should be completed by your treating physician.

PART II DYSTONIA HISTORY AND OTHER MEDICAL CONDITIONS

Age at onset of dystonia: _________

Best Clinical Diagnosis for dystonia:

☐ **Primary dystonia:**

☐ **Focal dystonias**:

☐ Cranial dystonia, blepharospasm only
☐ Cranial dystonia, upper and lower facial involvement
☐ Cranial dystonia, oromandibular dystonia
☐ Cranial dystonia, lingual
☐ Laryngeal dystonia, adductor type
☐ Laryngeal dystonia, abductor type
☐ Laryngeal dystonia, mixed
☐ Cervical dystonia
☐ Limb dystonia: Upper extremity dystonia
☐ Limb dystonia: Lower extremity dystonia

☐ **Segmental dystonia**

☐ **Hemi-dystonia**

☐ Left

☐ Right

☐ **Multi-focal dystonia**

☐ **Generalized dystonia**

☐ **Other dystonia syndrome:** ____

☐ **Non-dystonia syndrome:** ____

Received any surgical treatment for dystonia? ❑ Yes ❑ No ❑ Unknown

If yes, what type?

❑ DBS Date:__________ Target: ❑ Gpi ❑ Subthalamus ❑ Thalamus ❑ Other________

❑ Ablation Date:__________

Target: ❑ Pallidotomy ❑ Subthalamotomy ❑ Thalamotomy ❑ Other__________

❑ Selective denervation for cervical dystonia: Date: ____________________

❑ Myectomy for blepharospasm or spasmodic dysphonia: Date: ____________________

❑ Laryngeal nerve surgery for spasmodic dysphonia: Date: ____________________

❑ Other (specify date and details): ____

Documented causal gene:

☐ Yes (If yes is selected, please choose from the list below)

- ☐ DYT1 (GAG deletion)
- ☐ DYT1 (other TOR1A mutation, torsin A)
- ☐ DYT3 – lubag (TAF1, TATA-binding protein associated factor 1)
- ☐ DYT5a/DYT14 (GCH1, GTP cyclohydrolase)
- ☐ DYT5b (TH, tyrosine hydroxylase)
- ☐ DYT6 (*THAP1*, thanatos associated protein)
- ☐ DYT8 (*MFRP1*,Myofibrillo-genesis regulator protein-1)
- ☐ DYT11(*SCGE*, epsilon-sarcoglycan)
- ☐ DYT12 (*ATP1A*3, Na/K ATPase α3)
- ☐ DYT16 (PRKRA, protein kinase, interferon-inducible double stranded RNA dependent activator)
- ☐ DYT18 (SLC2A1, glut1 deficiency syndrome)
- ☐ Other: ___________________________

☐ Unknown (no screening done)

History of Dystonia Onset:

* If bilateral onset, please select left and right for that body part.
** In all body regions where there is currently evidence of dystonia or where dystonia has been in the past, please indicate age of onset for those body regions.

Body Region	Left	Right	Age of onset	Initial site of onset? Yes/No
Upper face				
Lower face				
Shoulder				
Upper arm				
Hand				
Upper leg				
Foot				
Tongue				
Jaw				
Neck				
Trunk				
Pelvis				
Larynx				

Other characteristics of dystonia:

1. Task-specific
   ☐ Yes (If yes is selected, please choose from the list below)
      ☐ writer's cramp
      ☐ typist's cramp
      ☐ musician's cramp
      ☐ sports related cramp
      ☐ speech related dystonia (spasmodic dysphonia)
      ☐ other______
   ☐ No

2. Did dystonia have abrupt onset (less than 1 week):
   ☐ Yes
   ☐ No
   ☐ Unknown

3. Was dystonia fixed at onset?
   ☐ Yes
   ☐ No

4. Is there any sensory trick effective?
   ☐ Yes If yes, please describe ____
   ☐ No

5. Has alcohol relieved the dystonia?
   ☐ Yes
   ☐ No
   ☐ Unknown

Medical History: Does the subject have a history of any of the following? (check all that apply):

Stroke ❑ Yes ❑ No

Depression ❑ Yes ❑ No

Anxiety-related disorders ❑ Yes ❑ No

Serious trauma or surgery to head ❑ Yes ❑ No (if yes, give year__________)

Serious trauma or surgery to face ❑ Yes ❑ No (if yes, give year__________)

Serious trauma or surgery to neck ❑ Yes ❑ No (if yes, give year__________)

Diagnosis of vocal cord paralysis ❑ Yes ❑ No (if yes, give year__________)

Serious trauma or surgery to limb(s) ❑ Yes ❑ No (if yes, give year__________)

URI just prior (within 2 weeks) to onset of dystonia ❑ Yes ❑ No

Tremor ❑ Yes ❑ No

Parkinson Disease or parkinsonism ❑ Yes ❑ No

Tics or Tourette Syndrome ❑ Yes ❑ No

Other psychiatric condition : ❑ Yes:______________________________ ❑ No

Medications (Name only) currently taken (Please remember to record botulinum toxin treatment if applicable & record when last dose was received. :

PART III: EXAMINATION: Please indicate which body regions are affected by dystonia and laterality if applicable. If tremor is present for any body region, please check 'yes' under "presence of tremor" and indicate whether the tremor is regular, irregular/jerky, or both. If both, on paper form check both boxes and in electronic data entry, select "both". If subject has tremor, but no dystonia in a particular body region, only check the boxes pertaining to tremor. Do not check the left or right boxes which pertain only to the subject's dystonia.

	Dystonia		Tremor		
Body Region	**Left**	**Right**	**Presence of Tremor Yes/No**	**Regular**	**Irregular/jerky**
Upper face					
Lower face					
Shoulder					
Upper arm					
Hand					
Upper leg					
Foot					
Tongue					
Jaw					
Neck					
Trunk					
Pelvis					
Larynx (choose all that apply)	☐ Adductor ☐ Abductor ☐ Muscular tension dysphonia ☐ Tremor ☐ Other dysphonia:		Confirmed by nasopharyngoscopy? ☐ Yes ☐ No		

If any of the tremor boxes are checked, then

a) **is this patient's dystonia dominated by tremor more than tonic or twisting movements?**
   - ☐ Yes
   - ☐ No

b) **did tremor precede onset of dystonia?**
   - ☐ Yes If yes, age of onset of tremor: ________
   - ☐ No

Other Medical Conditions:

- Myoclonus:
  - ☐ Yes
  - ☐ No

- Parkinsonism:
  - ☐ Yes
  - ☐ No

PART IV: FAMILY HISTORY

- **Ethnic category of grandparents: (ancestors' countries of origin):**

Maternal Grandmother	☐ American Indian/Alaska Native ☐ Asian ☐ Black/African American ☐ Native Hawaiian/ Other Pacific Islander ☐ Not Reported ☐ Unknown ☐ White/Caucasian ☐ Other ___________	Country of Origin
Paternal Grandmother	☐ American Indian/Alaska Native ☐ Asian ☐ Black/African American ☐ Native Hawaiian/ Other Pacific Islander ☐ Not Reported ☐ Unknown ☐ White/Caucasian ☐ Other ___________	Country of Origin
Maternal Grandfather	☐ American Indian/Alaska Native ☐ Asian ☐ Black/African American ☐ Native Hawaiian/ Other Pacific Islander ☐ Not Reported ☐ Unknown ☐ White/Caucasian ☐ Other ___________	Country of Origin
Paternal Grandfather	☐ American Indian/Alaska Native ☐ Asian ☐ Black/African American ☐ Native Hawaiian/ Other Pacific Islander ☐ Not Reported ☐ Unknown ☐ White/Caucasian ☐ Other ___________	Country of Origin

- **Dystonia present in any other family members?**
  - ❑ Yes (Complete two tables below.)
  - ❑ No (Skip two tables below.) ❑ Unknown (Skip two tables below.)

Family members	Age of onset	Type of dystonia	Observation or diagnosed

Mother			
Father			
Brother (s)			
Sister (s)			
Half-brother (Maternal)			
Half-brother (Paternal)			
Half-sister (Maternal)			
Half-sister (Paternal)			
Son (s)			
Daughter (s)			
Other			

Family members	Numbers affected	Numbers unaffected	Unknown
Brother (s)			
Sister (s)			
Son(s)			
Daughter (s)			

- **Documented causal gene present in family members with dystonia?**

  ❑ Yes ❑ Unknown (No screening done)

  If dystonic syndrome is present, please specify ____________________

  If yes, please select from the list below:

  ☐ DYT1 (GAG deletion)
  ☐ DYT1 (other TOR1A mutation, torsin A)
  ☐ DYT3 – lubag (TAF1, TATA-binding protein associated factor 1)
  ☐ DYT5a/DYT14 (GCH1,GTP cyclohydrolase)
  ☐ DYT5b (TH, tyrosine hydroxylase)
  ☐ DYT6 (*THAP1*, thanatos associated protein)
  ☐ DYT8 (*MFRP1*,Myofibrillo-genesis regulator protein-1)
  ☐ DYT11(*SCGE*, epsilon-sarcoglycan)

☐ DYT12 (*ATP1A*3, Na/K ATPase α3)
☐ DYT16 (PRKRA, protein kinase, interferon-inducible double stranded RNA dependent activator)
☐ DYT18 (SLC2A1, glut1 deficiency syndrome)
☐ Other: ______________________________________

➢ **Does any family member have the diseases listed below?**
**If yes, please mark which family members are affected.

		Mother	Father	Brother(s)	Sister(s)	Sons(s)	Daughter(s)	Other
Parkinson disease or Parkinsonism	☐ Yes ☐ No ☐ Unknown							
Tremor	☐ Yes ☐ No ☐ Unknown							
Myoclonus	☐ Yes ☐ No ☐ Unknown							
Other	Please specify:							

➢ **Family consanguinity:**
❑ Yes ❑ No ❑ Unknown

The Dystonia Brain Collective Organizations wish to acknowledge and thank
the Dystonia Coalition for permitting the use of their form, amended for use with the Brain Collective.