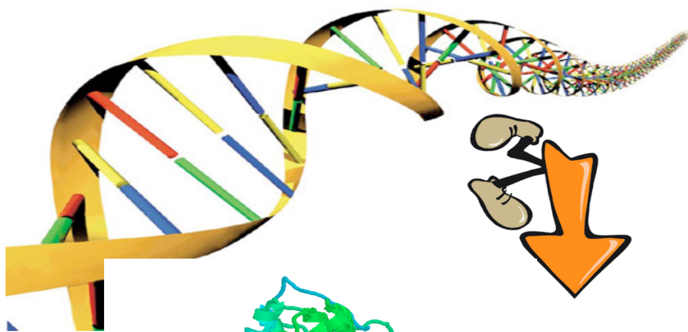
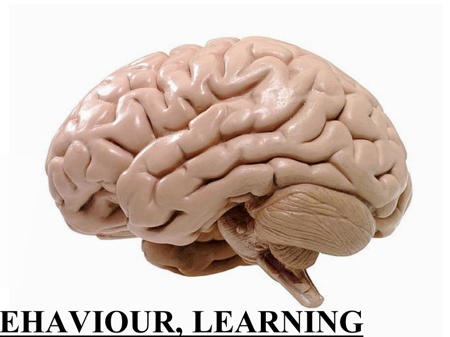
Psychological aspects involved in Duchenne Muscular Dystrophy
A guideline for parents and teachers

Gene

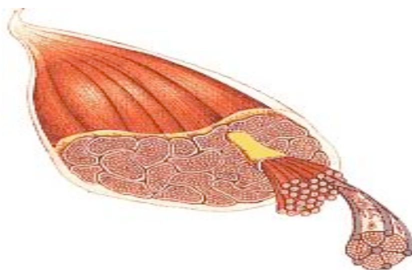


Protein(dystrophin)

Brain



<u>COGNITION, BEHAVIOUR, LEARNING</u>



<u>AND PHYSICAL FUNCTIONING</u>

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General introduction:

In contrast to the muscles and it's degeneration, who play a crucial role in Duchenne Muscular Dystrophy, there has not been written a lot about the psychological aspects of this disorder. Since the last decade scientific research on psychological aspects has been performed: initially on intelligence and learning, since five years also on behaviour and neuropsychiatry. Knowledge of these psychological aspects is of utmost importance: early detection can help ameliorating early intervention. Good and practical information is not always easily accessible to parents, caregivers and teachers. Therefore these guidelines were developed by the Dutch Parent Project Muscular dystrophy (DPP NL)- and reworked to the English language. We plan to give an understandable and practical discussion on a diversity of relevant psychological aspects that all have to do with DMD and hope it contains relevant information about psychology in DMD for parents and teachers. These guidelines consist of three parts. The first chapter deals with cognition, intelligence and reading inboys with DMD. The second chapter gives information about psychosocial adjustment involved in DMD. And the last and third chapter provides you with the information about the neuropsychiatric disorders that sometimes appear in (a subset of) DMD patients.

We hope that these guideline assist you in a better understanding of Duchenne and it's psychological aspects and dealing with them.

We wish you all the best and are convinced that your boy with DMD is a winner with his own unbelievable strengths.

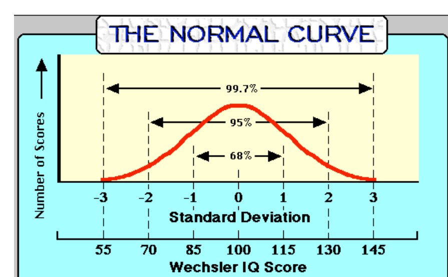
Chapter 1: Cognitive functioning in Duchenne Muscular Dystrophy.

1.1 Introduction:

First of all it is important to realise that, from a psychological point of view, the average boy with Duchenne Muscular Dystrophy does not exist. Every boy is unique with his own specific strengths and weaknesses. Boys with DMD often show, like every other child, a profile of specific strengths and deficits in their cognitive functions. Not every boy with DMD has to deal with cognitive disabilities, but the risk is higher than average.

We will first discuss about intelligence in DMD and subsequently inform you about the specific cognitive profile of strengths and weaknesses. Thirdly we will discuss the reading problems in boys with DMD. Finally we give some information about what parents and teachers can do in dealing with the reading difficulties.

Figure 1: Normal Distribution of intelligence scores. Note that an IQ between 85 and 115 is normal.



1.2 Intelligence in boys with DMD:

The average Full-Scale IQ (Intelligence Quotient) in general population is 100 with a normal range from 85 to 115 (figure 1). On the basis of an extensive study from Sue Cotton it can be concluded that boys with DMD have a low average mean IQ score of 80. This implies that 30% of the boys with DMD have an IQ score which is lower than 70. It can be concluded that boys with DMD have a mild cognitive impairment and that their IQ scores show a considerable heterogeneity with full scale IQ's ranging from a minimum of 14 to a maximum of 134.

It has been shown that Verbal IQ (language reasoning) is somewhat lower than Performance IQ (visual reasoning). Furthermore it has been shown that in younger children (till 9 years) there is a larger discrepancy between Verbal IQ and Performance IQ. As boys grow older the discrepancy diminishes and Verbal IQ seems to improve with age.

To summarize: boys with DMD show a low average Full-Scale IQ level. Unlike the progressive physical decline it is important to realise that there is no progressive decline in IQ. Actually, it looks like language thinking (Verbal IQ) is showing some improvement during age.

1.3 Cognitive strengths and weaknesses in boys with DMD:

The cognitive problems mentioned before are probably caused by the brain's lack of dystrophin. Dystrophin is as we know from recent research, also located in some specific areas of the brain: (1) the hippocampus, responsible for memory, (2) the cerebellum, which plays a crucial role in automatisation and (3) the prefrontal cerebral cortex responsible for planning and organisation (see figure 2). In DMD the dystrophin may be missing in these areas of the brain, and therefore probably causing the cognitive problems.

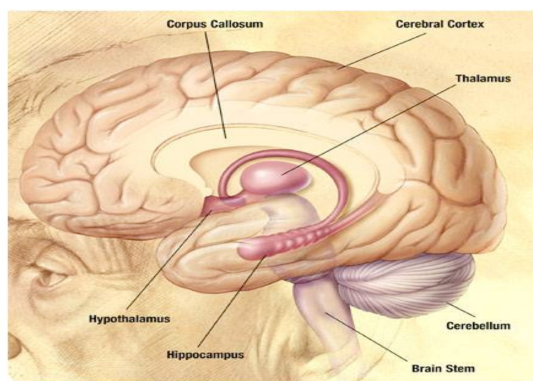


Figure 2: Brain areas containing dystrophin.

Language thinking proves to be especially at risk. Common problems that may occur are: late onset of speaking, word finding problems and influent speech. All of these are problems that have to do with language expression. It has been shown that in 40% of DMD boys without a family history of Duchenne, language problems manifest itself even before the onset of muscle weakness.

Another important cognitive problem involved in DMD is the short-term memory. This weakness has to do with the processing of recently heard oral information. One should remember that boys with DMD who may have a weak short-term memory only get a part of the message.

Attention is the third cognitive function that might be a weakness in boys with DMD. In one of our own studies we found that in 18,7 percent of boys with DMD parents reported attention problems in comparison to 7,5 percent in a control group of siblings. These concentration problems could be bothering boys with DMD especially at school. Overall it was shown that attention problems seem to diminish as the boys with DMD grow older.

Despite all the mentioned weaknesses in the previous paragraph, boys with DMD, also have cognitive strengths. Especially in visuospatial functions and the learning of rote material they may function above average. To summarise: the cognitive profile of boys with DMD:

Weaknesses	Strengths
Expressive language	Visuospatial capacities
Working memory	Rote memory
Attention	

1.4 Reading in boys with DMD:

Dyslexia is a learning disability that manifests primarily as a difficulty with written language, in particular with reading and spelling.

Boys with DMD are at increased risk for reading problems. Reading has to do with automatisation as one of the core cognitive functions. Beginning readers must learn to recognise the distinct visual features of different letters and must learn which sound (phoneme) corresponds with a specific letter. Elementary reading is completed when the child quickly and correctly decodes letters in sounds and subsequently in words.

Reading is therefore the automatisation of the decoding skills. In the Netherlands we performed an extensive study on reading in 25 DMD boys. It was found that twenty percent of these boys manifested serious reading problems; another twenty percent were having moderate reading problems. In total we found forty percent of the DMD boys having difficulty with learning to read. Note that in the general population around five percent of the children has reading problems.

In both reading and dyslexia the cerebellum (responsible for automatisation, which is important in reading) is assumed to play an important role. An, already mentioned, neurobiological mechanism is assumed: the dystrophin deficiency in the brain and especially in the cerebellum (see figure 2 again). Especially for boys with DMD early detection and prevention are therefore important as they rely more and more on communication by written words as they grow older.

1.5 How to deal with the reading difficulties?

In general, early detection and intervention for reading disabilities has been shown to be the most effective strategy of remediation in children with a higher risk for dyslexia on the basis of parental inheritance. For DMD boys we therefore suggest systematic screening at age 4 years to detect whether they are at risk for later reading problems. Screening should be aimed at sequential information processing and at phonemic awareness because these aspects are most likely to play a crucial role in later reading acquisition. Furthermore is it advised to get a psychological assessment of the boy's cognitive strengths and weaknesses.

Secondly, phonemic awareness (the awareness that words are made up of sounds), which is an important fundament of learning to read, should be trained. This can be done by formal reading instruction. But one can also start early with listening games, rhymes and ditties, playing with phrases and words, discovering the initial sounds of words and so on. Phonemic awareness can be trained in preschool programs and such training programs have a beneficial effect on later reading and prevention of reading problems. It is important to keep in mind that learning to read already begins before the first day in school. The best treatment

for reading problems is therefore starting as early as possible with reading, also called "emerging literacy". By training phonological awareness one can make an easier route into learning to read. Parents can in fact contribute significantly. Furthermore reading aloud also helps and has potential benefits:

- The boy establishes more precise phonological representations of words;
- The boy develops a richer vocabulary;
- They develop a lasting interest for reading.

Finally, it is important to enhance pleasure in learning to read and reading. Reading together should be fun and this works very stimulating,

Chapter 2: Psychosocial adjustment in Duchenne Muscular Dystrophy.

2.1 Introduction:

Research has shown that children with physical handicaps are at an increased risk of experiencing emotional or behavioural problems due to psychosocial adjustment. Around 20% of the children with a chronic illness and 30% of the children with DMD, will experience significant behavioural or emotional problems. This is a rate which is twice as high as that for physically healthy children. In some DMD boys, moreover, steroid medication may cause an increase in behavioural difficulties and outbursts. In this chapter we would therefore give some guidelines and strategies in dealing with the emotions and behaviour of your DMD boy. Finally we will give some guidelines to talk with your boy about DMD. But first of all we will start this chapter with discussion on psychosocial adjustment itself in DMD and the aspect of learned helplessness.

2.2 Psychosocial adjustment:

Psychological adjustment reflects the ways in which people psychologically adjust to difficult and stressful situations. Psychological adjustment has to do with coping strategies. In one of our research projects (Hendriksen, Poysky et al 2009) we found that 17% of the DMD boys had low- adjustment scores; in normal control group 2% had low adjustment scores. Nevertheless, 83 % of the Duchenne boys were reported to have adequate psychosocial adjustment.

This research also revealed that adjustment scores become better as boys grow older. The age-period from 8 till 10 year is characterized by significant lower adjustment scores. Clinically this is the age period when boys with DMD start realizing on the implications of their handicap and when walking becomes difficult.

There are six domains of psychosocial adjustment: peer relations, dependency, hostility, productivity, anxiety/depression and withdrawal. In the next table these domains are illustrated by an example:

Domain of psychosocial adjustment	Behaviour reflecting this domain
Peer relations	making friends without difficulty
Dependency	asking for help or being unable to decide things for himself.
Hostility	ignored warnings to stop unacceptable behaviour/ not responding to discipline
Productivity	working without being pushed or punished/ keeping on working when difficult

Anxiety/depression	saying that people do not care about him/saying that he couldn't do things right.
Withdrawal	acting afraid or apprehensive/ staring without doing anything

It has been shown that peer relations may become worse as boys grow older. Parents and teachers should take care to stimulate this area of adjustment as we know from other research that social support is an important protective factor in dealing with stress. Anxiety/depression and withdrawal also remain important aspects of follow-up as they remain stable over time. Dependency, hostility and productivity become better as boys grow older.

2.3 Learned helplessness:

Environments in which people experience events in which they have no control over what happens to them, may tend to foster learned helplessness. This model is of great relevance for chronical ill children and especially for children with a progressive disease, like DMD. It is important that parents and teachers are aware of this mechanism. A boy becomes learned helpless when he does a lot of effort without any subjectively experienced result (action-effect chains). For example doing intensive physical training and in the meanwhile experiencing physical detoriation. It is important to take care of what we call "action- effect chains": parents, teachers and therapists should always be aware of this mechanism: make actions of the boys effective. This refers to sense of control: having some control over a situation or a stressor (whether real control or imaginary control) will lessen the degree of impact on you by that stressor. Physical training for instance should always be followed by a tangible effect.

2.4 Guidelines and strategies to deal with emotional problems:

Emotional problems are an essential part of a normal and healthy psychological development. Growing up implies coping with normal developmental stressors (e.g. every child has to deal with his first school day or his first bad grade at school). For boys with Duchenne there are two periods which are known to be important: first there is the period of anxiousness in which they start to fall and won't be able to get up on their own power. Secondly there is a period of stress when the boys become wheelchair dependent. They can react with hostility or depression because of the loss of independence. One should alway keep in mind that there often is a reason for an emotional outburst, taking in account the illness related stressors the boys are confronted with and have to deal with.

Always keep in mind that child development experts advice that children with disabilities should be raised the same way other children of their age are treated. One important strategy for enhancing esteem in your boy is positive parenting. This implies that parents teach their child how to deal with stressful and emotional issues. Research has shown that emotion coaching is an important strategy and can help in building up positive self-esteem in your boy. It is important to be a coach of your child's emotions. It is also important to be consistent in the way you are dealing with you son. There are five essential steps for emotion coaching:

1. Emotional awareness: always try to be aware off what your child feels and acknowledge their anger, sadness or fear. Remember that awareness is the key to change.
2. Moments of learning: recognise the emotions as an opportunity for teaching. Negative experiences can serve as an opportunity of learning your child how to handle these feelings. Reasoning away your child's emotion with logic on the contrary rarely works.
3. Listening: empathic listening is the most important step in the emotion-coaching process. Empathic listening means in fact turning yourself in to your child's

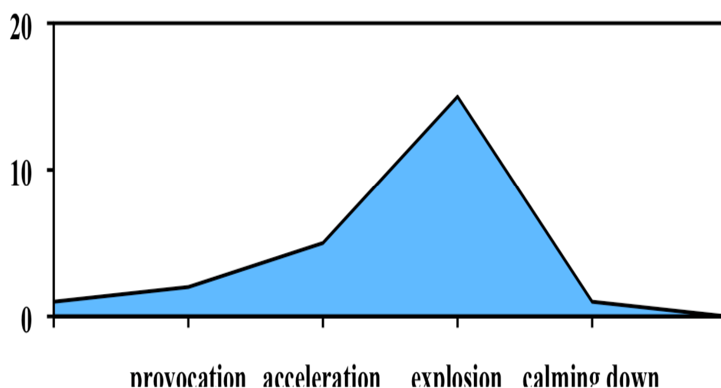
emotions. What does his body language tells you about the way he is feeling? If you want to understand your child you have to put yourself in his shoes.

4. Labelling: Help your child verbally labelling his emotions. Help you child labelling their emotions by giving it a name as they are having it. For example: ‘I can see that you are angry/sad’. Labelling goes hand in hand with empathy. Studies indicate that the act of labelling emotions helps children to recover more quickly form stressful incidents. If you observe that your child seems to be angry or disappointed you can help him to transform a scary, uncomfortable feeling into something identifiable and normal. Putting a name to the emotion not only helps children make sense of what they are feeling. Research studies suggest that it also helps calm their nervous systems and helps them recover faster from upsetting situations.
5. Problem solving and setting the limits. Once you have spent time listening to your child and helping him to label and understand his emotions, you will find yourself solving the problem. Work out possible solutions together with your child, evaluate them and help you child to choose a solution. Remember to set limits at this stage. Let your child understand that their feelings are not the problem but that their misbehaviour is. For example you can tell your boy that he may be angry at his brother, but that he may not tell naughty things about him. Feelings are acceptable, but not all behaviours are. Therefore set limits.

Emotion coaching is an important and easy way to help your boy coping with his emotions. It is a sort of basic attitude in parenting your child. Be aware of the strength of this strategy because it does improve your communication with your boy and build up his self-esteem.

Do NOT use the emotion coaching in all situations. Don’t use emotion coaching when: (1) you have no time, (2) others are present, (3) you are upset or tired, (4) you have to correct your child or set boundaries and discipline your child and (5) your child plays as if he is emotional.

Figure 1: The sequence of a tempur tantrum:



2.5 How to deal with behavioural problems:

Outbursts and tantrums are an important part of the behavioural problems. A tantrum is attention-getting behaviour and often unwanted by the parent. Children have tantrums because they want your attention. It does not matter if the attention is positive or negative. They just want 100 percent of it. So if you lose your control and have a tantrum yourself, you give them what you want. It’s best to ignore a tantrum. But that is not always possible. Every

tantrum is characterised by a sequence of (1) a provocation, (2) an acceleration, (3) the explosion and finally (4) the calming down.

During the phase of provocation and acceleration you should stick to a clear NO. Remember that during the phase of explosion it is difficult to react. After they have quieten down you may be able to negotiate with them by saying: ”I noticed your behaviour, but that won’t help in getting my attention. You need to use your words to get my attention”. Be consistent in dealing with these situations, and take care that both parents and other caregivers react the same!

You can use a time-out to help your child calming down. A time-out should take no more than a few minutes and helps him to cool down, preferably in a boring place. This is namely kind of disciplining your child. However the best way to discipline your child is taking away a privilege or something positive.

Remember eventually that positive feedback works the best: so reinforcement of positive behaviour is the best strategy: e.g. complementing your child when he succeeded in cooling down himself after a tantrum. Please also keep in mind that tantrums usually are not a reason for concern and they usually diminish on their own.

2.6 When to see a specialist?

We recommend consulting a specialist (e.g. child psychologist or paediatrician) if:

- The tantrums and outbursts are more frequent and longer lasting than anticipated;
- If you are uncomfortable with your reactions;
- If you keep giving up;
- If the tantrums cause a lot of bad feelings and family distress;
- If your boy frequently hurts himself or other persons;
- If you boy is destructive.

Finally, it might be necessary to check for medical problems that might contribute to tantrums.

2.7 Talking about DMD:

Emotions may arise when you (as parent or perhaps as teacher) are going to talk about things like the circumstances, DMD itself and the future. Telling children about their neuromuscular disease is a necessary but delicate conversation. Parents rarely seem to feel confident going in to these conversations. For parents it is important to set the time when to tell your boy about Duchenne.

One study on the worries of parents of boys with DMD in 1983 showed that parents worry most about when to tell what to their child . One can not give one good advice because it depends upon several factors. We will try to give you some guidelines (See Thompson, 1999). Do not tell your child about the muscle disease, but instead set the stage! Go for instance to a somewhat quiet place and create openings for a conversation, like: “you will be wondering about all those tests”. After this you should remain silent, listen to your child and answer the questions that come up.

How to react on questions, depends upon the illness perception of your boy: how does the child look at his disorder? Age is an important factor in this. The young child (younger than seven years), for example, is egocentric in his thinking. DMD is just one aspect of his life and he doesn’t realise the consequences of the disorder yet. The young child is thus focussed on the here and now. But parents, on the other hand, are focussed on the future. The young child doesn’t think about this because he just wants to play football, but his legs are naughty. As a parent one should therefore always react in the here and now: give emotional support and realise that the boy doesn’t want an answer. When boys grow older, they become less egocentric and they will also (like their parents) start thinking about the future. When boys with DMD are getting older, brief occasional talks can help a lot in talking with them about the several aspects of DMD. Some guidelines can be given:

- Remember to be honest but don’t talk too much: just provide your boy with the information he is asking for and not more; It might be reassuring to know that children don’t ask for more information than they can deal and cope with;
- Provide reassurance: e.g. “you are going to get what you need and we have all these doctors that are going to help us”;
- Anger, grief and other emotions are reasonable reactions to stressors. Don’t try these feelings to go away; show understanding and emotion coaching instead;
- If children don’t ask about the future don’t bring it on. If they do ask, answer honestly but keep the door open to possibilities: e.g. “We’ll do all the things that we can when things happen”

Chapter 3: Neuropsychiatric disorders in Duchenne Muscular Dystrophy.

In raising, educating and training a boy with DMD one might see behaviour problems that are associated with common neuropsychiatric disorders. This topic has come to interest of scientists and clinicians only since the last few years. In this chapter we shortly discuss the three neuropsychiatric disorders that may co-occur with DMD. It is important that parents, teachers, therapists are aware of this comorbidity, as it helps in dealing with the behaviour problems.

3.1 Autism spectrum disorders (ASD):

People with an Autism Spectrum Disorder have a triad of impairments in socialisation, communication (verbal as well as non-verbal) and imagination and behaviour. In ASD there are in general three problems:
- A failure to develop a relationship, eye contact or shared enjoyment;
- Delay in spoken language, idiosyncratic speech;
- Unusual preoccupation, repetitive behaviour and obsessions with particular objects and routines.

A recent US clinic study showed that the rate of Autism is significantly raised in boys with Duchenne. The prevalence rate was 3,7 % compared to a prevalence of 0,16% in general population. Data from our own research group (Hendriksen & Vles, 2008) resulted in a prevalence rate of 3.2%. Conclusion: there is a significant association between Duchenne and ASD. The already mentioned neurobiological mechanism is assumed again: the crucial protein dystrophin is lacking in the brain.

3.2 Attention Deficit Hyperactivity Disorder (ADHD):

Another important behavioural issue is ADHD, Attention Deficit Hyperactivity Disorder. One can distinguish two types of ADHD: with and without hyperactivity. ADHD without hyperactivity is called ADD. ADHD has to do with:

- Problems with emotional regulation;
- Problems with behavioural inhibition;
- Inflexible thinking;
- Poor planning and problem solving;
- Problems learning from own mistakes.

In boys with DMD we (Hendriksen &Vles, 2008) found a prevalence rate of ADHD of 11.8%. Epidemiological studies in general populations revealed an overall prevalence rate of 6.8%. So the prevalence of ADHD in DMD boys is somewhat higher than in the normal population.

3.3 Obsessive Compulsive Disorder (OCD):

Obsessive Compulsive Disorder are about intrusive thoughts, irrational or superstitious fears, worries, concerns with contamination and the need for evenness or symmetry. Compulsions (like checking, counting, tapping or redoing things) are then meant to relieve anxiety or to prevent a dreaded event. As such OCD might have a higher incidence in children confronted with progressive physical disabilities like DMD. Research shows that the OCD prevalence in DMD is twice as high as normal. In boys with DMD the OCD prevalence is 4.8% while in normal population it is 2.3%. Further research is needed to explore this neuropsychiatric comorbidity

3.4 Conclusion:

To summarize then, the data suggest that neuropsychiatric disoders are more prevalent in DMD boys. Recent studies show that especially ASD, ADHD and OCD are more common in boys with Duchenne Muscular Dystrophy. The possible causes are not yet fully understood. A brain-behaviour relationship is assumed: dystrophin being absent in the brains of people with Duchenne. However, there might also be psychological causes like coping with Duchenne, family stress, peer difficulties, school problems and fatigue. We believe that there is a relationship between genes and behaviour. Further research on neuropsychiatric comorbidity is of great importance for effective psychological treatment and follow-up of boys with DMD and their quality of life.