

Duchenne, ADHD, Dyslexia and Autism

Action Duchenne
It's Time to Stop Wasting

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How is DMD related to other disorders?

- DMD is diagnosed by its underlying cause
 - A genetic mutation that results in no dystrophin.
 - Person develops without dystrophin
 - Can affect many things including muscle and BRAIN
- ADHD, Dyslexia and Autism are diagnosed by the behaviors observed
 - Each one is defined by a cluster of symptoms
 - The cause is unknown and there may be different causes that present with the same symptoms

How are they related?

- Having DMD does NOT = having ADHD!
- Having DMD does NOT = having dyslexia!
- Having DMD does NOT = having autism!

- More people with DMD have each of these three disorders than is found among the general population

- Having DMD means there is an **INCREASED RISK** for having any of these diagnoses

How to determine?

- So, most children with DMD will NOT have these disorders
- Nonetheless, these behaviors are observed more often in the group and need to be considered
- ADHD, Dyslexia & Autism are diagnosed based on **observable behaviors**
 - Evaluate
 - Observe
 - Neuropsychological Testing
 - School Reports
 - Peer Interactions

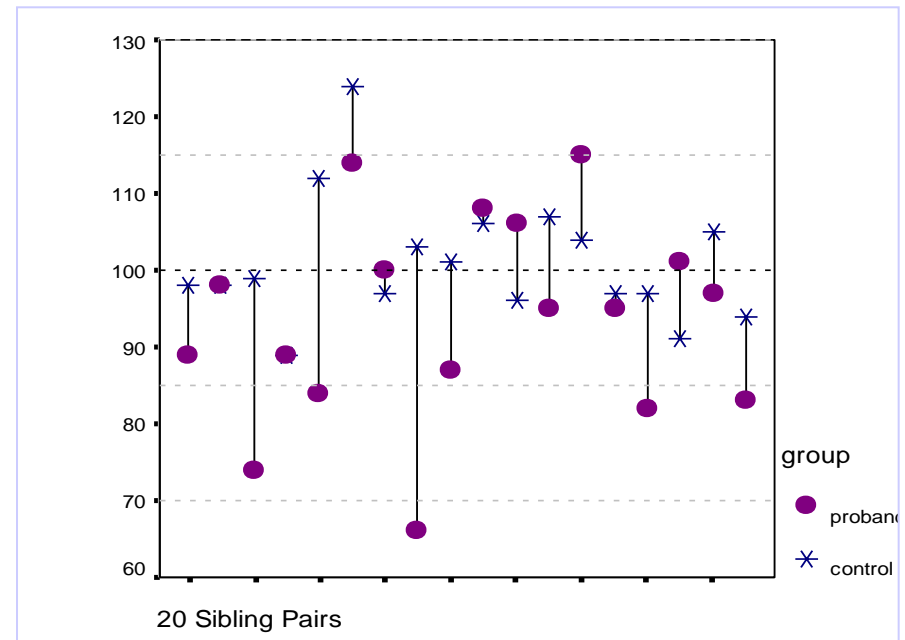
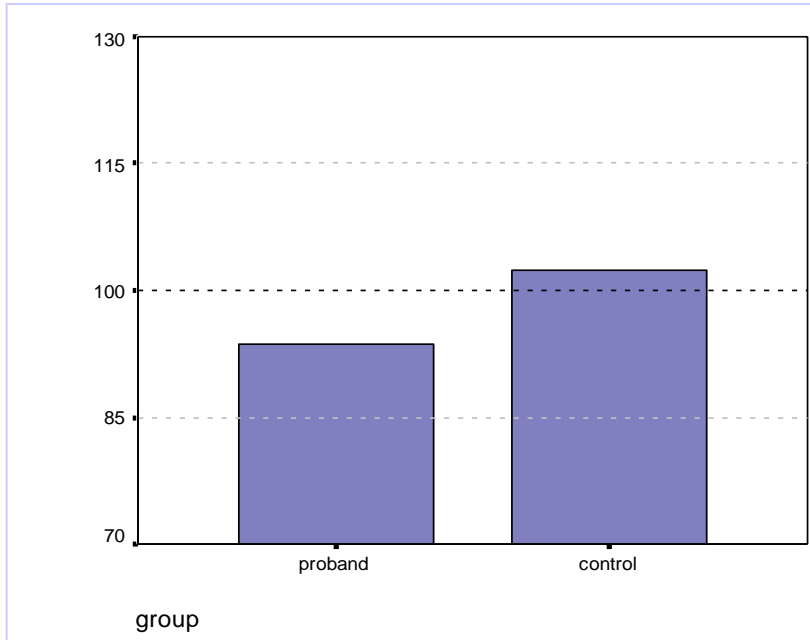
Developmental Dyslexia

- Reading ability is significantly lower than expected given child's intellect and other skills
- Difficulty learning to read; this can look different at different developmental time points
 - Delayed speech
 - Many sound substitutions in speech that persist (e.g., “fumb” for “thumb”)
 - Difficulty repeating speech sounds
 - Difficulty associating sounds with letters
 - Difficulty blending sounds together and “sounding out” written words
 - Difficulty becoming adept or automatic in reading (it remains effortful even after phonics are mastered) – slow readers

Research in Reading and DMD

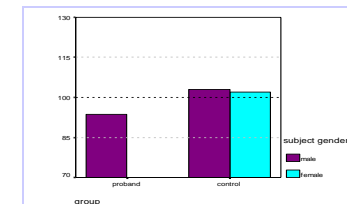
- Numerous studies have shown that reading skills are lower in boys with DMD as a group
 - When compared to children of same age
 - When compared to children with a different neuromuscular disorder
 - When compared to unaffected siblings
 - When compared to general intellectual level
- However, a diagnosis of developmental dyslexia must be based on an **individual's** performance
- **Each** child must be evaluated on the basis of **his own** performance and behavior

Boys with DMD have low reading scores



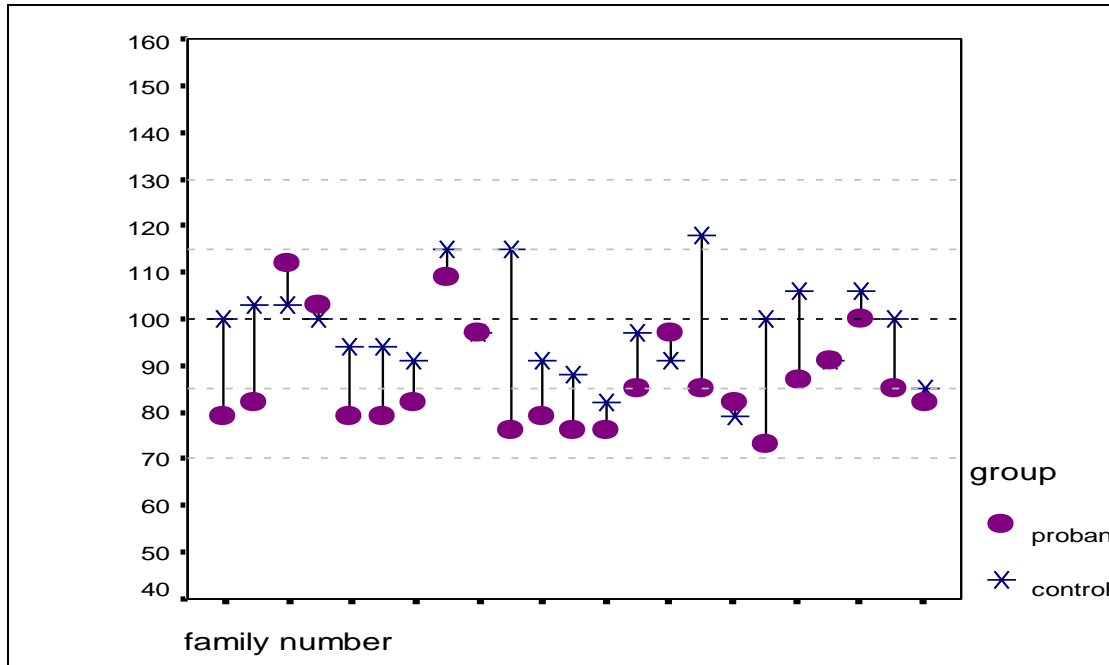
Even though group mean performance on WJ Reading Composite is within the low average range, boys with DMD perform significantly worse than their siblings.

DMD = 94 + 13, Controls = 102 + 9, $t = 2.5$, $p = .017$

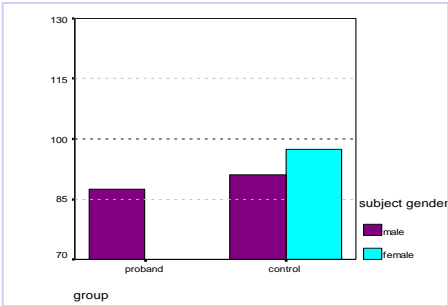


No gender differences

Boys with DMD do significantly more poorly than their siblings on tests of Phonological Awareness.

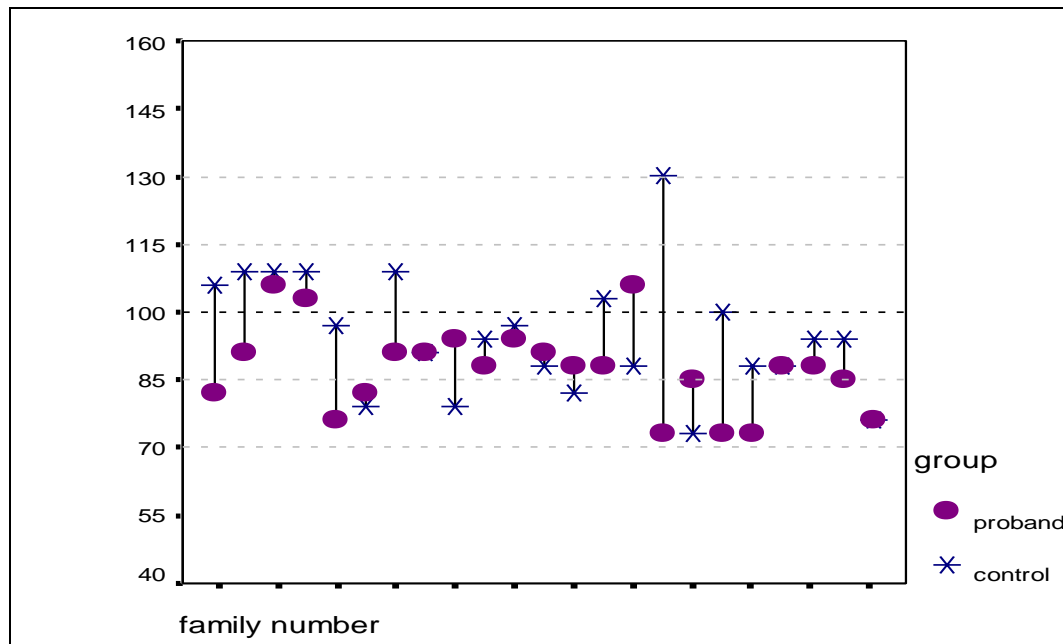


Mean + sd :
 DMD = 87 + 11, Siblings = 98 + 10 t = 3.47, p= .001



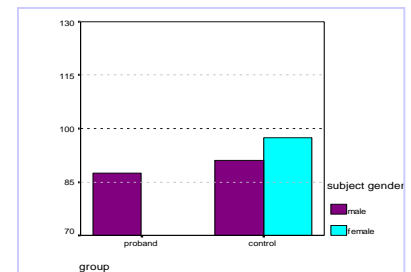
No gender differences were observed

Boys with DMD do significantly more poorly than their siblings on tests of Phonological Memory



Mean + sd :

DMD = 87 + 10, Siblings = 95 + 13, $t = 2.16$, $p = .036$



No gender differences were observed

- Even without evidence of “reading disability” boys with DMD tend to be weaker on the PHONOLOGICAL PROCESSING skills necessary for good reading acquisition
- Be aware of this possibility
- If concerned, have the child evaluated
- Treat the problem as dyslexia (not anything special for DMD)
- Interventions for improving reading are available
 - ask Janet Hoskin!

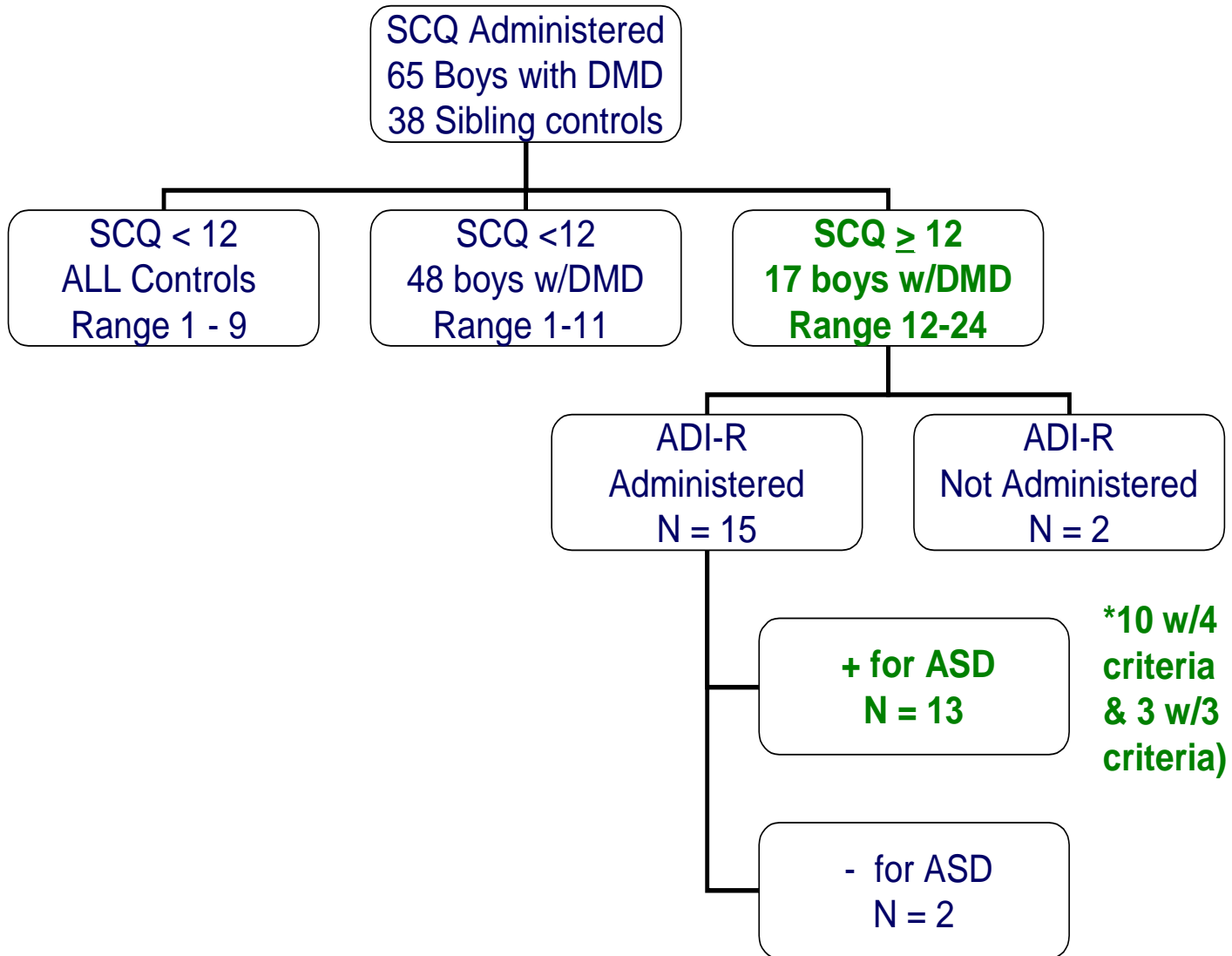
Autistic Spectrum Disorder

- Qualitative abnormalities in reciprocal social interactions
- Qualitative abnormalities in communication
- Restricted, repetitive or stereotyped behaviors
- Onset before age 3
- May look different at different developmental stages
 - Delayed speech
 - Lack of social gestures (e.g. waving “bye bye”)
 - Lack of sharing interests with others (e.g., no “look at me! Look at me!”)
 - Circumscribed interests (knows ALL about the underground system)

Research in Autism and DMD

- Case studies observing children with DMD & Autism
- Report showing that in a clinic in Boston DMD and autism occur together with greater than random frequency
- Studies showing increased social problems in DMD
- Study showing decreased affect recognition in DMD
- Studies showing decreased executive functions in DMD

- However, a diagnosis of Autism Spectrum Disorder must be based on an **individual's** performance
- **Each** child must be evaluated on the basis of **his own** performance and behavior



- 65 Probands
 - Range of scores 1-23
 - 12/58 \geq 15 (21%) – suspected autism
 - 19/58 \geq 12 (33%) – suspected pervasive developmental disorder
- 38 Controls
 - Range of scores 1-9
 - 0 fall in suspected autism range
- Parents of children with suspected autism spectrum disorder given a lengthy interview, the Autism Diagnostic Interview– Revised (ADI-R), the “gold standard” for diagnosis

- Results

- Higher than expected number of children
 - 15-19% of children with DMD/BMD + for ASD
 - 10 cases out of 65 met all criteria
 - 3 more met $\frac{3}{4}$ criteria
 - Selection bias?
 - There are other case reports in literature

Description of ASD Qualities in DMD

- Qualitative abnormalities in reciprocal social interactions that improve with age
 - Failure to use nonverbal behaviors for social interactions (poor eye contact, lack of social smiling)
 - Failure to develop peer relationships (lack of interest in children)
 - Lack of shared enjoyment (not directing other's attention to things of interest)
- Qualitative abnormalities in communication that improve with age
 - Delay in spoken language
 - Lack of varied, spontaneous make-believe play
 - Lack of reciprocal conversation

Description of ASD Qualities in DMD

(continued)

- Restricted, repetitive and stereotyped patterns of behavior in some children, but this is not the predominant characteristic
 - Circumscribed pattern of interest
 - Compulsive adherence to rituals
- No evidence of regression
- No evidence of unusual sensory interests
- No evidence of repetitive motor mannerisms
- No evidence of savant qualities
- Intellectual function within normal limits

How to put it all together?

- DMD affects development of brain structures – including cerebellum and cerebral cortex
- Without dystrophin in these areas, synaptic function may be impaired --- this may impact on verbal span --
- maybe the amount of verbal information that can be processed at any one time is a little less than that of people with dystrophin
- Additionally, without dystrophin, brain areas may be less resilient to environmental influences Brain may be at increased risk for damage after minor insults, such as hypoxia ---- this may account for the great range of impairment

- If brains lacking dystrophin hold less verbal information than expected, language acquisition will develop more slowly
 - Understanding of complex language will be particularly affected
 - This may impact on development of social skills too
 - This may interfere with phonological processing
 - Perhaps there is less space in the phonological loop?
 - It will impede reading acquisition
- Cerebellar involvement may suggest slowed learning of automatic processes

What to do?

- Be AWARE of the potential problems
 - Increased risk of developmental dyslexia and autistic spectrum disorders
- ASK if the child has any language, academic or behavioral difficulties
- Refer the child for neuropsychological testing
 - if there are any concerns
 - prior to starting school
- Get interventions/treatments as needed
 - Although there are no intervention studies in this group to offer evidence-based suggestions, (yet! Janet Hoskin is doing one!) targeting specific areas found to be weak will be helpful.
 - There are good interventions for dyslexia and autistic spectrum disorders
 - Always emphasize strengths!
- These are all things that can be improved and that impact on the child's, and the family's, life
 - Children with unaddressed cognitive problems often develop low self esteem, anxiety and depression



Let's ensure the best possible quality of life for all of these boys



Acknowledgments

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ADHD and Duchenne Muscular Dystrophy

Action Duchenne Annual Conference
November 12, 2010

James Poysky, PhD
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Objectives

- Describe ADHD and what it looks like.
- Examine the relationship between ADHD and DMD.
- Discuss clinical care of ADHD in DMD.

What is ADHD?

Prevalence in general population

- UK: 1%
- USA: 3-10%
 - 4 to 5 million school-aged children
 - Under diagnosed in low SES
 - Over diagnosed in high SES
- 2.1% of population on active medication treatment
- 2:1 male to female ratio (closer to 3:1 in UK)

Practice Parameter for the Assessment and Treatment of Children and Adolescents With Attention-Deficit/Hyperactivity Disorder, AACAP (2007)

Clinical Practice Guideline: Diagnosis and Evaluation of the Child With Attention-Deficit/Hyperactivity Disorder, AAP (2000)

National Clinical Practice Guideline #72, GDG/NICE (2008)

What is ADHD?

- Diagnostic and Statistical Manual (DSM-IV)
 - Symptoms of inattention and/or impulsivity and hyperactivity
 - Excessive for age
 - Present before 7 years old
 - Persistent across time
 - Pervasive across settings
 - Not explained better by other disorder
 - Causes impairment in multiple areas

What is ADHD? Inattention

- Focus
 - Careless mistakes, misses details
 - Easily distracted, needs redirection
 - Does not seem to listen, follow instructions
 - Daydreams
- Planning/Organization
 - Messy
 - Loses important belongings
 - Poor time management
 - Difficulty planning ahead
 - Difficulty prioritizing, “multitasking”
 - Problems sequencing multi-step tasks
- Memory
 - Forgetful, absent-minded
 - Poor short-term memory
- Motivation
 - Avoids mental effort, procrastinates
 - Difficulty connecting hard work with long-term rewards
 - Overly attracted to fun things



What is ADHD?

Hyperactivity/Impulsivity

- Impulsivity
 - Acts/speaks without thinking
 - Impatient
 - Interrupts/intrudes on others
 - Blurts things out and/or is overly loud
 - Talks excessively or at the wrong times
 - Acts silly or “over the top” inappropriately
 - Hard time stopping or changing a behavior despite feedback from others
 - Hard time keeping hands to self
- Hyperactivity
 - Fidgets with hands/feet
 - Difficulty playing quietly
 - **Runs or climbs excessively**
 - **On the go or driven, “busy”**
 - **Often leaves seat**



What is ADHD? Subtypes

- Inattentive Type
 - Primarily attention problems
 - Often overlooked
 - Diagnosed later , mid-elementary
 - Sluggish cognitive “tempo”
- Combined Type
 - Symptoms of both inattention and hyperactivity/impulsivity
- Hyperactive/Impulsive Type
 - Classic type
 - Easily recognized
 - Diagnosed early, preschool
 - Early hyperactivity
 - Persistent impulsivity

What is ADHD? Impact

- ADHD symptoms can interfere with:
 - Academic progress
 - Missing information
 - Not completing work
 - Interactions with peers
 - Making/keeping friendships
 - Targeted for bullying
 - Home life
 - Parent/family stress
 - Conflict
 - Self-esteem
 - “Always in trouble” no matter how hard he tries
 - Attributed to character flaws: immature, lazy, kid is a “brat”, “just doesn’t care”

ADHD and DMD

- Approximately 12-30% of boys with DMD may have symptoms of ADHD

All patients seen in our clinic (5% refused participation):

- Hyperactive/Impulsive and Combined = 31%
- Inattentive = 4%
- Oppositional, argumentative, explosive behavior = 52%
- Medication may have had an impact on behavior, but no statistical correlation

Hendriksen & Vles (2008); Hinton et al. (2006); Poysky & Lotze (unpublished data)

ADHD and DMD

- In DMD, kids with Dx of ADHD have poorer psychosocial adjustment than those without ADHD:
 - Peer relations
 - Dependency
 - Hostility
 - Productivity
 - Anxiety/depression
 - Social withdrawal

(Hendriksen & Poysky 2006 – unpublished data)

*This does not necessarily mean that AD/HD is the *cause* of poor psychosocial adjustment in DMD

ADHD and DMD: Clinical Care

- Pliszka S and AACAP Work Group on Quality Issues. Practice Parameter for the Assessment and Treatment of Children and Adolescents With Attention-Deficit/Hyperactivity Disorder. J. AM. ACAD. CHILD ADOLESC. PSYCHIATRY, 46:7, JULY 2007:894-921.
- American Academy of Pediatrics. Clinical Practice Guideline: Diagnosis and Evaluation of the Child With Attention-Deficit/Hyperactivity Disorder. PEDIATRICS Vol. 105 No. 5 May 2000:1158-70.
- 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. Lancet Neurol 2010;9(1):77-93.

ADHD and DMD: Clinical Care

- Making a Diagnosis:
 - Clinical interview to collect information:
 - History of the symptoms and level of impairment
 - Patient's developmental, medical history
 - Family history
 - Other problems (comorbid conditions)
 - Medical tests to rule out potential medical condition (e.g., thyroid problems), if indicated
 - Other sources of information (if available)
 - Teacher narratives, letters, reports
 - Standardized rating scales (have to be specific to ADHD, not global measures of behavior)
 - Research does not support use of medical/neurological tests (e.g., EEG, MRI) or neuropsychological testing (e.g., continuous performance tests) in making ADHD diagnosis
 - Psychological/neuropsychological testing can help identify comorbid conditions

ADHD and DMD: Clinical Care

- Psychotherapy
 - Parent Training
 - Information about ADHD
 - Appropriate use of reward strategies and time outs
 - Manage behavior in public settings
 - Anticipate future behaviors
 - Daily school behavior note/chart
 - Group Therapy
 - Social skills training for kids with ADHD
- Therapy is the “first line” treatment for mild cases of ADHD

ADHD and DMD: Clinical Care

- Medication
 - Most effective form of treatment
 - Approx 85% of kids with ADHD get benefit from medication
 - Methylphenidate most commonly used
 - Side effects common, but generally well-tolerated
 - Consult with cardiologist first, monitor status
- Medication is “first line” treatment for moderate to severe cases of ADHD

ADHD and DMD: Clinical Care

- Accommodations at school to reduce the impact of ADHD on academic progress:
 - Extra time for tests and assignments
 - Quiet test-taking environment (another room)
 - Reduced amount of homework
 - Preferential seating
 - Give directions one step at a time
 - Not penalized if forgets to turn work in to teacher
 - Teacher's aide

ADHD and DMD

Thanks for paying attention!!!

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