PSYCHOSOCIAL ISSUES IN DMD

End Duchenne Tour

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OUTLINE

1) Causes
2) Learning Problems
   - General Learning Problems
   - Specific Learning Disorders
3) Neurobehavioral Disorders
   - Externalizing
   - Internalizing
4) Coping
5) What to do about it

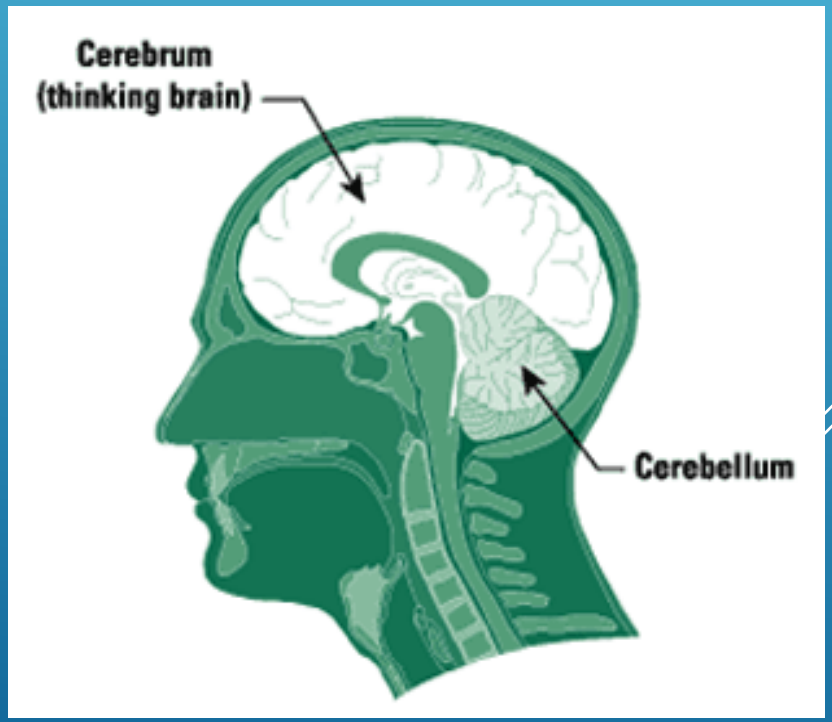
POTENTIAL CAUSES

- Psychological
  - Coping with DMD
- Psychosocial Factors
  - Family stress/conflict
  - Parenting
  - Peer interactions
  - Teachers/adults
- DMD impact on brain functioning
- Medical factors
  - Steroids
  - Fatigue/sleep
  - Medical procedures
  - Blood sugar
  - Clinical Trials

FULL- LENGTH DYSTROPHIN (DP 427)

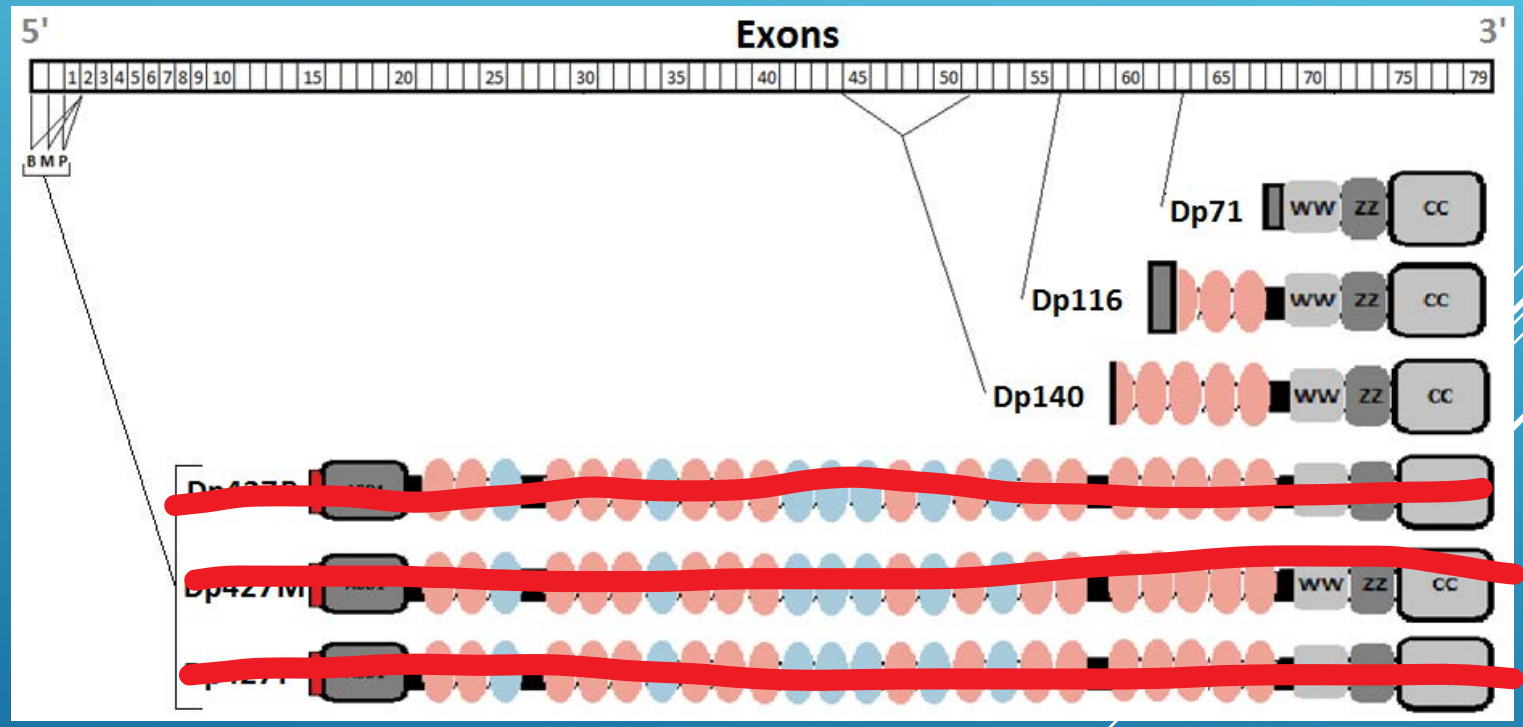
Usually found in different brain areas:

- Cerebral Cortex
- Subcortical structures
- Hippocampus
- Cerebellum – Purkinje Cells
- GABA neurons – primary inhibitory mechanism of brain
- Also glutamate, acetylcholine, NMDA



DYSTROPHIN IN THE BRAIN

- Everyone with DMD is missing Dp427



MISSING DYSTROPHIN IN THE BRAIN

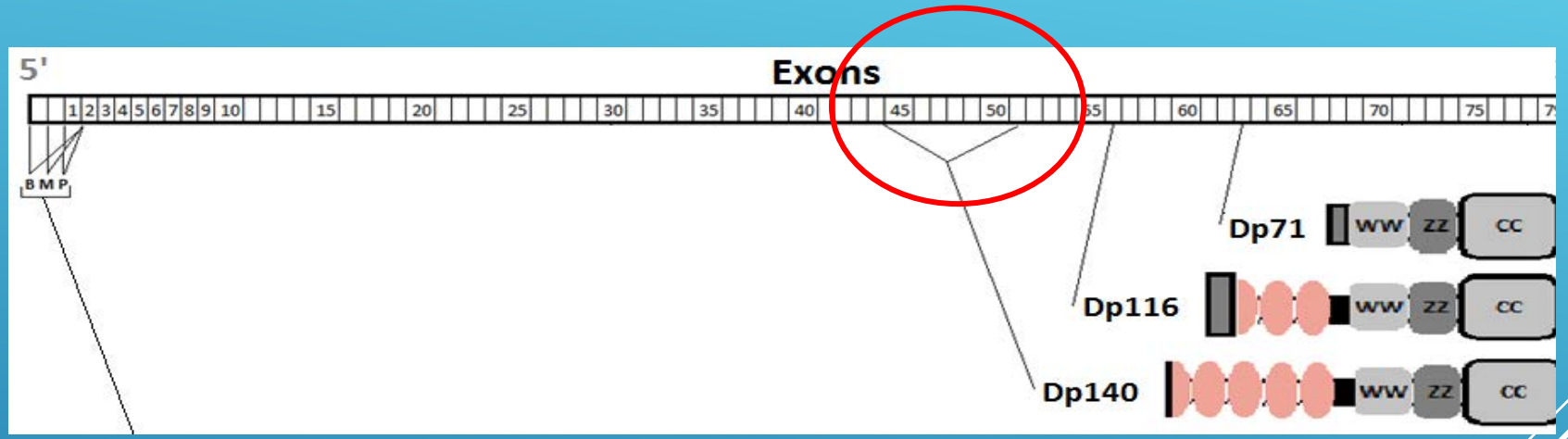
- Neurons
  - Ion channels not clustered correctly
    - less efficient in sending signals to each other
    - less ready for new signals
  - Changes in shape and size
  - Fewer neurons
  - Less new “connections”

- Structural
  - Blood brain barrier less “tight”
  - Smaller grey matter
  - Less white matter density

- Functional
  - Differences in metabolism
  - Increase in choline-containing compounds
  - Higher incidence of abnormal EEG

Knuesel et al., 1999; Vaillend & Billard, 2002; Kueh, Head, Morley, 2008; Doorenweerd et al., 2014

MISSING DYSTROPHIN IN THE BRAIN



- Some may also be missing smaller versions
- 55% of mutation breakpoints occur between introns 45 - 51

Pane et al 2012

- Region for Dp140
  - Missing Dp140 related to smallest brain grey matter
  - Greater risk for cognitive/behaviour problems

DYSTROPHIN IN THE BRAIN

- WHY DOES MY SON/DAUGHTER WITH DMD HAVE PROBLEMS……(fill in the blank)?

ANSWER: IT'S NEUROLOGICAL

GENERAL LEARNING PROBLEMS: INTELLIGENCE

- Most have IQ in the normal range
- Average IQ score lower (80)
- Increased amount in the intellectual disability range (35% below 70)
- Verbal scores lower than visual-spatial scores

Cotton, S, N. Voudouris, and K. M. Greenwood. 2001
Cotton, S, N. Voudouris, and K. M. Greenwood. 2005.

GENERAL LEARNING PROBLEMS: LANGUAGE

- Greater chance of delays in language development
- Greater problems in younger children
- Normal vocabulary
- Problems with:
  - complex comprehension and verbal fluency
  - About two years behind

Cyrulnik, S.E., Fee, R., Batchelder, A., Kiefel, J., Goldstein, E., & V.J. Hinton. 2008;
Cyrulnik, S., Fee, R., De Vivo, D.C. & V. J. Hinton.. 2007

GENERAL LEARNING PROBLEMS: SHORT-TERM MEMORY

- Weaknesses in short-term memory
  - Worst for verbal information
  - Pattern seen at all IQ levels and ages

Hinton, V. J., N. E. Nereo, D. C. DeVivo, E. Goldstein, and Y. Stern. 2000.

Hinton, V. J., N. E. Nereo, D. C. DeVivo, E. Goldstein, and Y. Stern. 2001.

Hinton V. J. , Fee R., Goldstein, E.& D.C. De Vivo. 2007.

SPECIFIC LEARNING DISORDERS IN DMD

- About 40% have one or more learning disorders:
  - Dyslexia: Difficulty learning to read
  - Dyscalculia: Difficulty learning math
  - Dysgraphia: Difficulty with writing

Hendriksen & Vles 2006

- Anxiety related to specific subject
- Shutting down, overwhelmed
- Frustration, exploding
- Poor performance relative to other subjects
- Says “I hate _____!” (reading, math, writing)

OTHER SIGNS OF A LEARNING DISORDER

NEUROBEHAVIORAL DISORDERS IN DMD

Attention-deficit disorder: 12% – 44% in DMD (with or without hyperactivity-impulsivity)

Signs to look for:

Impulsive

Blurts things out

Interrupts

Impatient

Fidgets

Easily frustrated

Too loud

Avoids work

Overly focused on fun

Easily distracted

Messy and disorganized

Forgetful

Daydreams

Difficulty following directions

Hendriksen & Vles 2008; Poysky & Lotze, 2008; Hinton et al. 2006; Pane et al. 2012; Ricotti et al 2015

NEUROBEHAVIORAL DISORDERS IN DMD

Autism: 3-21%

Signs to look for:

- Delayed language development
- Excessive and unusual interests/obsessions and routines
- Impaired understanding of social interactions

Wu et al. 2005; Hendriksen & Vles, 2008; Darke, Bushby, Le Couteur, McConachie, 2006; Hinton et al 2009; Ricotti et al 2015

NEUROBEHAVIORAL DISORDERS IN DMD

Oppositional, argumentative, & explosive behavior:

15% - 52% of boys with DMD?

- Difficulty controlling emotional reactions
- Easily irritated, angry outbursts
- Blames others
- Hard time predicting consequences
- Punishment increases anger/bad behavior
- Rigid expectations
- Difficulty adjusting to unexpected outcomes

Poysky, Hodges, Lotze – unpublished data; Ricotti et al 2015; Conway et al 2015

NEUROBEHAVIORAL DISORDERS IN DMD

- INCREASED RISK OF ANXIETY
- Generalized Anxiety
  - Worrying, stressing, feeling tense, about what might happen or has already happened
    - May seek reassurance from others repetitively
    - Too hard on themselves for mistakes
    - Keep replaying things in their mind
    - Has to know what is going to happen
  - Easily "thrown off", agitated, overwhelmed by change in routine or unexpected events
    - Often presents as anger, criticism of others, arguing
    - Associated with being rigid/stubborn in thought process
  - Changes from day to day

NEUROBEHAVIORAL DISORDERS IN DMD

- INCREASED RISK OF ANXIETY
- Social Anxiety
  - Very shy, uncomfortable around people they don't know well
  - Avoids eye contact, won't say much, may ignore other's when they say "What's up?"
  - Worried about others watching/judging them
  - Afraid they will do/say something embarrassing
  - Avoid/leave situations where they might have to interact
  - Use electronics so they don't have to have conversations

NEUROBEHAVIORAL DISORDERS IN DMD

- INCREASED RISK FOR ANXIETY
- Obsessive-Compulsive Disorder
  - Rituals and excessive routines
  - Very particular about things being even, lined up, etc.
  - Repetitive behaviors
  - Intrusive thoughts/images
  - Too sensitive to how things feel

“HANGRY” = Hungry + Angry

- Blood sugar starts to drop
  - Angry
  - Irrational
  - Mean/aggressive
  - Emotionally sensitive/labile

  - Don’t feel hungry
  - Blood sugar may still technically be in the normal range

COPING WITH DMD

How are the boys coping?

- Same as boys with other chronic medical conditions
  - Being sad and frustrated at times due to DMD is a normal reaction
  - Coping gets better with age
  - Ages 8-10 and adolescence might be extra difficult
  - Some boys may become depressed/distressed

Hendriksen, Poysky, Schrans, Shouten, Aldenkamp, Vles, 2008; Fitzpatrick et al 1986; Liebowitz et al 1981

COPING WITH DMD

- Many boys and young men not as “independent” as they could be

- Increased focus on transition to adulthood
  - Living independently
  - Making decisions in medical care
  - Employment
  - Romantic relationships*

PEER INTERACTIONS IN DMD

Social Problems: 34%

- Social skills weaknesses
- Social anxiety
- Teasing/bullying
- Peer inclusion

Peer relationships decline with age

Hinton, Nereo, Fee, Cyrulnik, 2006;
Hendriksen et al 2008

DEPRESSION

- 24% have “Internalizing” problems (anxiety and depression) – Ricotti et al 2015
- 19% of adult men with DMD report symptoms of depression – Pangalila 2015

SIGNS OF DEPRESSION

- Depression
  - Increased irritability, moodiness
  - Loss of interest in fun activities
  - More withdrawn (socially), wanting to be alone
  - Feeling “blah”, not happy but not sad, “no emotions”
  - More negative about things (everything is terrible, not right, not good enough)
  - Guilty feelings
  - Low self-esteem
  - Crying spells
  - Changes in appetite (more/less)
  - Drop in motivation
  - Changes in sleeping (more/less)

FAMILIES AND DMD

- Family Adjustment
  - Increased rates of parental depression and isolation
  - Behavior problems can be as stressful for parents as physical aspects of DMD
  - Sibling adjustment
- **Stress related to clinical trials**

Abi Daoud, Dooley, Gordon 2004; Bothwell , Dooley , Gordon , MacAuley, Camfield 2002; Poysky & Kinnett , 2009; Nereo, Fee, Hinton, 2003

LEARNING AND BEHAVIOR TREATMENT RECOMMENDATIONS

Bushby K, Finkel R, Birnkrant DJ, Case L, Clemens P, Cripe L, Kaul A, Kinnett K, McDonald C, Pandya S, Poysky J, Shapiro F, Tomezsko J, Constantin C, DMD Care Considerations Working Group. **The diagnosis and management of Duchenne muscular dystrophy – part 1. Diagnosis, pharmacological and psychosocial management.** The Lancet Neurology 2010;9(1):77-93.

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. **Diagnosis and management of Duchenne muscular dystrophy, part 3: primary care, emergency management, psychosocial care, and transitions of care across the lifespan** The Lancet Neurology Published online: January 23, 2018

Colvin MK, Poysky J, Kinnett K, Damiani M, Gibbons M, Hoskin J, Moreland S, Trout CJ, Weidner N. **Psychosocial Management of the Patient with Duchenne Muscular Dystrophy** Pediatrics 2018 12(s2) – In Press

TREATMENT RECOMMENDATIONS

- Effective treatments! Same as for people without DMD
- Don’t wait if you have concerns
- Mental health professional
  - Does not need to be “expert” in DMD
  - Willing to learn from, and listen to, patient, parents and other professionals
  - It is helpful if they have worked with other medical conditions

- **Evaluations**
  - Neuropsychologist
  - School Psychologist
  - Special Education Case Manager
  - Psychiatrist
- There may be more than one problem going on
- Don't wait to see if he/she will "grow out of it"

LEARNING PROBLEMS: WHAT CAN I DO ABOUT IT?

PREVENTION (so it doesn't become a problem)

INTERVENTION (trying to fix the problem)

ACCOMMODATION (minimizing the impact)

MODIFICATION (changing teaching and/or evaluation methods)

NEUROBEHAVIORAL TREATMENT RECOMMENDATIONS

Psychotherapy

- **Parental behavior management training**
  - Noncompliance, disruptive behavior, temper meltdowns
- **Individual therapy**
  - Low self-esteem and depression, anxiety, obsessive-compulsive disorder, coping
- **Group therapy**
  - Social skills deficits
- **Applied Behavior Analysis**
  - Autism

TREATMENT RECOMMENDATIONS

Psychiatric Medication

- Remember, it is neurological!
  - Stimulants or alpha-agonists for ADHD
  - SSRI's for anxiety, depression, emotional reactivity

Ritalin + Prozac combination becoming more common

ADDITIONAL RESOURCES:

treat-nmd.eu
TREAT NMD: Family Care Guidelines

parentprojectmd.org
"DMD Learning and Behavior Guide" (Poysky)
"Psychology of Duchenne" (Hendriksen)

A Guide to Duchenne Muscular Dystrophy: Information and Advice for Teachers and Parents -Janet Hoskin – Editor. (Amazon)

The Explosive Child – Ross Greene