

Learning potential and psychosocial adjustment in boys with DMD

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Boys with Duchenne not only have physical and motor problems but also may have some specific associated psychological problems. Two problems are known to be of significant influence on the general functioning of these boys: problems with learning potential and with psychosocial adjustment. The aim of our contribution is to highlight these aspects which are so relevant to the boys and their families, and warrant further research.

1. Learning potential in DMD: evidence for specific learning disabilities.

The debate as to whether these boys have a cognitive deficit has been going on since the disease was originally described by Duchenne in 1868, when he reported five patients with some degree of cognitive impairment (Brody and Wilkins 1968). Although initially attributed to the environmental and psychological consequences of a chronic debilitating disorder, there is now an overwhelming amount of evidence supporting Duchenne's original observation that these boys show a mild, non progressive, cognitive impairment (Cotton, Voudouris et al. 2001; Emery and Muntoni 2003). More particularly, it is now well established that the affected boys show specific neuropsychological deficits involving specific language delays. Thus, boys with DMD appear to have sub-average cognitive capacities (average IQ of 85). In a recent meta-analysis of studies on cognitive functioning in DMD, consisting of a total of 1224 boys, Cotton and colleagues (Cotton, Voudouris et al. 2005) found that Full Scale IQ and Performance IQ did not change with age in boys with DMD. On the other hand, significant age group differences were found for Verbal IQ, with mean scores improving with age. This suggests that verbal impairments are more prevalent in younger boys with DMD. It is also reported that boys with DMD may first present with language delays, which may be more apparent to the parents initially than the associated motor delays (Essex and Roper 2001). In a recently published study (Hendriksen and Vles 2006), the information-processing capacity, reading performance, and behavioral functioning of 25 Dutch boys with Duchenne muscular dystrophy (mean age 10.1 years) were systematically assessed. Five of the boys had serious reading problems and another five had moderate reading problems, which means

that reading problems are significantly more common in boys with Duchenne muscular dystrophy (40%) than in boys from a normal population (3 to 10%). It was also found that these reading problems were independent of the level of information processing and behavioral functioning. It is suggested that reading problems in DMD boys likely occur as the result of expression of the dystrophin gene in the central nervous system, more specifically in the cerebellum (Muntoni, Torelli et al. 2003; Anderson, Head et al. 2004). The cerebellum is a brain structure which has been shown to play a role also in language and reading (Bishop 2002).

Reading is important for later education, especially in boys with DMD who have progressive motor problems and rely more and more on communication by written words (reading and spelling). The current emphasis on computer skills in many work settings also underscores the importance of reading. Early detection of and interventions for reading disabilities can ameliorate the development of later reading performances in boys who are at risk for reading problems.

2. Psychosocial adjustment and behavioral functioning

Research has documented that children with physical handicaps are at an increased risk for experiencing emotional or behavioral problems. Between 20% and 30% of children with a chronic illness or disability will experience significant behavioral or psychological problems at some point during childhood or adolescence, a rate twice that of physically healthy children (Witt, Riley et al. 2003). The psychosocial adjustment of children with Duchenne has some special areas of impact and is related to the age of the boy and the physical decline. These children often experience more anxiety when they start to fall and are not able to get up again on their own. Furthermore, as they grow older they discover that more exercise does not improve their physical condition. The decline in condition and regular medical assessments can increase their anxiety for physical decline. Boys with DMD, together with their families, are thus confronted with a lot of stressors with which they have to cope. This may result in a greater prevalence of clinically significant stress and behavioral/emotional morbidity.

However, very little evidence is available concerning psychosocial adjustment and behavioral functioning in boys with DMD. In reviewing the literature (PSYLIT and MEDLINE) we found only 11 studies since 1980 on this subject. In 1986 Fitzpatrick was the first author to systematically evaluate the prevalence of psychiatric problems in boys with Duchenne (Fitzpatrick, Barry et al. 1986). He found that symptoms of depression of a varying degree of severity were common among 52% of the older boys with DMD. Leibowitz and Dubowitz report rates of 30% of emotional disturbance in their 1981 study (Leibowitz and Dubowitz 1981). Another study reported that 89% of the children were classified by parent report as having behavior problems (Thompson, Zeman et al. 1992).

There are only a few standardised instruments available that have been successfully used in children with a chronic illness. The Child Behaviour

Checklist (Achenbach and Edelbrock 1983) which is widely used in children, is not recommended for use in children with chronic physical conditions (Witt, Riley et al. 2003). The Personal Adjustment and Role Skills Scale (PARS) III was developed by Stein and Jessop in 1990 (Stein and Jessop 1990) to assess psychosocial adjustment in children with chronic physical illnesses (Witt, Riley et al. 2003). The questionnaire has good psychometric properties and assesses six domains that are associated with patterns of psychosocial maladjustment in children with a chronic physical illness. These domains are: peer relations, dependency, hostility, productivity, anxiety/depression and withdrawal.

We performed a survey on psychosocial adjustment in boys with DMD. The parents from the Dutch and American Duchenne Parent Project were asked to participate in a questionnaire study. The parents of 351 DMD boys (mean age 12.2 years; ages ranging from 3 to 38 years) completed a questionnaire on psychosocial adjustment and behavioral functioning. To the best of our knowledge this is the first study to examine psychosocial adjustment in a large-scale sample of boys with DMD. Some of the results of this study will be presented.

3. In conclusion.

Boys with DMD are not only thwarted by the inevitable physical deterioration of the disease but also by possible emotional and behavioral problems, impaired cognitive functioning and specific learning difficulties. It is important for parents, physicians, and other caregivers to remain cognisant of the psychological vulnerabilities of these boys. These psychological aspects need to be taken in account in the (early) diagnosis, treatment and rehabilitation of the boys and their families, because their quality of life can be affected by these problems. Further research is needed to help elucidate these issues.

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