Males with Duchenne muscular dystrophy have sub-average cognitive capacities and may manifest more specifically language-related deficits. In the current study, the information-processing capacity, reading performance, and behavioral functioning of 25 Dutch males with Duchenne muscular dystrophy (mean age 10.1 years) were systematically assessed. This study relied on the use of a new battery of tests to explore more precisely reading disabilities in males with Duchenne muscular dystrophy. Five of the males had serious reading problems and another five had moderate reading problems, which indicates that reading problems are significantly more common in males with Duchenne muscular dystrophy than in males from a normal population. These reading problems were independent of the level of information processing and behavioral functioning. Implications of these findings and possible directions for future research are discussed, especially with regard to the early detection and treatment of reading problems in males with Duchenne muscular dystrophy.

Cognitive and Academic Impairments in Duchenne Muscular Dystrophy

Recently, Cotton et al. [9] summarized the results of 32 studies published since 1960. The studies involved in total 1224 males with Duchenne muscular dystrophy (mean age 12 years and 4 months, range 2 to 27 years). The mean full-scale intelligence quotient was 80.2, with scores ranging from a low of 14 to a maximum of 134, describing a considerable heterogeneity and a great deal of variety in cognitive abilities among males with Duchenne muscular dystrophy. Generally, Verbal intelligence quotient scores tended to be somewhat lower than Performance intelligence quotient scores: mean Verbal intelligence quotient was 80.4; mean Performance intelligence quotient was 85.4. The mean Verbal intelligence quotient – Performance intelligence quotient of −5.0 is clinically not significant, where a discrepancy of 10 intelligence quotient-points is required at the 0.05 level [9]. Thus males with Duchenne muscular dystrophy appear to have sub-average cognitive capacities. In a further meta-analysis of these data, Cotton et al. [10] found that Full-scale intelligence quotient and Performance intelligence quotient did not change with age in males with Duchenne muscular dystrophy. On the other hand, significant age group differences were found for Verbal intelligence quotient, with mean scores improving with age. This suggests then that verbal impairments are more prevalent in younger children.

Males with Duchenne muscular dystrophy also manifest specific cognitive deficits besides this general intellectual impairment: more specifically, there seems to be a language-related deficit [11]. The relative impairment in verbal abilities seems to be caused by a defect in auditory working memory [12,13]. Essex and Roper reported on the late diagnosis of Duchenne muscular dystrophy presenting itself initially as a global and language-related delay [14]. Of 18 males without a family history of Duchenne, who were diagnosed with Duchenne muscular dystrophy, 8 (44%) were first referred to health professionals because of concerns about language and cognitive development. Other authors [15,16] have reported the occurrence of speech language delay or specific language impairment even before the onset of muscle weakness.

Children who fail to develop language normally in the absence of explanatory factors, such as neurologic disorders, hearing impairments, and adequate environmental stimulation of language, are clinically described as having specific language impairment. It has been demonstrated that genetic factors are important in the etiology of specific language impairment. It has been argued [17] that “developmental dyslexia and specific language impairment are now regarded as different manifestations of the same underlying problem, differing only in severity of developmental stage” (page 858). From a neuropsychologic point of view, language and reading skills are strongly related and might be considered as a continuum.

Children with a delay in language development are at risk of developing reading problems at a later age [5,6,17]. Dyslexia is traditionally defined [18] as “a disorder in children who despite conventional classroom experience, fail to attain the language skill of reading, writing and spelling commensurate with their intellectual abilities” (pages 21-22). Several hypotheses have been formulated to account for the full range of difficulties exhibited by dyslexic children. The phonological deficit hypothesis states that dyslexia is a language disorder in which phonological processing is deficient [17]. Another theory is the cerebellar deficit hypothesis [19], which holds that difficulties in skill automatization, as is the case in reading and spelling, are directly associated with the role of the cerebellum.

Until now there has been no systematic research on the prevalence of specific language impairment and dyslexia in males with Duchenne muscular dystrophy. Only a few studies have reported on specific neuropsychologic defects [12,13,20-22]. Auditory working memory, expressive language, and attentional processes appear to be relatively weak whereas executive functioning and visuo-motor functioning are relatively strong. Wicksell et al. concluded that the short-term memory deficits probably play a critical role in the cognitive impairment and intellectual development of these males, but also found that the Duchenne muscular dystrophy group performed lower on visuo-spatial tasks and executive functions [12]. There is thus still debate on the specific cognitive profile in males with Duchenne muscular dystrophy. Even fewer studies have been carried out on reading performances [8,23,24]. Dorman et al. reported that half of the 15 Duchenne muscular dystrophy males had deficient reading or spelling skills [23]. Billard and coworkers reported that males with Duchenne muscular dystrophy [8] had reading difficulties (mean reading quotient of 85%), which is in accordance with the observation of Leibowitz and Dubowitz who already in 1981 observed [24] that “a considerable number of Duchenne muscular dystrophy children were particularly handicapped in reading” (page 586).

Reading performance varied considerably between the 21 Duchenne muscular dystrophy males, who were more likely to have a delay in reading acquisition than children with spinal muscular atrophy and normal children. Billard et al. [8] used both an age-matched control group and a second control group with severe motor impairment. A psychoaffective explanation for the reading problems was excluded, although the children were not behaviorally assessed [8]. We used the Child Behavior Checklist to assess a possible relationship between reading and behavioral functioning [25].

Patients and Methods

Twenty-five Dutch males with Duchenne muscular dystrophy participated in the study. Males were recruited from the members of the Dutch Duchenne Parent Project, and were living in different parts of Holland.
There were 112 parents, with males of different ages, who received a letter sent by the Duchenne Parent Project, informing them about the aim and procedure of the study. The local medical ethics committee approved the research project. Thirty-eight parents of school-age males with Duchenne muscular dystrophy agreed to participate in the study. Only males who had had more than 20 months of reading instruction \((n = 25)\) were selected for the study because they could be tested for the existence of reading problems. All testing was performed at the patients’ home or school setting. The ages of the patients varied from 8 to 12 years with a mean age of 10.1 years \((S.D. 1.2)\). Eleven of the males attended a normal school; 14 visited a special school for children with a physical handicap. Motor problems varied among the participants.

For this study, reading problems or dyslexia were defined as poor reading ability in the context of broadly normal intellectual capacities. Tests were selected which would be minimally influenced by motor impairment. The Kaufman Assessment Battery for Children was used to assess information-processing capacities \([26,27]\). Two different types of information processing can be tested with this test, namely sequential processing and simultaneous processing. Sequential processing refers to solving problems in which the emphasis is on the serial order of the stimuli, as in language. Simultaneous processing refers to a gestalt-like or holistic approach, as in visuo-spatial information processing \([26]\). In accordance with the difference between verbal and performance intelligence quotient, we expected that sequential information processing would be lower than simultaneous processing in males with Duchenne muscular dystrophy. The total score \(\text{(mental processing composite)}\) was also computed. Mean scores for the Kaufman Assessment Battery for Children were 100; \(S.D. = 15\).

Reading performance was measured by using the “three minutes test,” a standardized Dutch reading test, in which children have to read three cards of progressively more difficult words \([28]\). The number of words correctly read was counted, so that both reading accuracy and reading speed were assessed. The reading quotient was computed by dividing reading age by chronological age. A child is considered reading disabled if he scores 85% or below on the reading quotient. A quotient of 70% or below is indicative of serious reading problems. The “Klepel test,” a Dutch test for non-word reading skills \([29]\), was also used. Mean scores for the Klepel test were 10; \(S.D. = 3\). Non-word reading is thought to be of special value in the assessment of children with reading problems and dyslexia \([30]\).

To take behavioral functioning into account, the Child Behavior Checklist was completed by parents \([31]\). This self-report questionnaire measures two broad-band scales \(\text{(internalizing and externalizing behavior)}\) and eight narrow-band scales \(\text{(withdrawal, anxiety/depression, social problems, somatic complaints, thought problems, attention problems, destructive behavior, and aggression)}\). The cutoff point for a clinical range in the broad-band scales is 63; in the narrow-band scales this cutoff point is 70.

**Results**

The mean score of the 25 patients on the Kaufman test of information processing was low: mean = 87.3 \((S.D. = 13.6)\). The difference between simultaneous and sequential information processing was significant in the expected direction, with sequential processing scores being statistically significantly lower \((\text{paired-samples} t \text{test}; t \text{value} = -4.1; P < 0.001)\). The average reading quotient was 70% \((S.D. = 32.9)\). The average score for the non-word reading test was 6.8 \((S.D. = 3.8)\). The mean scores on the broad-band scales for internalizing and externalizing problem behavior of the Child Behavior Checklist were within the normal range. None of the scores for the narrow-band scales were in the clinical range. Results are presented in Table 1.

Patients were then classified according to the severity of their reading problems. Reading problems were classified as severe when one of the scores for information processing \(\text{(mental processing composite, sequential or simultaneous)}\) was in the normal range \(\text{(score >85)}\), and both scores for reading quotient and non-word reading were 2 \(S.D.\) below the average \(\text{(reading disabled group)}\). Five males fulfilled these criteria. Reading problems were classified as moderate when information processing was again in the normal range; when reading quotient was 2 \(S.D.\) below average and non-word reading was between 1 and 2 \(S.D.\) below average, 5 males fulfilled these criteria \(\text{(moderate disabled males)}\). Ten patients had normal reading scores and normal intelligence. Two males were not able to read at all and their information processing capacities were 2 \(S.D.\) below average, suggesting mental retardation. Three patients had reading problems but their reading performance was in accordance with their information processing capacity \(\text{(nondiscrepant reading)}\). In Table 2 the main characteristics of the five groups are described.

### Table 1. Information processing scores, reading scores, and Child Behavior Checklist scores for the 25 Duchenne muscular dystrophy males

<table>
<thead>
<tr>
<th></th>
<th>Mean</th>
<th>S.D.</th>
<th>Min</th>
<th>Max</th>
</tr>
</thead>
<tbody>
<tr>
<td>Mental processing composite</td>
<td>87.3</td>
<td>13.6</td>
<td>58</td>
<td>117</td>
</tr>
<tr>
<td>Simultaneous processing</td>
<td>91.4</td>
<td>13.9</td>
<td>64</td>
<td>121</td>
</tr>
<tr>
<td>Sequential processing</td>
<td>81.6</td>
<td>15.7</td>
<td>48</td>
<td>113</td>
</tr>
<tr>
<td>Reading quotient</td>
<td>69.5</td>
<td>32.9</td>
<td>20</td>
<td>115</td>
</tr>
<tr>
<td>Non-word reading</td>
<td>6.8</td>
<td>3.8</td>
<td>1</td>
<td>13</td>
</tr>
<tr>
<td>Internalizing behavior</td>
<td>58.9</td>
<td>8.6</td>
<td>41</td>
<td>72</td>
</tr>
<tr>
<td>Externalizing behavior</td>
<td>52.3</td>
<td>11.8</td>
<td>40</td>
<td>77</td>
</tr>
</tbody>
</table>

### Table 2. Mean scores and standard deviations for information processing, reading scores, and behavioral functioning in the five subgroups of reading problems

<table>
<thead>
<tr>
<th></th>
<th>Normal Reading ((n = 10))</th>
<th>Reading Disabled ((n = 5))</th>
<th>Moderate Reading Problems ((n = 5))</th>
<th>Mentally Retarded ((n = 2))</th>
<th>Nondiscrepant Reading ((n = 3))</th>
</tr>
</thead>
<tbody>
<tr>
<td>Mental processing</td>
<td>97.2 (12.2)</td>
<td>82.0 (3.7)</td>
<td>88.8 (6.6)</td>
<td>61.5 (19.2)</td>
<td>77.7 (3.1)</td>
</tr>
<tr>
<td>Simultaneous processing</td>
<td>101.0 (13.4)</td>
<td>90.2 (5.4)</td>
<td>90.6 (9.9)</td>
<td>68.5 (6.4)</td>
<td>79.0 (2.6)</td>
</tr>
<tr>
<td>Sequential processing</td>
<td>92.1 (12.2)</td>
<td>72.2 (5.2)</td>
<td>87.0 (10.7)</td>
<td>50.0 (2.8)</td>
<td>75.0 (8.7)</td>
</tr>
<tr>
<td>Reading quotient</td>
<td>101.4 (14.3)</td>
<td>30.3 (10.4)</td>
<td>50.2 (6.6)</td>
<td>NA</td>
<td>52.7 (16.3)</td>
</tr>
<tr>
<td>Non-word reading</td>
<td>10.4 (1.5)</td>
<td>3.2 (0.9)</td>
<td>8.0 (1.0)</td>
<td>NA</td>
<td>6.3 (2.1)</td>
</tr>
<tr>
<td>Internalizing behavior</td>
<td>59.2 (9.6)</td>
<td>53.2 (8.6)</td>
<td>60.2 (9.0)</td>
<td>61.0 (9.9)</td>
<td>62.0 (6.0)</td>
</tr>
<tr>
<td>Externalizing behavior</td>
<td>48.0 (12.3)</td>
<td>58.5 (13.9)</td>
<td>51.6 (12.9)</td>
<td>47.5 (14.4)</td>
<td>57.0 (15.7)</td>
</tr>
</tbody>
</table>
The patients with reading disabilities had a low score for sequential information processing. Moreover, sequential information processing scores were significantly different in the reading disabled and the moderate reading groups (independent samples t test: $t = -2.8; P = 0.02$), which was not the case when the normal group was compared with the moderate reading problems group (independent samples $t$ test: $t = 0.8; P = 0.44$). Simultaneous information processing scores were not significantly different in the reading disabled and moderate reading problems groups, neither in the normal reading and moderate reading problem group (independent samples $t$ test: $t = 1.5; P = 0.15$). Thus the groups appeared to differ not only in reading ability but also in sequential information processing ability. None of the behavior problem scores on the Child Behavior Checklist (broad-band and narrow-band scales) were significantly different in the normal reading, reading disabled, and moderate reading disabled groups (Table 2). Reading quotient and non-word reading were furthermore not significantly associated with scores on the Child Behavior Checklist when using bivariate correlations.

### Discussion

In accordance with the literature [9,10], males with Duchenne muscular dystrophy were found to have a low level of information processing with considerable heterogeneity, and relatively low scores for sequential processing capacity. Whereas most studies of children with Duchenne muscular dystrophy have used the Wechsler Intelligence Scale for Children [32], we used the Kaufman Assessment Battery for Children, which provides information on information processing instead of intelligence [26,27]. Scores for simultaneous processing (visuospatial information processing) were relatively good, whereas those for sequential processing (language processing) were relatively poor, especially in the reading disabled group.

In contrast to other studies [8,23], this study used the discrepancy criterion to classify reading problems by their severity [5,17]. As there is a continuing debate on the diagnostic criteria of dyslexia [17,33,34], we restricted ourselves to technical reading problems. Spelling problems were not taken into account, which may be a separate problem. Individuals with reading problems usually also have spelling problems. The spelling problems in males with Duchenne muscular dystrophy warrant further research, as there might also be Duchenne muscular dystrophy males with isolated spelling problems [23].

The mean reading quotient in the 25 Duchenne muscular dystrophy males in the present study was 70%, which is significantly lower than the 85% reported by Billard et al. [8]: one-sample test $t = -2.308; P = 0.03$. Thus the reading disabilities of Duchenne muscular dystrophy males appear to be even more serious than previously thought. The scores for the reading quotient in the study of Billard et al. yielded less variation (range = 67-124) than in the present study (range = 20-115). Because both Billard et al. and we used standardized and validated instruments to measure reading performance, the discrepant findings cannot be explained by the different instruments used. Other factors therefore play a role, such as different recruitment of the participants and different methodology.

Of the 25 Duchenne muscular dystrophy males who finished elementary reading instructions, five had serious reading problems in technical reading not in accordance with their level of information processing, and another five patients had moderate reading problems also not in accordance with their normal information processing capacities. We can therefore conclude that males with Duchenne muscular dystrophy definitely exhibit a higher rate of serious reading problems than do males from a normal population, where the incidence of dyslexia is estimated between 3% and 10% [5,17,34]. A nonparametric binomial test with 10% as the hypothesized value (i.e., incidence in the normal population) yielded a significant difference in the prevalence of reading problems in Duchenne males and the normal population ($P < 0.000$). When compared with the results from Dorman et al., we find comparable results with 40% of the Duchenne muscular dystrophy males in the present study having reading problems [23]. Although a control group was not used in this study, which might be a methodological drawback, the data were analyzed in comparison with existing data (i.e., normal incidence of dyslexia and the results of Dorman et al. and Billard et al.). We therefore believe our data have clinical importance. More extensive studies are required to further clarify the nature of reading and spelling problems in Duchenne muscular dystrophy males.

Behavioral functioning as rated by parents was not associated with reading scores. Billard et al. [8] already ruled out a purely psychoaffective explanation for the reading problems. However, they did not systematically evaluate the behavioral functioning of the males in their study. We used the Child Behavior Checklist [25,31], which is a commonly used instrument to detect behavioral problems.

In conclusion, males with Duchenne muscular dystrophy are at a higher risk of developing reading problems, independent of their level of information processing and behavioral functioning. This finding has several important implications. First, the presumed role of the cerebellum in both Duchenne muscular dystrophy and dyslexia is theoretically important: it explains the higher incidence of reading problems in Duchenne muscular dystrophy. The brain involvement which was assumed by Duchenne de Boulogne in 1868 is now beyond doubt. As such, the findings of the present study also underscore the involvement of the cerebellum in higher-order cognitive processes and skill automatization [4,6,19]. Secondly, reading is important for later education, and especially in males with
Duchenne muscular dystrophy who have progressive motor problems and rely more and more on communication by written words (reading and spelling). Early detection of and interventions for reading disabilities have been demonstrated to be effective in children with a higher risk for dyslexia on the basis of parental inheritance [34]. This finding is also important in males with Duchenne muscular dystrophy. We therefore suggest systematic screening of Duchenne muscular dystrophy males at age 4 years to detect whether they are at risk of later reading problems. Screening should be aimed at sequential information processing and at phonetic awareness because these aspects most likely play a crucial role in later reading acquisition [5,17,33,34]. Moreover, phonological awareness can be trained in preschool programs and such training has a beneficial effect on later reading in children at risk for reading problems [33,34]. Research on intervention strategies and reading problems in Duchenne muscular dystrophy has not yet been described but is required.

We wish to thank the Duchenne Parent Project Netherlands for their support and participation. We also wish to thank Brigitte Vugs and Monique Thijsse, masters in psychology, for their participation in the data collection. Finally we want to thank the “Stichting ter behartiging van de belangen van het gebrekkige kind” for supporting the study financially.

References