THE PSYCHOLOGICAL COMPLEXITY OF MYOTONIC DYSTROPHY

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Introduction

- DM1 affects more than physical function
- Psychological component
  - Cognitive functions
  - Psychosocial functioning
- Why is this important?
  - Impacts affected individual, family, and caregiver
  - Physical changes may contribute to psychological changes
  - Foster healthy social and emotional growth
Defining the Psychobable

- **Cognitive Functioning**: means and mechanisms of acquiring knowledge (i.e., reasoning, memory, perception, awareness, attention, judgment, and language)

- **Executive Function**: cognitive processes necessary for the cognitive control of behavior

- **Psychosocial Functioning**: psychological function of individual in the context of his/her social environment

- **Behavioral Functioning**: behavior in the context of social environment
Physical Concerns

- Progressive muscle weakness
- Slow motor development
- Tire easily
- Sit to stand difficulties
- Decreased muscle power
- Myotonia and locking up
- Falls
- GI
- Eating and Swallowing
Implications

- **Loss of muscle and strength**
  - Fatigue
  - Tired
  - Falls
  - Anxiety
  - Depression

- **Physical symptoms that may impact psychological functioning**
  - Fatigue, sleep problems, incontinence, GI complaints, pain, headache, swallow
  - May contribute to anxiety, depression, attention, memory, daily functioning, executive function problems
Addressing Physical Concerns

- **Communication**
  - Express concerns and feelings
  - Active listening

- **Adapt environment to disease progression**
  - Adapted environment contributes to feelings of success and not feelings of loss
  - Increase social interactions
  - Decrease apathy, depression, anxiety

- **Adaptive devices, power chairs, AFOs**
  - Sense of freedom and stability
  - Increase confidence and social interactions
  - Less fearful of falling
Cognitive Function

- Thinking
- Memory
- Language
- Attention
- Executive Function
  - Planning
  - Organization
  - Inhibition
  - Shift
  - Emotional control
  - Initiation
  - Working memory
  - Monitoring behavior
Research

- **Health Endpoints and Longitudinal Progression in Congenital Myotonic Dystrophy (HELP-CDM)**
  - Nicholas Johnson, MD, University of Utah
  - Craig Campbell, MD, Western University, Canada
  - 3 year study following 50 children with CDM
  - Cognitive assessments: IQ, executive function, adaptive behavior, autism, QOL, sleep

- **Health Endpoints and Longitudinal Progression in Myotonic Dystrophy (HELP-DM)**
  - Nicholas Johnson, MD, University of Utah
  - 1 year study with f/u at 3 months of 22 adults with DM1
  - Cognitive assessments: IQ, executive function, adaptive behavior, memory, visuospatial abilities, processing speed, QOL, sleep, anxiety, depression, apathy
What did we learn from HELP-CDM?

### Cognitive Functions: CDM vs Control

<table>
<thead>
<tr>
<th>Construct</th>
<th>Measure</th>
<th>CDM $\bar{x}$ (sd)</th>
<th>Control $\bar{x}$ (sd)</th>
<th>t-score (p-value)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Intelligence (IQ)</td>
<td>WPPSI-III/WAIS-IV</td>
<td>66.1 (18.1)</td>
<td>100 (15)</td>
<td>-10.60 (0.000)</td>
</tr>
<tr>
<td>Adaptive Behavior</td>
<td>Vineland-II</td>
<td>70.0 (16.2)</td>
<td>125.6 (15.5)</td>
<td>-10.15 (0.000)</td>
</tr>
<tr>
<td>Executive Function</td>
<td>BRIEF parent</td>
<td>63.3 (11.1)</td>
<td>$\leq$ 65</td>
<td>6.20 (0.000)</td>
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<tr>
<td></td>
<td>BRIEF teacher</td>
<td>72.9 (13.8)</td>
<td>$\leq$ 65</td>
<td>6.80 (0.000)</td>
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<tr>
<td>Autism Traits</td>
<td>ASSQ</td>
<td>15.0 (9.3)</td>
<td>$\leq$19</td>
<td>6.10 (0.000)</td>
</tr>
<tr>
<td>Social Communication</td>
<td>SCQ</td>
<td>12.4 (7.1)</td>
<td>$\leq$15</td>
<td>5.33 (0.000)</td>
</tr>
<tr>
<td>Repetitive Behavior</td>
<td>RBS-R</td>
<td>16.4 (14.7)</td>
<td>$\leq$ 13</td>
<td>NA</td>
</tr>
</tbody>
</table>
What did we learn from HELP-CDM?

Parent and Teachers Responses on Behavior Rating Inventory of Executive Function

Mean score ≥ 65 indicate clinical significant
What did we learn from HELP-DM?

- IQ: $x = 88.15$ (9.7), average (norm 85-115)
- BDI: $x = 11.68$ (7.4), moderate depression
- BAI: $x = 11$ (8.0), moderate anxiety
- EF: Shift $x = 67$ (WM & initiate- trending)
- Slowed processing speed
- Memory impairment
What does this look like in daily life?

- **EF/CF problems affect language, memory, and concentration**
  - Automation, planning, and learning
    - Task completion
    - Time orientation
    - Initiative
    - Resistance to change
    - Switching tasks
    - Behavior
    - Mood
Important to know

- Cognitive function issues are not one of physical function or motivation.

- Individuals are not unwilling.

- Unable to complete tasks and follow directions in the same way as others.

- Rethink the way we approach giving tasks/asking for things.
What can you do to help?

- Strategize to find ways that work for individuals
- Memory: short sentences, clear instructions, divide information
- Visual cues to help process information
- Check in- Does she understand?
- Let him repeat what he’s been told to synthesize and build verbal memory
- Summarize and repeat information
- Introduce new information in steps
What can we do to help?

- Extra time for instructions
- Break tasks into smaller parts (two 10 minute vs. one 20 minute)
- Quiet location with limited interruptions
- Make lists and check off completed tasks (individuals can do)
- Agenda training (individuals can plan their days, weeks, etc.)
- Extra time for transitions
- Help get things started
- Create a system for organizing and planning
- Use lists and calendars
- Create system with check lists for self-monitoring
Important to know

- Emotional and behavioral problems are normal to healthy socio-emotional development
- Emotional reaction to disease progression is normal for affected individuals and family

  - Stages of grief
    - Denial
    - Anger
    - Bargaining
    - Depression
    - Acceptance
Psychosocial Functioning

- Emotional and behavioral concerns
  - Important clinically because children/adults recognize consequences of their disease

- Psychosocial adjustment related to:
  - Relationships w/ friends (↓ w/ age and/or disease progression)
  - Dependence, hostility, productivity (↕↓ w/ disease progression)
  - Anxiety, depression, and withdrawal (↑ w/ age or disease progression)
Psychosocial Functioning

- **Learned helplessness**: when an action does not have an impact, inclination to give up hope and make no further effort
  - Can lead to
    - Emotional reaction (apathy and depression)
    - Behavior problems (passivity, tantrums)
- **How to help**
  - Activities that individual can complete (create a positive experience)
  - Increased sense of control leads to increased motivation to continue
Psychosocial Functioning

- **Emotional concerns**
  - Increase when symptoms increase; associated anxiety
  - Increase when there is a loss of independence and/or control-
    - response: hostility, depression, relief
  - Increase when peers are moving on and individuals feel stuck because of their DM

- **How to help**
  - Important to respond as you would to any child/adult
  - Normalize emotional response given circumstances
  - Mourn the loss to help move toward acceptance
    - Stages: denial, anger, bargaining, depression, acceptance
  - Address emotions, learning opportunity, listen, name emotions, solve problems and set limits
  - Work together to be successful!
Psychosocial Functioning

- **Tantrums/Wallowing**
  - Normal process of growing up
  - Want 100% of your attention
  - Best to address tantrum with boundaries

- Normal course of tantrum: 1) reason no matter how small or meaningless, 2) escalation, 3) explosion, 4) cooling down

- **How to help**
  - Set limits, respond clearly, allow tantrum to run its course w/o intervention or reaction, time out to cool down, discuss emotions
  - Accept person’s emotions, but set limits on negative behavior
Caregivers burn-out is a real thing

- Take time for yourself-this is not being selfish
  - Relaxation, sleep, exercise
  - Date nights
  - Time outs

You are a better caregiver when you take time for yourself!
Resources

- Myotonic Dystrophy Foundation
  - www.myotonic.org

- MDA
  - www.mda.org
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Questions?