

# Brain and Behavior in Dystrophinopathy and BELS

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**Parent** **Project** **Muscular** **Dystrophy**  
JOIN THE FIGHT.  
END DUCHENNE.

# Why Brain and Behavior Matter

## Psychosocial Management of the Patient With Duchenne Muscular Dystrophy

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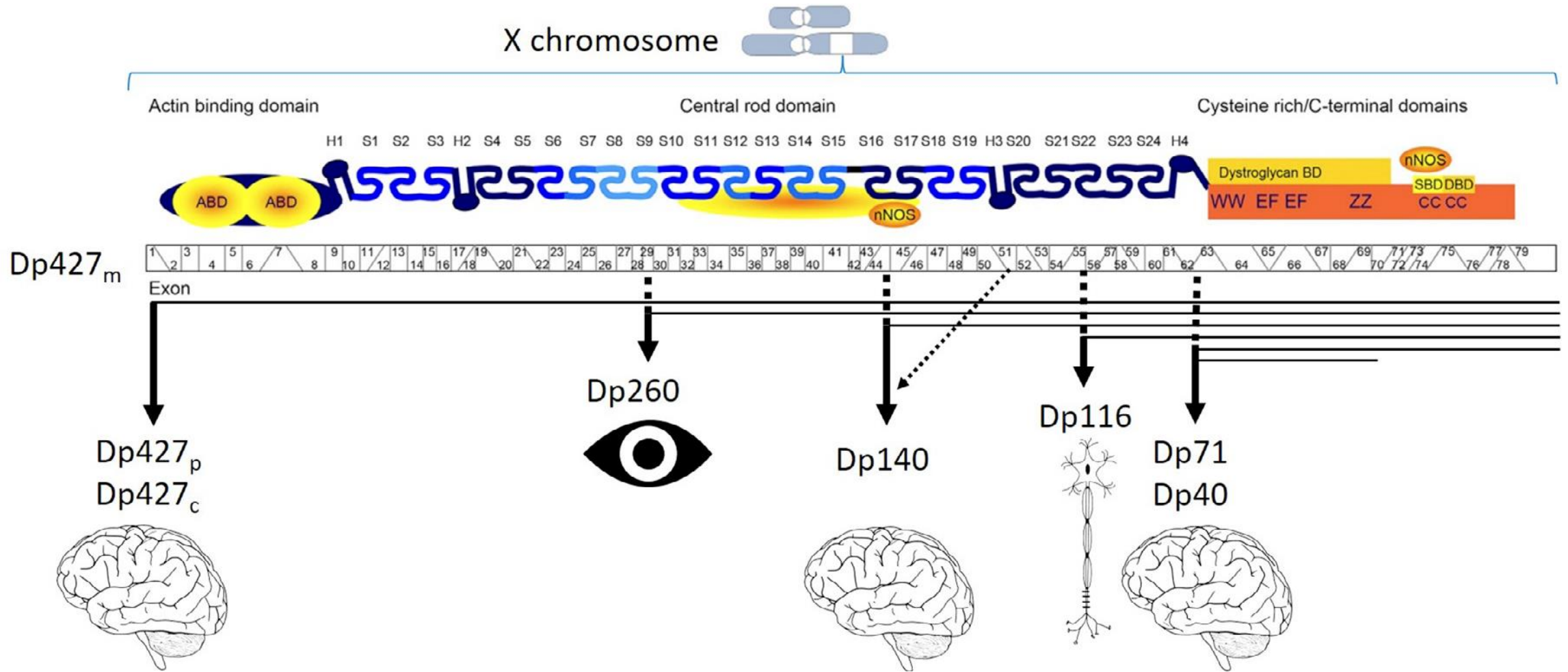
## Diagnosis and management of Duchenne muscular dystrophy, part 3: primary care, emergency management, psychosocial care, and transitions of care across the lifespan

*David J Birnkrant, Katharine Bushby, Carla M Bann, Susan D Apkon, Angela Blackwell, Mary K Colvin, Linda Cripe, Adrienne R Herron, Annie Kennedy, Kathi Kinnett, James Naprawa, Garey Noritz, James Poysky, Natalie Street, Christina J Trout, David R Weber, Leanne M Ward, for the DMD Care Considerations Working Group\**

- Many children with Duchenne or Becker have learning, emotional, and social challenges that deserve support
- Screening and check-ins help identify these needs early, so support can start sooner
- As children with Duchenne and Becker live longer, this understanding improves quality of life for them and their families

# How Dystrophinopathy Can Affect the Brain

# The same gene shapes both muscles and the brain



## Understanding the Neurodevelopmental Disorder Due to Dystrophinopathy



### 1. GENETIC ALTERATION

Pathogenic variants in the *DMD* gene lead to absent or reduced dystrophin.

### KEY POINT

Dystrophin is expressed in the brain and plays a critical role in neural development and function.



### 2. ALTERED BRAIN DEVELOPMENT AND FUNCTION

Changes in neural connectivity, synaptic signaling, and brain networks impact multiple neurodevelopmental domains.

### SUPPORTING EVIDENCE

Neuroimaging and neuropathological studies demonstrate structural and functional differences in individuals with dystrophinopathy.



### 3. NEURODEVELOPMENTAL VULNERABILITIES

Vulnerabilities emerge in key domains that affect learning, behavior, emotion, and social functioning.

### CORE DOMAINS IMPACTED

- Social communication
- Attention and executive function
- Learning and academic skills
- Emotional regulation
- Anxiety/OCD vulnerability
- Mood regulation



### 4. FUNCTIONAL IMPACTS

Challenges become apparent in daily life across home, school, and social settings, affecting independence, self-esteem, and quality of life.

### EXAMPLES

- Academic underachievement
- Social difficulties
- Emotional distress
- Behavioral challenges
- Family stress



### 5. CLINICAL PRESENTATIONS

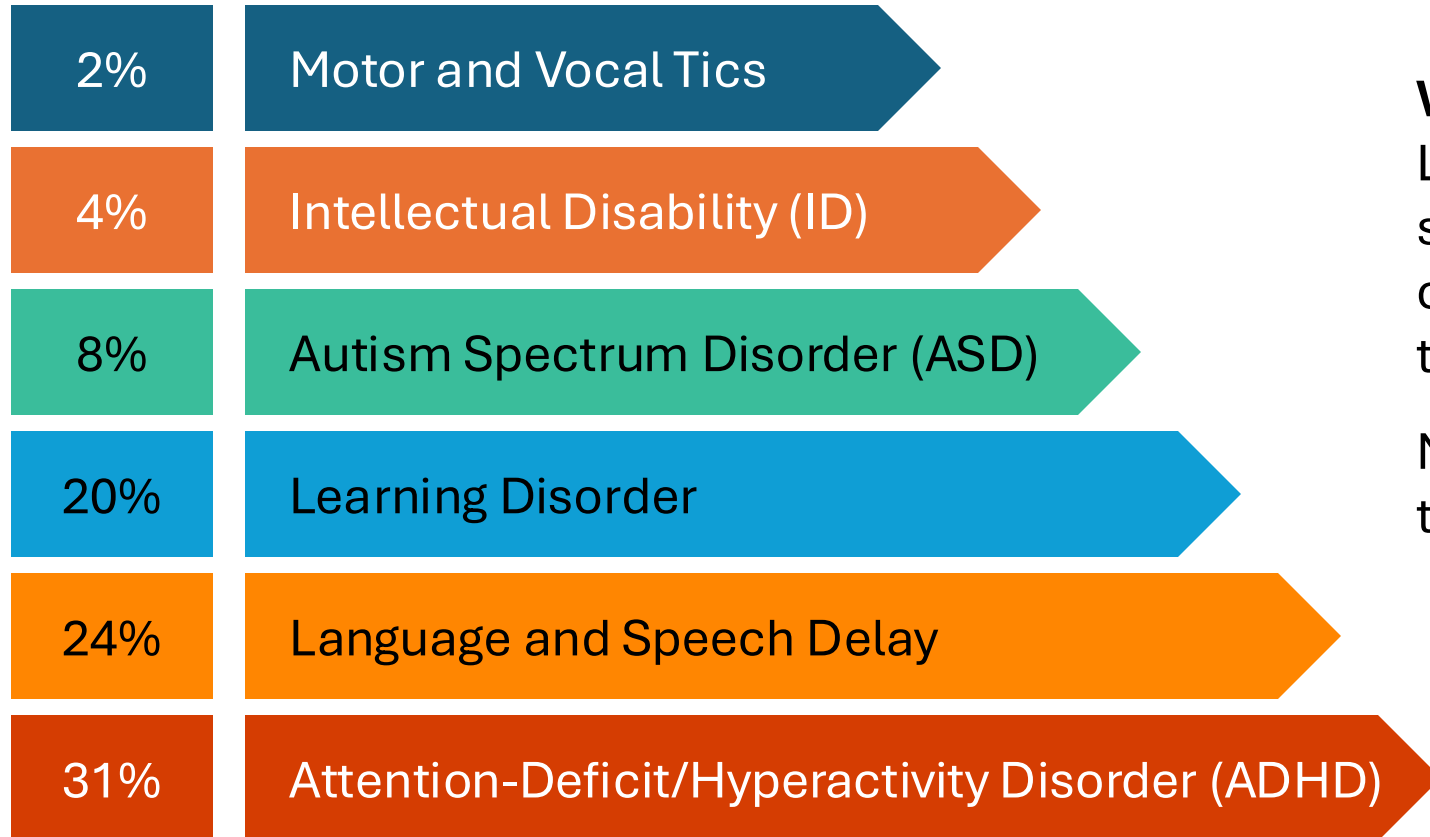
(Examples, not all will be present)  
May meet criteria for ADHD, ASD, learning disorders, anxiety/OCD disorders, mood disorders, etc.

### IMPLICATION FOR CARE

Screen broadly, monitor over time, and treat proactively to optimize development and well-being.



# Clinical Signs of Brain Involvement



## **What this means for families:**

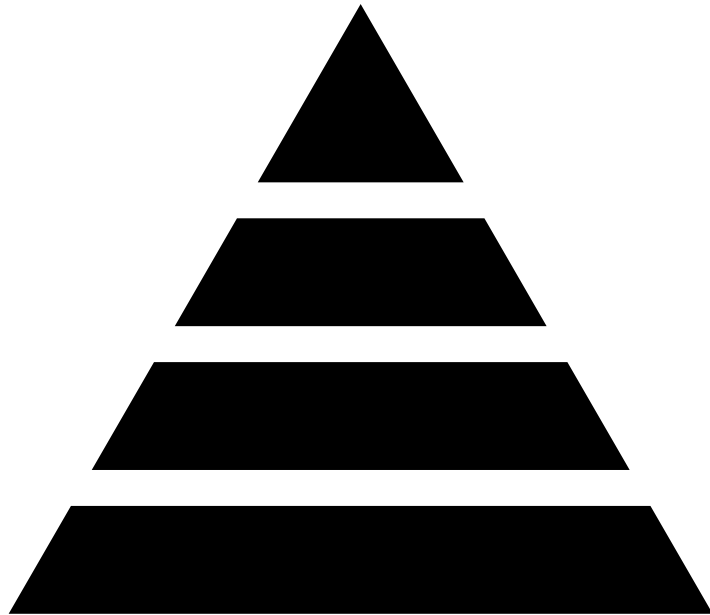
Learning, attention, language, and social differences are more common in Duchenne and Becker than in other children.

Noticing them early opens the door to support.

# Common Parent/Caregiver Concerns

	Caregiver Response (% , N = 320)
<b>Anxiety</b>	50
<b>ADHD</b>	32
<b>OCD</b>	26
<b>Depression</b>	22
<b>Autism Spectrum</b>	21
<b>Sensory Processing</b>	19
<b>Oppositional Behaviors</b>	10

# Foundation for a Developmental Hierarchy



Language Processing

Motor Skills

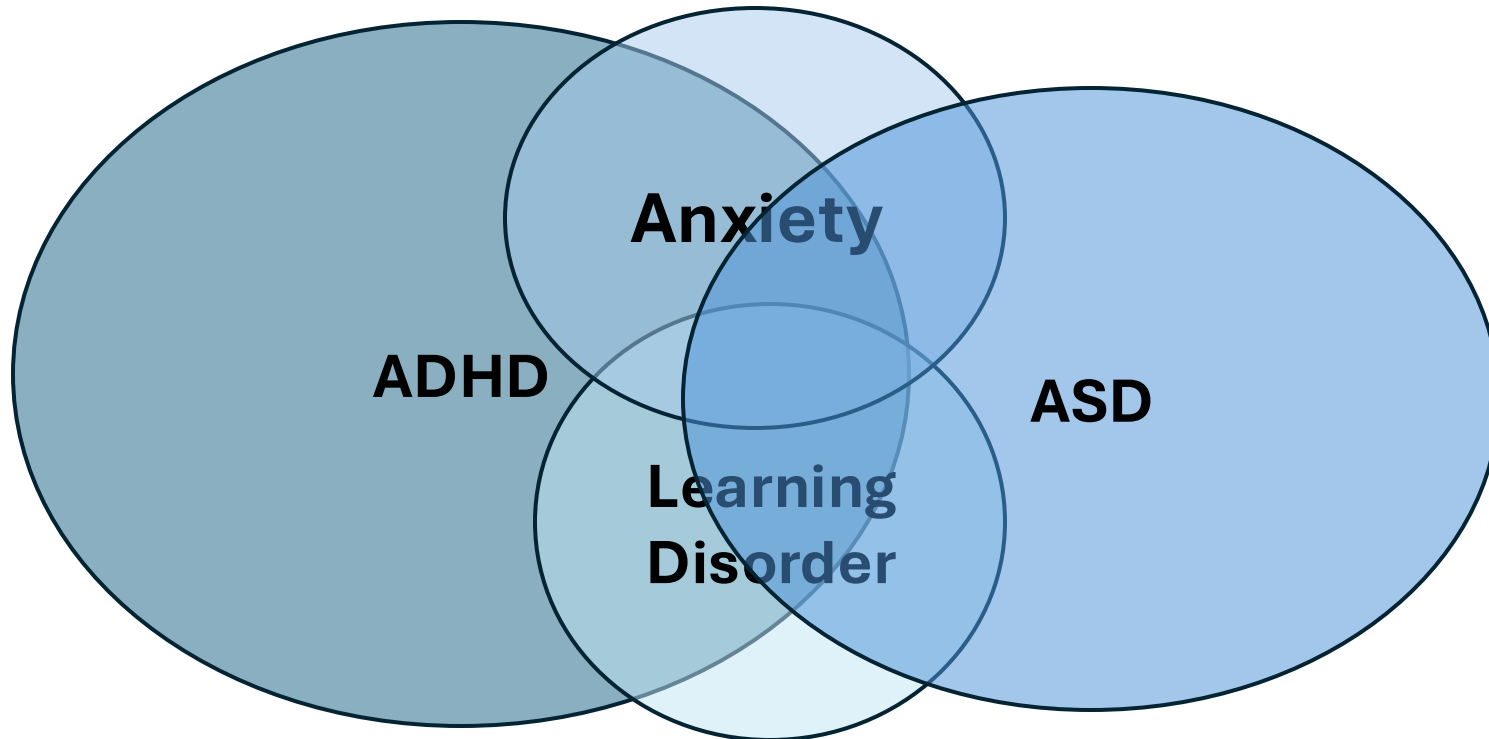
Sensory Processing

Emotional Regulation

Attentional Regulation

Social Communication

# “Alphabet Soup” Diagnoses Often Overlap



# FIGURE 1. THE FULL SPECTRUM OF IMPACT

## Neurodevelopmental Disorder Due to Dystrophinopathy

### WHAT IS READILY SEEN

The underlying genetic neuromuscular condition is often recognized.

### DYSTROPHINOPATHY

Duchenne or Becker muscular dystrophy and related dystrophinopathies



#### FOR PROVIDERS:

Know this.  
Screen for it.  
Treat it.  
Support the whole patient.

### WHAT IS OFTEN HIDDEN

Neurodevelopmental vulnerabilities are common, impact quality of life, and may require identification, intervention, and ongoing support.

### COMMON NEURODEVELOPMENTAL FEATURES AND CHALLENGES



#### SOCIAL COMMUNICATION DIFFERENCES

Challenges with social understanding, pragmatics, and peer relationships



#### ATTENTION / EXECUTIVE DYSFUNCTION

Inattention, hyperactivity, impulsivity, planning, organization, working memory



#### LEARNING CHALLENGES

Difficulties with reading, writing, math, and information processing



#### EMOTIONAL REGULATION DIFFICULTIES

Challenges with frustration tolerance, flexibility, and managing emotions



#### ANXIETY / OCD SYMPTOMS

Worry, perfectionism, obsessions, compulsions, and anxiety



#### MOOD SYMPTOMS

Low mood, irritability, loss of interest, sadness

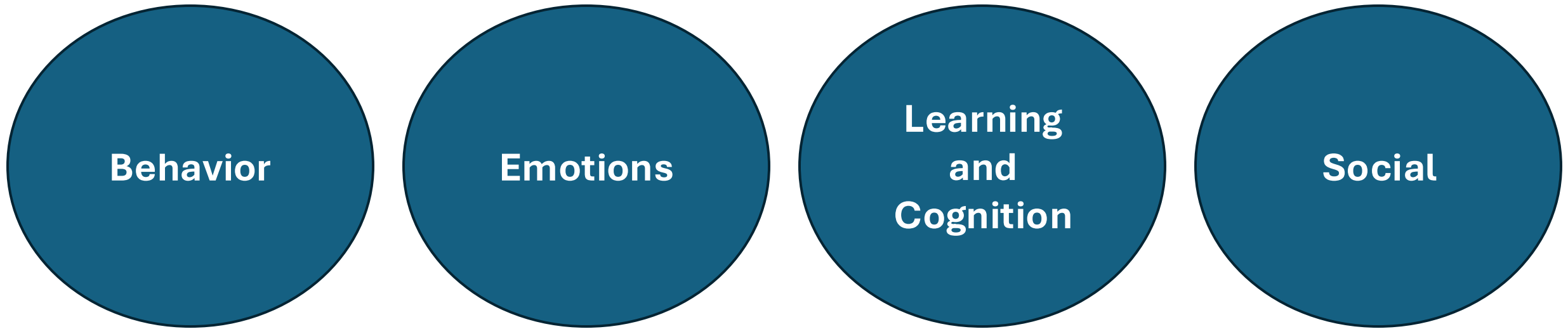
### LOOK BENEATH THE SURFACE.



### HOLISTIC, MULTIDISCIPLINARY CARE IMPROVES OUTCOMES

Early identification • Appropriate treatment • School & community support • Family partnership

# BELS: Towards a Functional Framework



# A Functional Framework

## Behavioral

- Sensory processing
- Motor Skills

## Emotional Functioning

- Anxiety
- Oppositionality
- Temper tantrums
- Frustration Tolerance

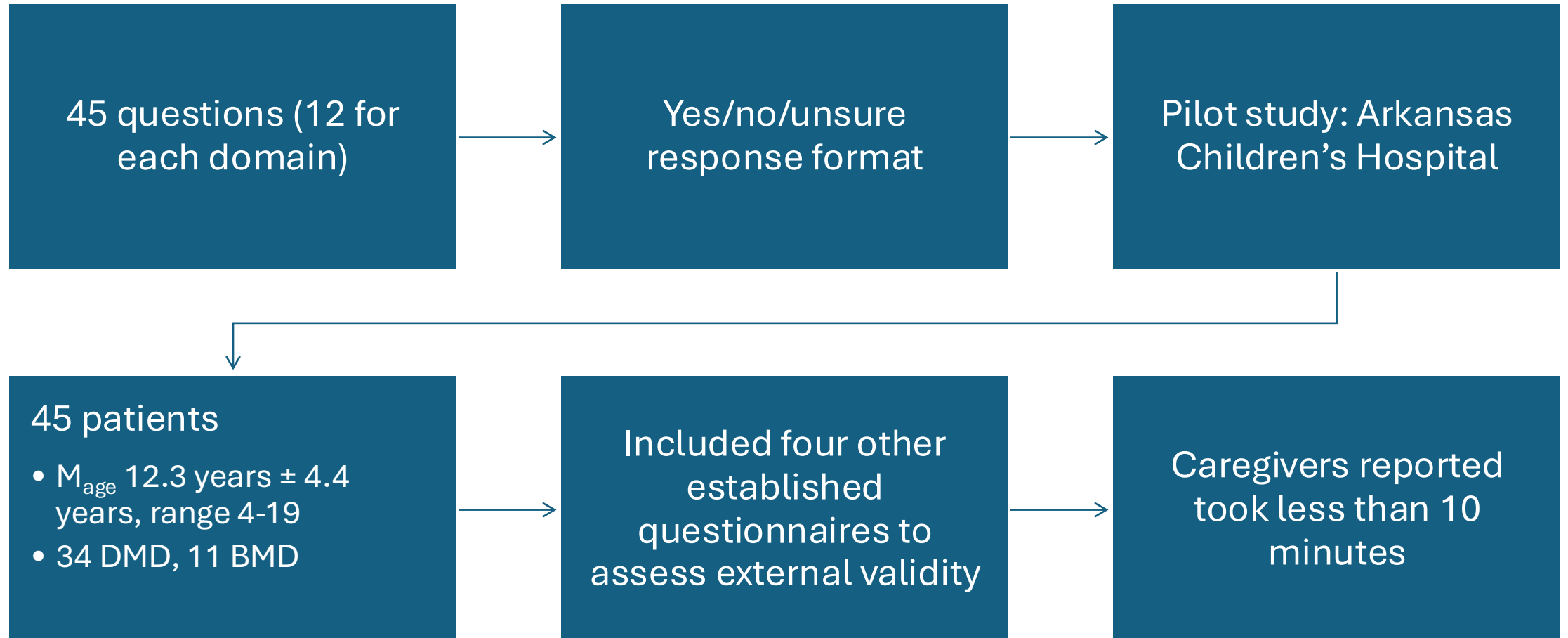
## Learning and Cognition

- Language
- Attention

## Social Skills

- Engagement and motivation
- Play skills

# BELS Development



Diagnosed Condition	Reported Symptoms on BELS
50% ADHD	> 60% distractibility, impulsivity, and restlessness
35% anxiety	50% anxiety symptoms (e.g., worries, fears)
12.5% ASD	> 1/3 social communication and peer relationship concerns
1 patient with LD; 2 with IDD	50% difficulties with reading, writing, and math

Truba, et al., *Muscle and Nerve*, 2025

# BELS Current Research

- **National validation study** in dystrophinopathy comparing BELS to other standardized screening questions
- **Targeted validation study** in pediatric patients with other neurodevelopmental disorders (autism, learning disorders, ADHD) to provide benchmarks for scoring
- **Comparing BELS to other tools** that are being developed in dystrophinopathy

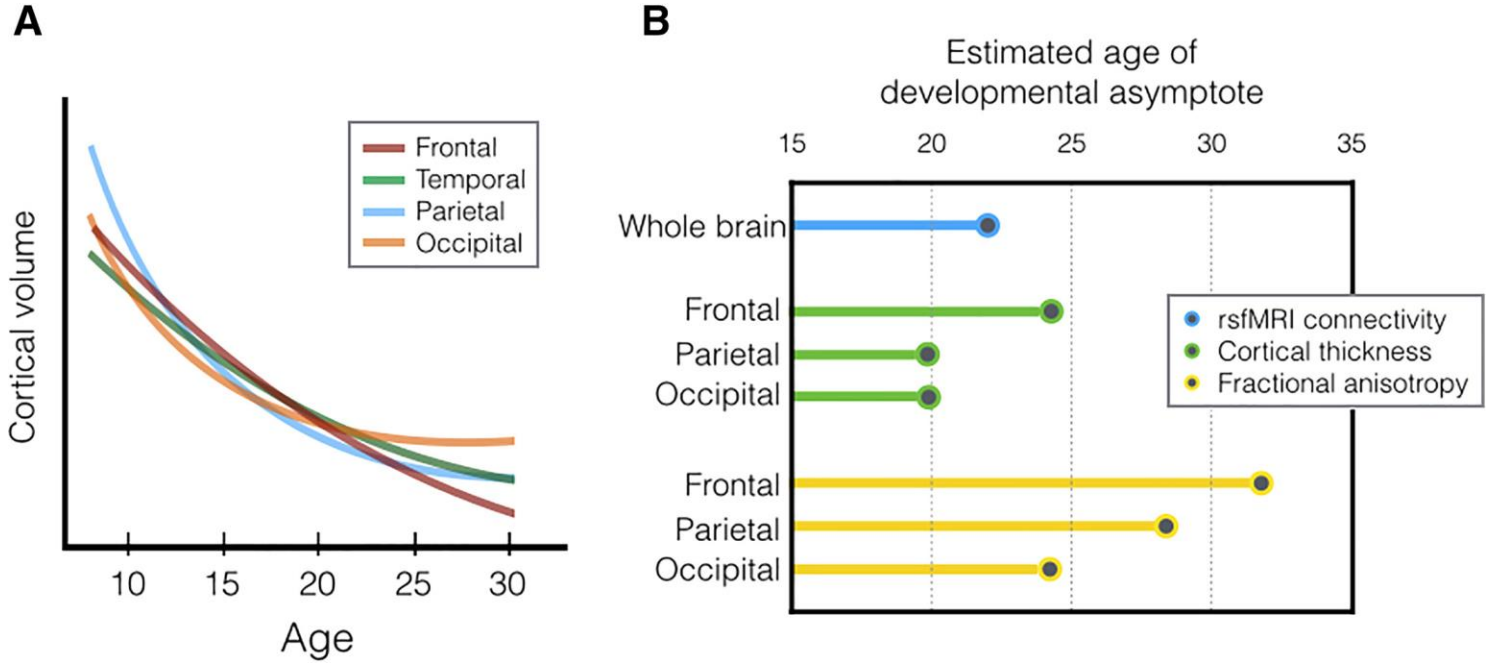
# Getting Help: Assessment and Support

# Importance of Early Intervention

- Improved functional outcomes
- Improved family functioning
- Lower caregiver stress
- Fewer barriers to medical treatment



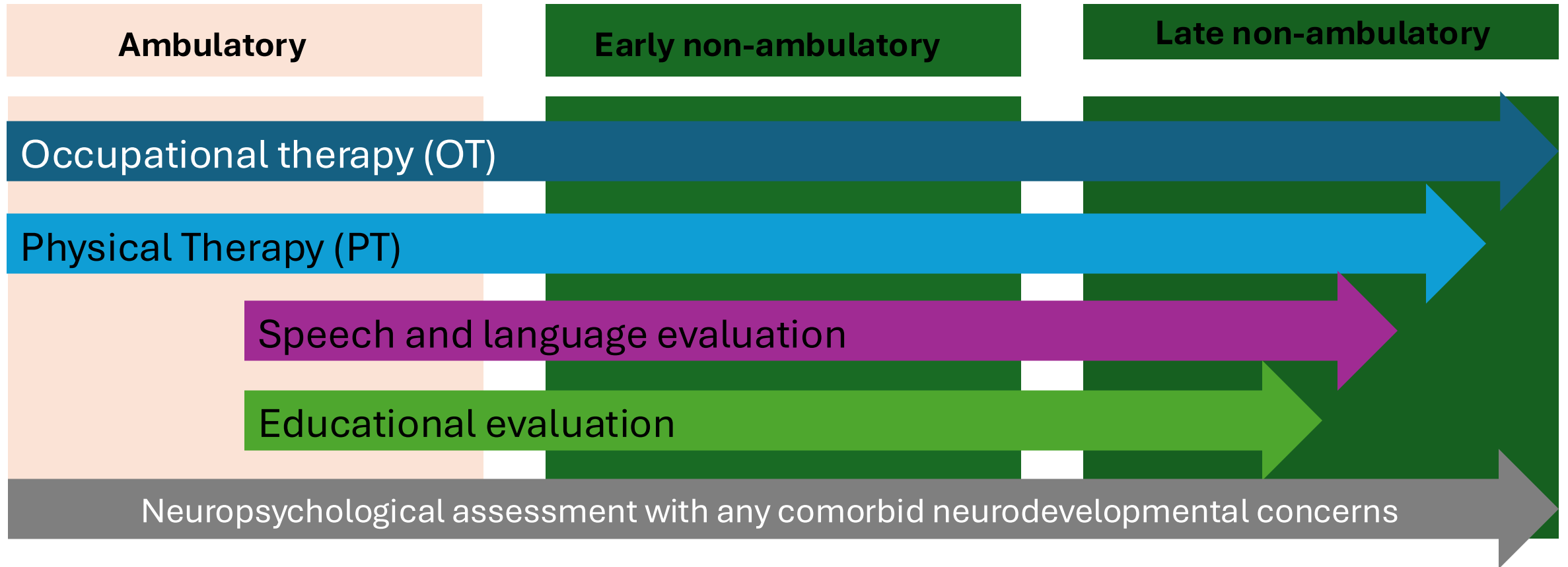
# Developmental Context



From Somerville

**Brain is still developing over the course of childhood and adolescence in Duchenne and Becker**

# Types of Developmental Assessment



Neuromuscular  
Clinic

Pediatrician



Psychiatry  
(medication)

Psychotherapy

Neuropsych  
Evaluation

School  
Services

Occupational  
Therapy

Physical  
Therapy

Speech and  
Language

Social Skills/  
ABA

# Why These Concerns Are Often Missed

- **Why concerns slip through the cracks**
  - Families may not realize these symptoms are connected to Duchenne or Becker
  - The pediatrician may assume the neuromuscular team is handling it — and the neuromuscular team may assume the pediatrician is
  - Not every specialist is trained to look for these concerns
  - Families may have limited access to evaluations
- **What helps**
  - Routine screening designed for Duchenne and Becker
  - Sharing the changes you notice at home and at school

Support and Treatment That Helps

# What Might this Look Like?

- **Whole Child Approach to Multidisciplinary Care:**
  - **Patient:** cognition/learning, emotional/behavioral, social relationships, sensory processing
  - **Family:** hope, parenting strategies and understanding, home environment, parenting philosophy, dynamics of primary caregivers
  - **Environment/Culture:** access to care, community and government supports
- Need for providers specializing in **fetal and infant development** and **transition to adulthood** (impact of newborn screening and longer lifespan)

# Support That Can Help

- **Therapies that build everyday skills**

- Occupational therapy for daily living and motor skills
- Behavioral therapy to build coping skills and ease worries — for children and caregivers
- Support for attention, memory, and problem-solving
- School and educational supports

- **Care that includes the whole family**

- Parenting strategies and support for parents and caregivers
- Building on each child's strengths

# Takeaways and Questions for Your Care Team

- **Remember**

- Learning, attention, emotional, and social differences are common in Duchenne and Becker
- Noticing them early opens the door to support and better outcomes
- You know your child best — your observations matter

- **Questions to ask your care team**

- Who on the team checks development, learning, and mental health?
- Would a developmental or neuropsychological evaluation help my child?
- What school supports or therapies could help right now?
- How can we support our whole family's wellbeing?

# Thank you

- All of you!
- Pat Furlong
- Kathi Kinnett
- Rachel Schrader
- Our patients and their families
- Our colleagues and collaborators
- Industry sponsors

**Parent** **JOIN THE FIGHT.**  
**Project** **END DUCHENNE.**  
**Muscular**  
**Dystrophy**