

THE  
2ND DECADE



MYOTONIC  
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## 2017 MDF Annual Conference

Sept. 8–9, 2017 San Francisco



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Care and a Cure

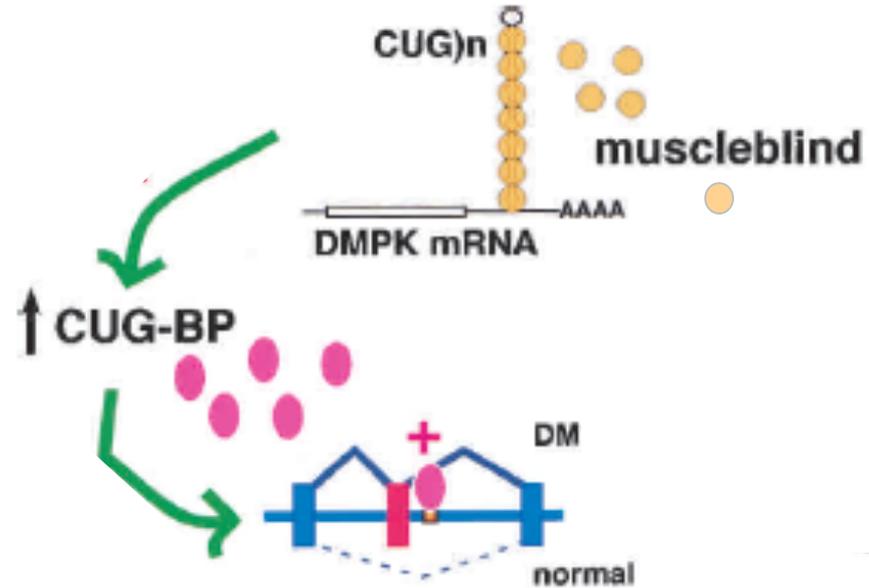
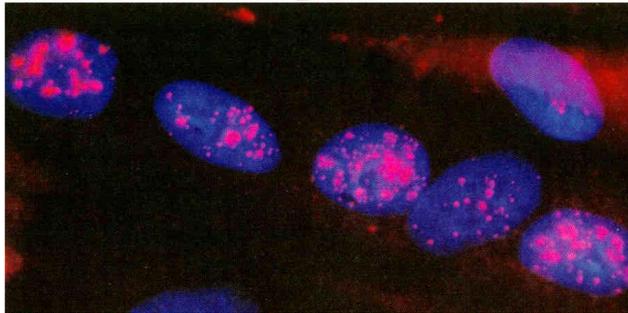
# DM AND THE CENTRAL NERVOUS SYSTEM

John W. Day, MD, PhD



# Understanding DM Effects on Muscle

... CTGCTGCTGCTG ...



*Ranum, et al., Ann Rev Neurosci, 2006*



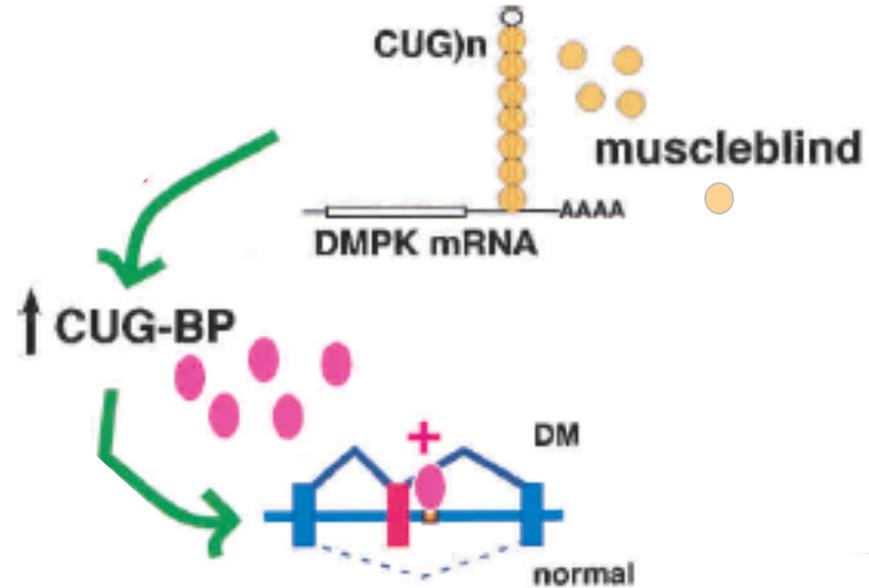
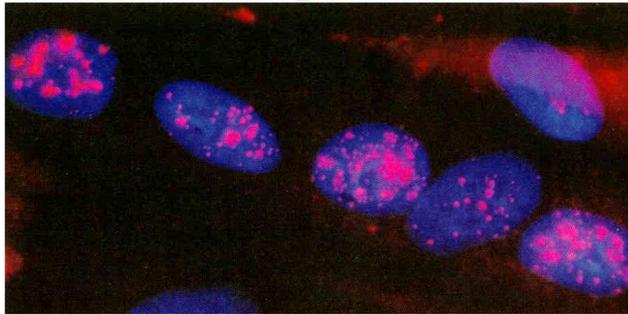
... CCTGCCTGCCTG ...



**Changes in Proteins,  
Changes in Function**

# Helps Explain DM Effects on CNS

... CTGCTGCTGCTG ...



*Ranum, et al., Ann Rev Neurosci, 2006*

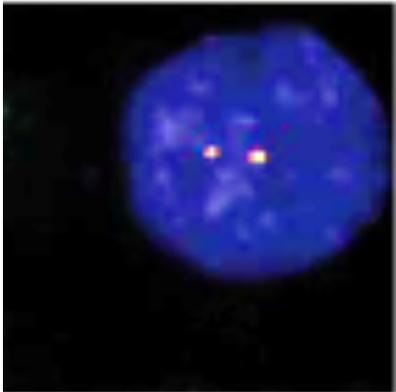
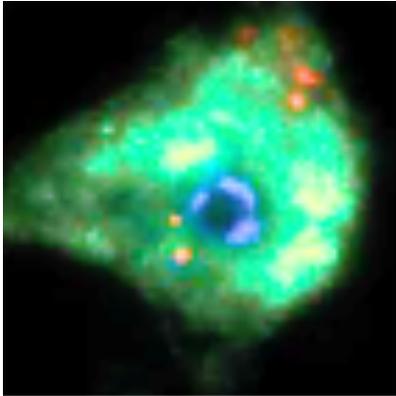


... CCTGCCTGCCTG ...



**Changes in Proteins,  
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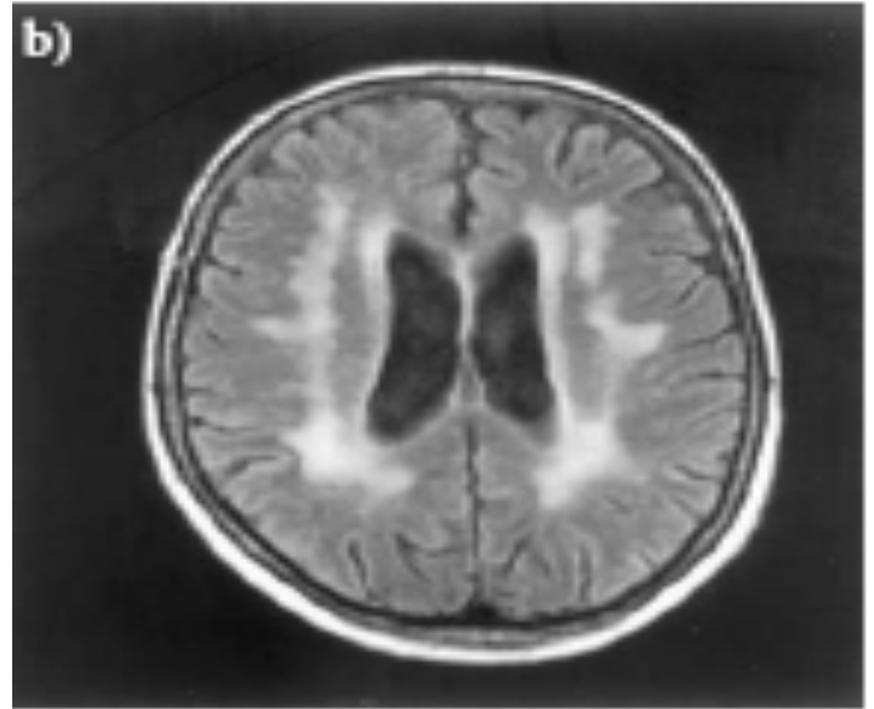
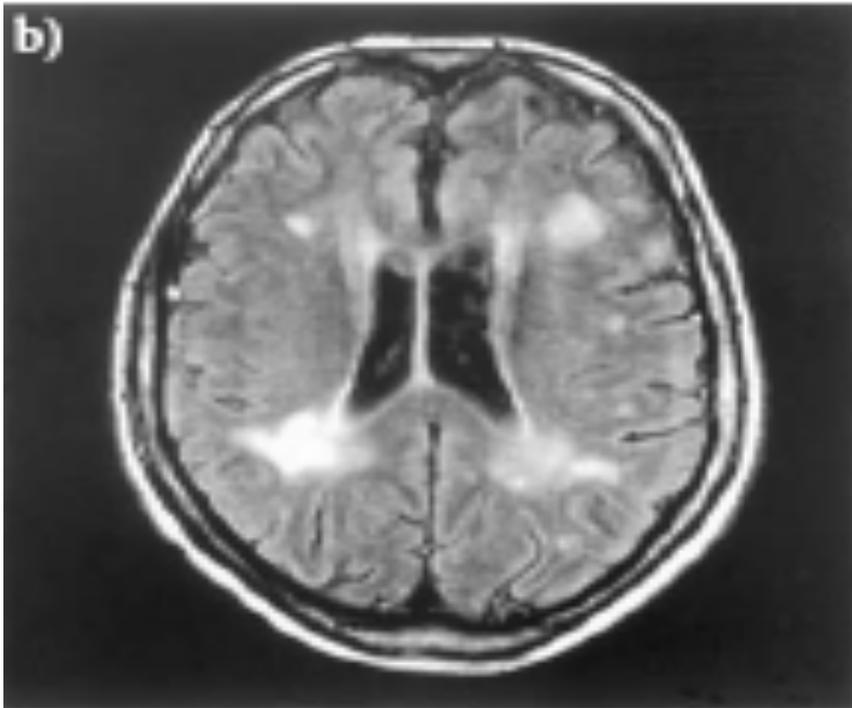
# Helps Explain DM Effects on CNS



**Changes in Proteins,  
Changes in Function**

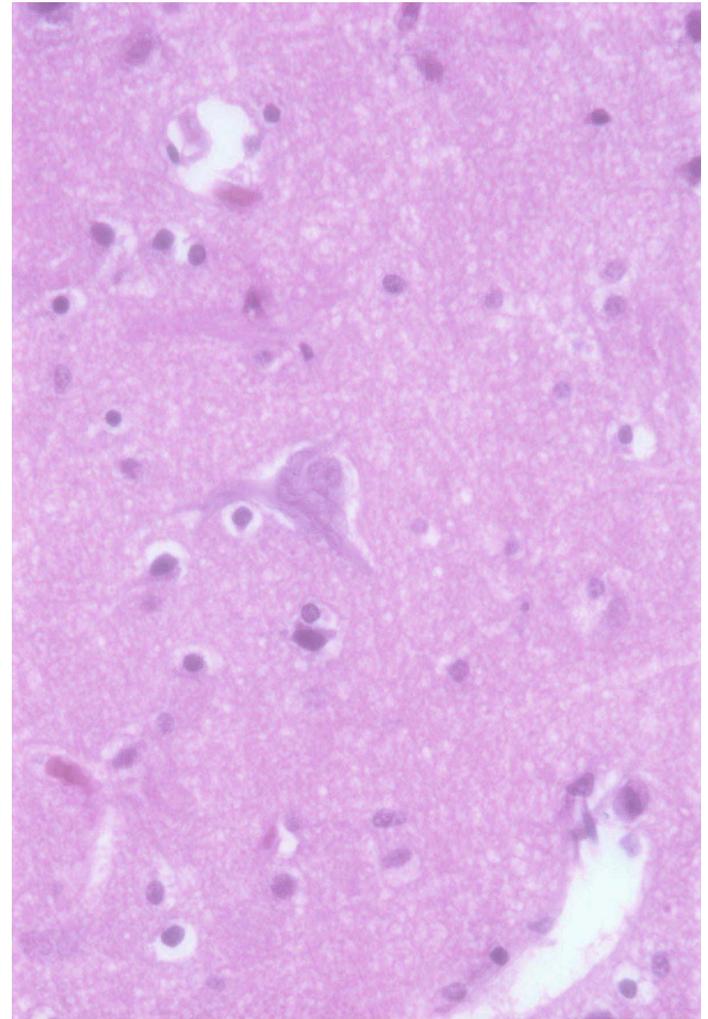
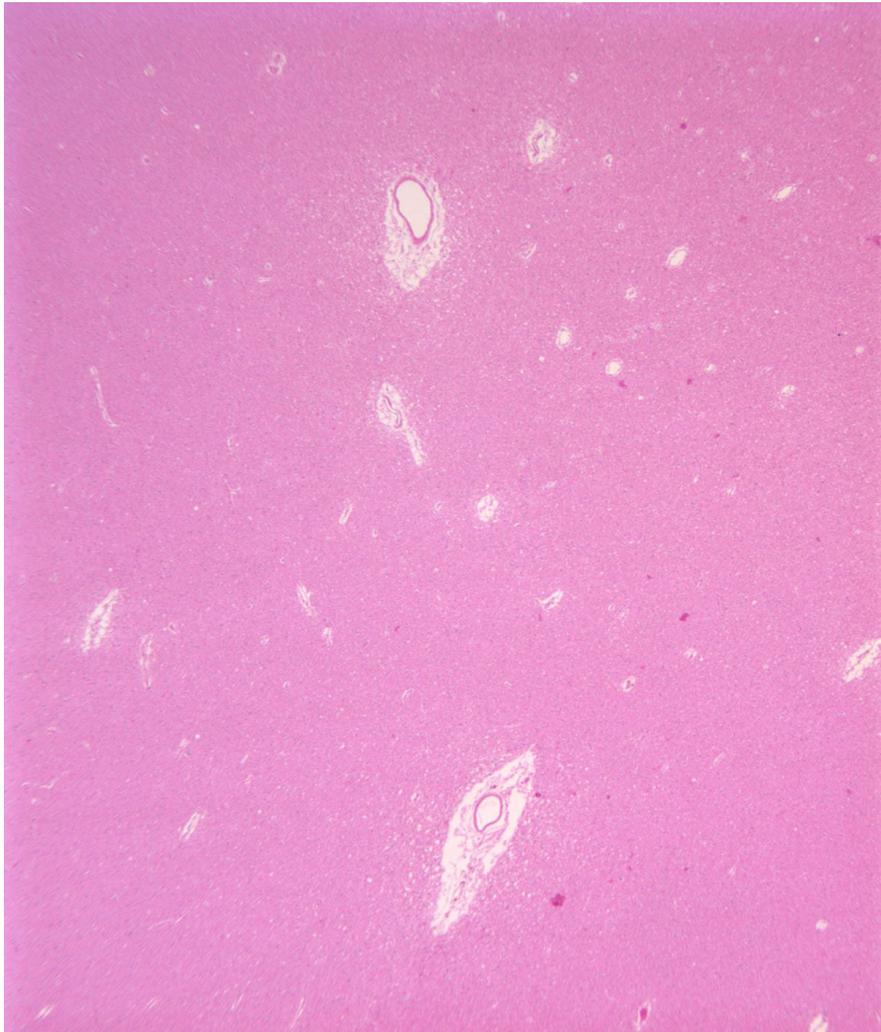
*Jiang, et al., 2004*

# MRI Changes

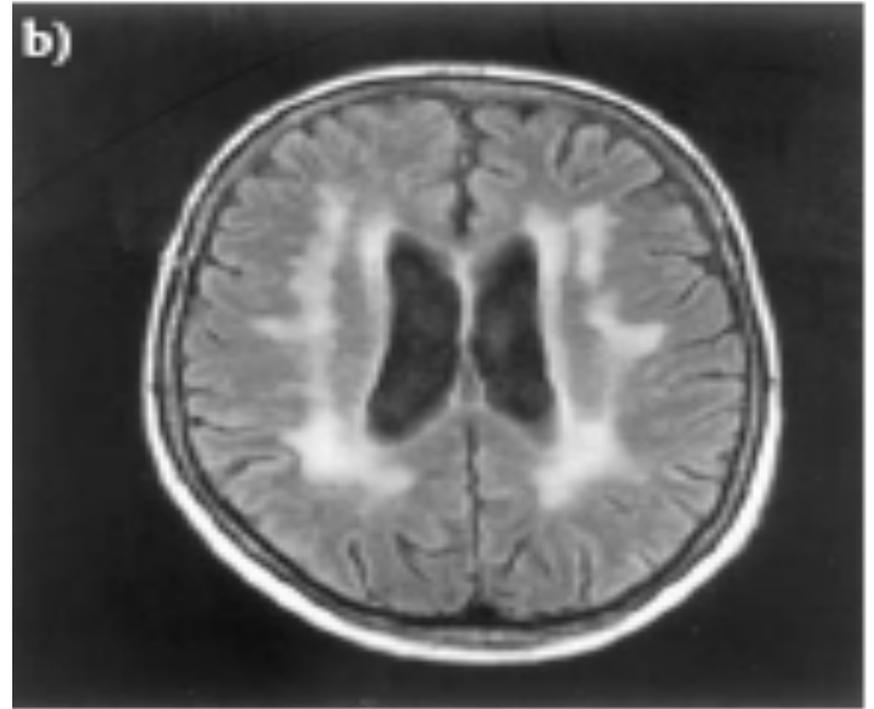
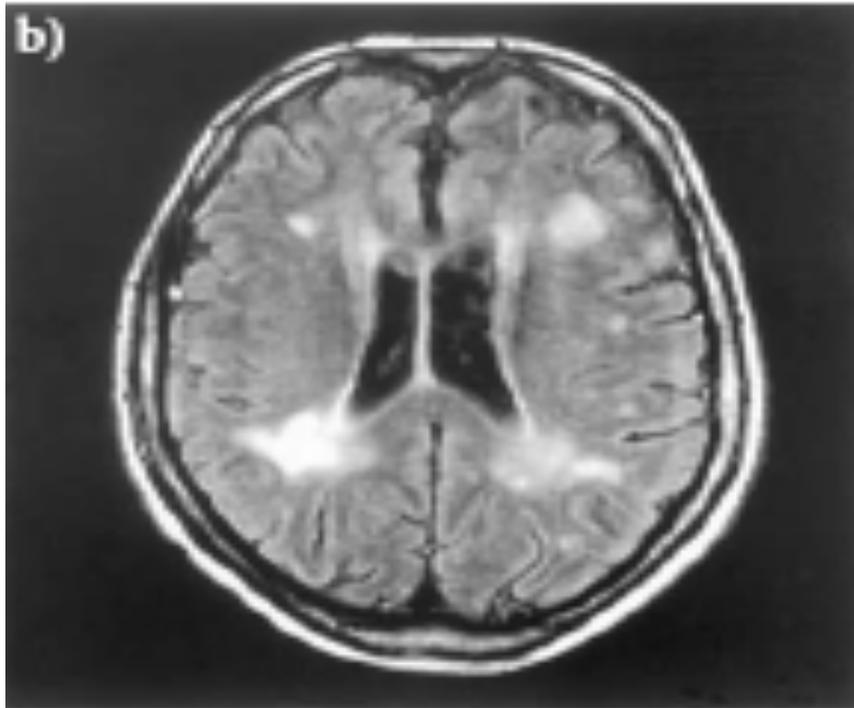


*Kornblum, et al, J. Neurol, 2004*

# MRI Changes Do Not Predict Microscopic Changes

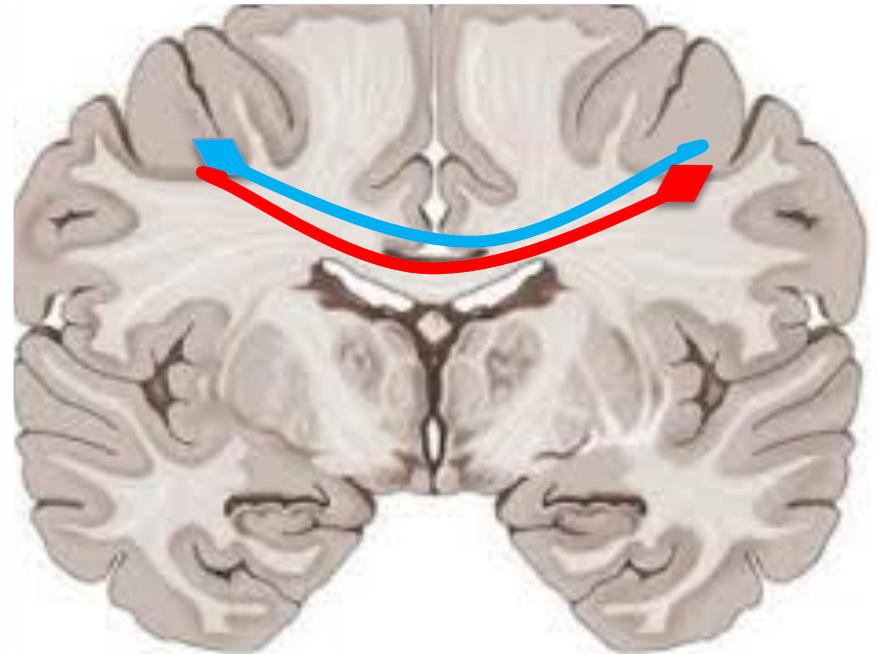
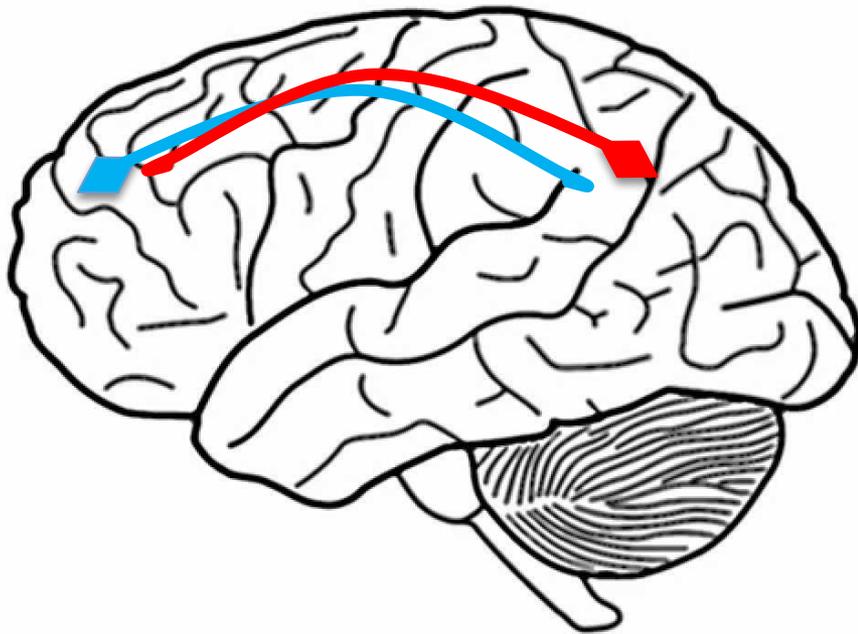


# But Maybe those MRI Changes



*Kornblum, et al, J. Neurol, 2004*

# Underlie Changes in Connectivity?



# And Account for Changes in Processing?

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Wechsler Test of Adult Reading

Executive Function Verbal Fluency

Conceptual reasoning

Stroop Color Test

Trail Tests

Visual Puzzles

Purdue Pegboard Test

Frontal Systems Behavior Scale

Depression Inventory

# Do test changes relate to patient descriptions?

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- Brain Fog
- Good Days and Bad Days
- Low Energy
- Fatigue
- Sleepiness
- Inability to focus
- Trouble connecting

# But What Accounts for Patient Experience?

**Molecular?**  
**Changes**



**MRI**  
**Changes**



**Psychology**  
**Test Changes**



**Patient**  
**Experience**

**Plus what effect on brain is caused by DM changes on muscle? Heart? Vision? GI?**

# Patients and Investigators Need to Work Together to Determine

What symptoms are most important

How do we quantify those symptoms

How do we measure those symptoms

How do we improve those symptoms

# What can you do?

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- MDF Registry
- Rochester DM Registry
- Participate in Research
- Donate specimens
- Spread the word