Emotional/Behavioral Health and Neurocognitive Functioning in DBMD

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Duchenne and Becker Muscular Dystrophy are neurodevelopmental conditions as much as they are neuromuscular conditions

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DMD Isoform Profile

- Often involves loss of multiple isoforms (Dp427 ± Dp140 ± Dp71, depending on mutation site)
- Compared to BMD (generally):
  - Intellectual disability (~20–25%)/Broader cognitive deficits more common
  - OCD and emotional dysregulation more common
- Implication of mutation site
  - More “global” neuropsychiatric phenotype when Dp140/Dp71 are affected

BMD Isoform Profile

- Typically, partial dystrophin expression is preserved
- Isoform involvement is variable but often less extensive than DMD
- Clinical presentation (generally):
  - Less global cognitive impairment
  - BUT still with substantially high rates of:
    - ADHD
    - Anxiety and mood disorders

Neurodevelopmental and psychiatric disorders: DMD vs BMD

Condition	DMD Prevalence	BMD Prevalence	
ADHD	~18%	~28%	Normative Rates
Autism	~7%	~6%	
Anxiety	~24%	~25%	
Depression	~11%	~7%	
OCD	~12%	~7%	

1. Vaillend C, Aoki Y, Mercuri E, et al. Duchenne muscular dystrophy: recent insights in brain related comorbidities. Nat Commun. 2025;16(1):1298. Published 2025 Feb 3. doi:10.1038/s41467-025-56644-w;
2. Truba N, Sorensen S, Bearden R, et al. The BELS questionnaire: a novel screening tool for neurodevelopmental and psychiatric symptoms in pediatric dystrophinopathy. Muscle Nerve. 2024;71(2):153-158
3. Pascual-Morena C, Cavero-Redondo I, Reina-Gutiérrez S, et al. Prevalence of neuropsychiatric disorders in Duchenne and Becker muscular dystrophies: a systematic review and meta-analysis. Arch Phys Med Rehabil. 2022;103(12):2444-2453

Labels Matter: Diagnoses versus "Features"

- Emotional/Behavioral dysfunction
  - **DMD = 38.7%**
  - **BMD = 38.6%**
- Inattention/hyperactivity-like
  - **DMD = 31.4%**
  - **BMD = 35.7%**
- OCD features
  - **DMD = 25%**
  - **BMD = 20%**
- Anxiety features
  - **DMD = 32.6%**
  - **BMD = 21.4%**
- Depressive features
  - **DMD = 15.9%**
  - **BMD = 5.7%**
- Motor and/or Vocal Tics
  - **DMD = 2.4%**
  - **BMD = 5.6**
- Language/speech delays
  - **DMD = 24.4%**
  - **BMD = 35.7%**
- Global intellectual delay
  - **DMD = 3.7%**
  - **BMD = 11.4%**
- Specific learning delays/ special education needs
  - **DMD = 19.6%**
  - **BMD = 31.4%**
- Autistic features
  - **DMD = 7.6%**
  - **BMD = 11.4%**

Darmahkasih et al. (2020). Neurodevelopmental, behavioral and emotional symptoms common in Duchenne muscular dystrophy. *Muscle & Nerve, 61,* 466-474; Lambert et al. (2020). Neurodevelopmental, behavioral and emotional symptoms in Becker muscular dystrophy. *Muscle & Nerve, 61,* 156-162

General Brain & Mental Health Takeaways

- Mental health and cognitive features are intrinsic to dystrophinopathies - not merely psychosocial reactions to having a disability
- CNS involvement is genotype-driven, not severity-driven
  - Brain dystrophin isoforms are affected depending on mutation location, not just disease type (DMD vs BMD)
- BMD is not “mild” neurologically
  - Even with milder muscle disease, BMD patients still show substantial CNS involvement
- Different mechanisms across domains
  - Neurodevelopmental disorders → largely biological/genetic
  - Depression/anxiety → biological + psychosocial interaction

Early Childhood (~2yrs-6/7yrs)

Shared features (DMD + BMD)

- ADHD and executive dysfunction
- Autistic traits / ASD diagnoses
- Language delays and learning difficulties

Differences: BMD vs DMD

- BMD may show equal or higher ADHD prevalence
- DMD may show more global cognitive impairment (lower IQ shift)

Conceptual framework

- Primarily neurodevelopmental and genetic (brain dystrophin deficiency)
- Symptoms often precede significant physical disability, especially in DMD
- Early differences between DMD and BMD are small, reinforcing that neurodevelopmental vulnerability is shared across dystrophinopathies

Late Childhood→ Adolescence (~6/7yrs-12yrs)

Shared features (DMD + BMD)

- Anxiety disorders
- Emotional dysregulation (irritability, frustration)
- Social difficulties

Differences: DMD vs BMD

- Minimal prevalence differences between DMD and BMD
- However: DMD patients often face earlier functional decline → greater psychosocial stress
  - ➢ This may amplify emotional symptoms

Conceptual framework (more mixed)

- Neurobiological (brain dystrophin effects)
- Psychosocial (school challenges, peer differences, emerging disability)
- This period reflects the interaction of brain-based vulnerability + lived experience, with DMD showing higher burden qualitatively (idiographic contextual factors), not dramatically quantitatively (functional impact of weak muscles)

Transitional Adult→ Early Adulthood (19yrs- 35yrs)

Shared Features (DMD + BMD)

- Depression
- Anxiety (persistent or worsening)
- Grief
- Complex psychiatric presentations (e.g., mood dysregulation, rare psychosis-risk states)

Differences: DMD vs BMD

- Depression higher in DMD
  - ➢ Likely driven by:
    Progressive loss of ambulation
    Increased dependency
    Reduced life expectancy (historically)

Conceptual framework

- Strong psychosocial contribution layered onto:
  - ➢ Ongoing CNS vulnerability
  - ➢ Transition-related stress (independence, identity, future planning)
- This is where DMD diverges most clearly from BMD, with greater internalizing burden reported by patients with DMD

Periods of Increased Vulnerability

Developmental transitions ↔ Functional transitions

- Late childhood/early adolescence → increased functional impact d/t disease progression (e.g., falling more)
  - Increased awareness of weak muscles and how this makes them different from their friends/peers/siblings
    - Risk of internalizing symptoms like anxiety, depression, & self-doubt
    - Parental reactions/responses often misperceived or misinterpreted
  - Familial narrative around DBMD dx → how DBMD is integrated into one's life narrative & how development of sense of self increasingly more important

Periods of Increased Vulnerability

Developmental transitions ↔ Functional transitions

- Adolescence → puberty; increasing need for caregiver involvement/support to complete things like ADLs; increasing dependence on technology (e.g., power chair)
  - Social environmental and ease of which they can access & engage alongside peers starts to be more challenging (e.g., friends' homes not accessible, community spaces not designed adequately)
  - Desire to fit in/be liked ("be normal") & social dynamics increasingly more important
    - peers more influential than family
  - Sleep and other health habits might change (physio and beh drivers)
  - Desire more independence (normative) but increasing need for support (abnormal barrier to independence)

Periods of Increased Vulnerability

Developmental transitions ↔ Functional Transitions

- 17/18+ yrs → transition into adulthood; increased caregiver and technology dependency and functional independence may start declining
  - Social opportunities often diminished if not pursing additional education (college/trade)
  - Hopes/goals for the future, meaning-making and finding one's value (existential)
  - Normative adult transition stressors
    - changes in health/more decision-making demands
    - financial/family planning (e.g., aging parents, planning for long term financial stability)
  - Physical impact of chronic steroids (e.g., short stature, lack of mature adult features)
  - Need for new/more robust technology (e.g., wheelchair, cough assist, nighttime bipap, gtube, ostomy, trach/vent)

Managing Needs into Adulthood

Early-Late Childhood

- Early monitoring and early intervention for autism, ADHD, anxiety, developmental delays (e.g., language), sensory integration issues
- Social skills training (formal and informal)
- Behavioral Science education/training for parents
- Promoting developmentally appropriate independence
  - Demands/expectations for behavior and investing in being independent
  - Appropriate limit setting

Adolescence-Adulthood

- Actively shaping independence is incredibly important
  - Critical thinking/problem solving, directing care & learning how to direct others in caring for them, doing things without parents/caregivers immediately available
- Holding space for possibilities while learning to ground self in probabilities
- Normalize proactive and ongoing management of mental/behavioral health (anx, dep, grief; healthy habits)
- Promoting focus on meaning-making and living a valued life; social connection & hobbies

THANK YOU ☺

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