The Big Picture

- Most boys with DMD will show steady developmental progress in the acquisition of cognitive, emotional, and social skills, even with loss of motor skills.

- Boys with DMD will lead fulfilling lives, pursue areas of interest and expertise, and have meaningful social relationships.

- Improved medical management of DMD has resulted in improved quality of life.

- Assessment and treatment of concerns related to psychosocial health should be part of routine clinical care.

- While many boys have age-appropriate neuropsychological abilities, there is increased risk of psychiatric and neurodevelopmental conditions.
Mental Health Risks
Coping with Chronic and Progressive Medical Illness

• Medical Factors
  – Chronic pain and fatigue
  – Medication effects
  – Restriction of activities
  – Need for help to execute daily activities

• Emotional, Social, and Cognitive Stress
  – Vigilance to potential health threats or physical changes
  – Rumination on past or future
  – Guilt
  – Social isolation from peers
Family Stress

• Don’t forget parents, siblings, and extended family

• Caregivers do generally report a high degree of life satisfaction
  – Also have increased risk of anxiety and depression, especially around transition points
  – DMD may influence parenting decisions

• Demands on Limited Resources (Financial and Time)
  – Medication and appointment copays
  – Managing medications and appointments
  – Adaptation for specialized equipment
  – Insurance costs
  – Employment restrictions due to caregiving demands

• Interventions:
  – Increase mother’s resilience and social support
  – Enhance positive perceptions of the disease: hope, caring, empathy
  – Consider the emotional impact of genetic testing on the mother
  – Sibling support
Common Psychiatric Conditions in Patients

- Anxiety, especially social anxiety
- Depression
- Obsessive compulsive disorder (OCD)
- Oppositional or explosive behaviors
- Social communication problems
Psychiatric Treatment Approaches

- Psychotherapy Formats
  - Individual
  - Family
  - Group (Peers)

- Psychotherapy Modalities
  - Cognitive Behavioral Therapy (CBT)
  - Acceptance and Commitment Therapy (ACT)
  - Collaborative Problem Solving
  - Behavioral Therapies (e.g., Applied Behavior Analysis, social skills training)

- Medication

- There is very little empirical data examining efficacy of treatments in DMD
Anxiety

• All children worry, especially kids with health-related concerns

• Concerns are Clinically Significant when:
  – Frequency is high
  – Intensity is high
  – Interferes with daily functioning

• Specific phobias
  – Can be medically-related (e.g., swallowing pills, needles, etc)

• Social anxiety
  – Excessively shy, fear of doing something embarrassing or wrong
  – Becomes tense or rigid in new situations
  – Reluctant to engage with new people
  – Avoids social interactions: leaves situations or uses technology
    – Important to rule out underlying social communication problems
GAD and OCD

- **Generalized Anxiety Disorder (GAD)**
  - Worries about what has happened or will happen
  - Excessive tension, difficulty relaxing or sleeping
  - Frequently seeks reassurance

- **Obsessive Compulsive Disorder (OCD)**
  - “Get stuck” on particular thoughts or behavioral routines
    - Obsessions: death, dying, religion, illness
    - Compulsions: checking, evening up, confessing
  - Perfectionism, high expectations
  - Can become very upset if rituals are interrupted or prevented
Depression

• Risk Factors:
  – Coping with DMD
  – Social Isolation
  – Thinking Patterns (e.g., rigidity)

• Symptoms:
  – More withdrawn or irritable, crying spells
  – Negative thought processes
  – Guilt, low self-esteem
  – Self-injurious behaviors or suicidal ideation

• Risk may increase in adolescence
Oppositional and Explosive Behavior

- Behavioral Rigidity
  - Adherence to Routines
  - Difficulty Quickly Adjusting to Change

- Heightened Emotional Reactivity
  - Easily irritated or angered

- Reduced Sensitivity to Consequences or Punishment

- Often co-occurs with ADHD, ASD, mood, and anxiety concerns

- Treatment Options:
  - Psychotherapy (CBT, Collaborative Problem Solving, sometimes ABA)
Neurodevelopmental Risks
Brain Development in DMD

• Dystrophin plays a big role in muscle development and function, but it is also expressed in brain

• Depending on the nature of the genetic mutation, there can be changes in brain development
  – Smaller total brain volume
  – Smaller gray matter volume
  – Changes in the structure of white matter pathways
Brain Development in DMD

- Dystrophin is expressed in higher levels in certain brain regions important in learning.
- These changes may contribute to learning differences in DMD.
- May relate to the higher prevalence of neurodevelopmental conditions in DMD.

Doorenweerd (2017)
DMD Neuropsychological Profile

• Potential Strengths
  – Acquired knowledge of the world (e.g., vocabulary, facts)
  – Visual pattern recognition and spatial skills
  – Retention of learned knowledge over time

• Potential Weaknesses
  – Aspects of executive functions (processing speed, working memory, mental flexibility)
  – Aspects of higher-order language processing
    • Phonological processing that supports reading and spelling
    • Language comprehension
    • Pragmatic (social) language
  – Social skills and emotional regulation
Behavior

Executive Functions

Emotional Functions

Social Functions

Language Processing

Medical Factors
Recognizing Neurodevelopmental Disorders

• Intellectual Disability (17-27%)
• Learning Disabilities (26%)
• Attention Deficit Hyperactivity Disorder (ADHD, 32%)
• Autism spectrum disorder (ASD, 15%)
Intellectual Disability

• Steady developmental progress but skills uniformly fall below age-based expectations

• Warning Signs:
  – Delayed acquisition of skills in all areas (global delay)
  – Seems younger than same-aged peers in all areas

• Important to support the development of functional living skills
Learning Disability

• Many aspects of functioning, including overall cognitive abilities, are within normal limits for age

• Certain academic skills are harder to learn than others
  – Reading (e.g., dyslexia)
  – Writing (e.g., dysgraphia)
  – Math (e.g., dyscalculia)

• Warning Signs:
  – May be general (e.g., difficulties focusing in school)
  – May be specific (e.g., trouble with reading)

• May need specialized academic supports
Attention Deficit Hyperactivity Disorder (ADHD)

• Inattentive Symptoms
  – Can focus on preferred activities
  – Hard time focusing on nonpreferred activities, especially if too easy or too challenging

• Hyperactivity
  – Restlessness
  – “can’t sit still”

• Impulsivity
  – “acts without thinking”

• Treatment Options:
  – Medication
  – Psychotherapy (Cognitive Behavioral Therapy, parent guidance)
Autism Spectrum Disorder (ASD)

• Difficulties relating to other children and/or unusual play interests
  – Prefer to play alone
  – Play tends to be repetitive and scripted
  – May play with toys in unusual ways
  – Shows reduced awareness or understanding of others’ emotional experiences

• Prefers to follow routine, may have difficulty adjusting to change; can be associated with outbursts and explosive behaviors

• May have difficulties with language processing and/or language delays

• Sensory sensitivities

• Treatment
  – Behavioral Therapy (ABA)
  – social skills training
  – specialized academic programs
What Can Clinicians Do?
Discuss Psychosocial Functioning

• Topics to Cover:
  – Individual Functioning
  – Family Functioning
  – School or Vocational Concerns
  – Interactions with peers (friendships, dating)
  – Access to Resources and Support

• How Cover Them:
  – At any age, patients should have a developmentally-appropriate understanding of DMD.
  – Patient involvement in medical care should be encouraged and be developmentally-appropriate.
  – Raise difficult topics in a sensitive way that allows time to make decisions and behavioral shifts.
Screen for Neuropsychiatric Conditions

• Anxiety
• Depression
• Social Communication Difficulties/Autism Spectrum Disorders
• Explosive or “Oppositional” Behavior
• OCD
• ADHD
• Learning Disabilities
• Language Delays
• Global Cognitive Delay
Address Mental Health in the Neuromuscular Clinic

• Care Coordinator
  – bridge to the family’s local community and national resources
  – Provides caregiver and patient support between clinic visits

• Standardized Mental Health Screening Forms
  – At every visit and recorded in the medical record to allow for longitudinal tracking
  – Pediatric Patients: PARS-III, Strengths and Difficulties Questionnaire
  – Adult Patients: PHQ-9 (depression), GAD-7 (anxiety)

• Mental Health Clinician
  – Annual Formal Assessment
  – Further Evaluation when Screening Forms are Positive
Know Your Available Mental Health Resources

• Psychiatrist
  – Medication management in consultation with other specialties (e.g., cardiology, neurology)

• Psychologist
  – Individual therapy
  – Family therapy

• Neuropsychologist
  – Clinical psychologist with specialized training in assessment, neurology, and psychiatry
  – Assess for neurodevelopmental, learning, and/or psychiatric conditions
  – Develop comprehensive treatment plans (need for psychiatric services, educational interventions, targets for further monitoring)
  – Recommended for all pediatric patients within first 12 months of diagnosis or at school-age
  – Recommended for all patients if change in functioning, especially decline
Understand Special Education Services

• Early Intervention Services (infancy to age 3)

• Types of U.S. Public School Education Plans
  – 504 Plan – Accommodations only
  – Individualized Education Program (IEP) – Accommodations and services
  – Plans can change over time and should be assessed annually

• Available Public School Services
  – Special Education (generalized and/or in a single subject)
  – Speech and Language Therapy
  – Occupational Therapy
  – Physical Therapy
  – Counseling and/or social skills training
  – Behavioral Therapy (e.g., applied behavior analysis)

• Parents can request evaluation for service eligibility from their public school
  – Purpose is to determine whether child has skills/behavior to access the curriculum.
  – This is not a neuropsychological evaluation.
Know State and National Resources

• Advocacy organizations
• Vocational Training
• Transportation Assistance
• Housing Assistance
• Financial and Legal Planning
• Disability Requirements for those with Neurodevelopmental Disorders
<table>
<thead>
<tr>
<th>ASSESSMENTS</th>
<th>AMBULATORY/CHILDHOOD</th>
<th>EARLY NON-AMBULATORY/ ADOLESCENT/YOUNG ADULTHOOD</th>
<th>LATE NON-AMBULATORY/ ADULTHOOD</th>
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<tbody>
<tr>
<td></td>
<td>Consider baseline evaluation during first year of diagnosis</td>
<td>Neuropsychological evaluation to identify cognition/learning issues with concerns about school performance</td>
<td>Evaluations when there are concerns about change in functioning or ability to manage daily affairs.</td>
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<td></td>
<td>Developmental (&lt; 4 years old) or neuropsychological evaluation (&gt; 5 years old) when social/emotional concerns or cognitive delays</td>
<td>Neuropsychological evaluation when transitioning to adulthood if state-based assistance may be needed</td>
<td>Evaluation by speech/language pathologist for older patients with loss or impairment of functional communication ability; refer patients having issues with chewing or dysphagia.</td>
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<td></td>
<td>Evaluation by speech and language pathologist for children with suspected delays in speech/language development.</td>
<td>Social worker evaluation of the needs of the family/patient.</td>
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<td></td>
<td>Social worker evaluation at diagnosis and then as needed.</td>
<td>Refer for psychotherapy and/or psychopharmacology when mental health concerns are identified for the individual or family.</td>
<td>Assist with continuing education, vocational training, extended transitional education with IEP until 22 years old.</td>
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<td>INTERVENTIONS</td>
<td>Refer for psychotherapy and/or psychopharmacology when mental health concerns are identified for the individual or family.</td>
<td>Implement formal accommodations at school for health, safety, and accessibility; plan for health-related absences.</td>
<td>Assist with adjustments to accommodate job requirements.</td>
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<td>Provide parents with resources to educate teachers, school psychologists, and other school personnel about DMD.</td>
<td>Provide parents with resources to educate peers about DMD. Refer to psychologist for social skills training if needed.</td>
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<tr>
<td></td>
<td>Provide parents and patients with resources to educate peers about DMD. Refer to psychologist for social skills training if needed.</td>
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<td>Encourage patients/families to stay active and engaged.</td>
<td>Promote patient self-advocacy and independence.</td>
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<td>Arrange for home health care services if patient's health is at risk because sufficient care cannot be provided in current setting.</td>
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<td>Notify patients/families about availability of palliative care.</td>
<td>Assist with arranging respite care for caregivers.</td>
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<td>Make hospice care available for end-stage patients.</td>
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Questions?

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