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Reading and spelling impairments in children and adolescents with infantile myotonic dystrophy

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Abstract

This study investigated reading and spelling difficulties in subjects with the juvenile form of myotonic dystrophy (MD). Twenty-three consecutive patients with juvenile MD who were referred to a special clinic were assessed for reading and spelling skills (phonological processing, word identification, narrative comprehension (two tasks), information seeking in a document (TV schedule), and spelling). Reading impairments were frequent (63–84% of the subjects being below the level of literacy depending on the tasks), even in subjects without mental retardation (22–66%) despite normal word identification scores. All but two subjects had spelling difficulties. The severity of these learning difficulties was correlated with longer mutation size and maternal transmission, but could not be related to phonological deficit, suggesting that other brain dysfunction might be involved (e.g., attention, working memory, naming speed, executive function). Children

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with the juvenile form of MD should systematically be assessed for reading and spelling problems, and correlations with basic cognitive functioning explored.

Keywords: Myotonic dystrophy; Children; Reading disability; Spelling disability

1. Introduction

For the last 20 years, there has been a growing interest in promoting a multidisciplinary approach for genetic diseases. For instance, in the field of autism and mental retardation that are frequently associated with a genetic cause, a multidisciplinary evaluation involving a child psychiatrist, a neuropediatrician, and a geneticist is now recommended (for a review see Cohen et al., 2005). More recently, genetic abnormalities with a more subtle phenotype have been investigated with promising results in terms of links between cognition, learning and brain functioning. For e.g., Molko et al. (2003) found that women with Turner's syndrome, that was considered, until recently, a genetic abnormality associated with no or little cognitive dysfunction, had dyscalculia with structural and functional abnormalities in specific areas, including the horizontal intra-parietal sulcus (HIPS).

Myotonic dystrophy (MD) is an autosomal dominant inherited disorder secondary to an expanded and unstable CTG repeat sequence of the DMPK gene on chromosome 19 (Harper, 1998). The prevalence of the DM-mutation is estimated to be 1 in 8000 (Hunter et al., 1992). Its phenotype is particularly complex and characterized by a wide variation in neuromuscular symptoms and multisystem involvement: myotonia, progressive muscular weakness, cataract, smooth muscle disturbances, subfertility, mental retardation, depression, somnolence, diabetes and cardiac conduction defects (Harper, 1989). Considering its clinical manifestations and their age of onset, Harley et al. (1993) proposed a classification for MD consisting of four types: (i) the mild form of MD with cataract and no or minimal muscular symptoms in middle or older age; (ii) the classical form with typical neuromuscular symptomatology in adolescence or early adult life; (iii) the juvenile form with learning disabilities often prominent in early life (before the age of 10 years), but with neuromuscular signs mild or sometimes even absent at first diagnosis and (iv) the congenital form with clinical symptoms present at birth or in utero: hypotonic cerebral palsy, respiratory and feeding problems, and mild to moderate mental retardation in survivors. This wide variation in phenotype is partly explained by molecular mechanisms: (1) a correlation is reported between the size of the trinucleotide repeat and the clinical severity (Harley et al., 1992); (2) the congenital form is caused by maternal transmission although a few exceptions were reported (Ashizawa et al., 1994).

To date, only a limited number of studies have been conducted on the clinical phenotype of infantile form of MD. In several papers, Steyaert et al. (1997), Steyaert, de Die-Smulders, Goossens, Willekens, and Fryn (2000) and Goossens, Steyaert, de Die-Smulders, Willekens, and Fryns (2000) have described general cognitive functioning and the psychiatric phenotype of 24 children and adolescents with infantile MD using a systematic and standardized evaluation. The full-scale intelligence quotient (IQ) of the subjects varied between 50 and 97 (mean 72; SD = 14) with the lower IQ correlating with the transmitting parent's sex. Half of the subjects had problems with attention and social

interaction and were withdrawn. Fifteen received a psychiatric diagnosis, the most frequent being attention deficit hyperactivity disorder and anxiety disorder. Finally, all except four attended a special education program or worked in a sheltered workshop. We failed to find in the literature a systematic study of learning impairments in children with juvenile MD, although they were noted by Harley et al. (1993) in their seminal report.

Reading is acquired during childhood through education, and involves several visual, auditory, and motor skills. In terms of cognitive processing, one of the crucial steps is written word identification (Goswami & Bryant, 1990). Two word identification strategies or routes are used: the first being a grapho-phonologic conversion, or assembling strategy, which depends on converting grapheme into phonemes and then blending those phonemes to form a word; the second being an orthographic or lexical strategy, which enables readers to target the whole word in a stock of familiar written words and to access the corresponding lexical entry (Humphreys & Evett, 1985). While necessary, word identification is not sufficient for reading. Reading also involves the use of syntax rules and the assimilation of specific characteristics of written texts. This is why literacy measures usually assess sentence and connected text comprehension as well as word identification. Comprehension reading tasks can be classified into two main groups: the first requiring high-level processing with a fine-grain analysis of syntactic, semantic and pragmatic cues; the second requiring low-level processing—such as use of graphical cues or understanding of the logic of a table or list—in order to extract selective information in specific forms, lists, tables or programs (Rivière, 2002). Regarding cognitive correlates, several studies conducted on school-age children have established that reading acquisition appears to be influenced by several cognitive competencies: phonological awareness (Goswami & Bryant, 1990; Plaza & Cohen, 2003), working memory (Crain, Shankweiler, Macaruso, & Bar-Shalom, 1990; Torgensen, 1985), and naming speed (Plaza & Cohen, 2004; Wolf et al., 2002). Of note, in children with normal intelligence, IQ does not predict reading skills whatever the task (global reading, syntax or word identification) (Share, McGee, & Silva, 1989; Siegel, 1989); and similarly, reading disability is not related to IQ score (Siegel, 1989) unless for IQ in the range of mental retardation (Cohen et al., 2001).

The current study aimed (i) to determine the frequency and profile of reading and spelling impairments in 23 children and adolescents with infantile MD using both basic and global reading tasks; (ii) to assess whether the difficulties are the consequence of mental retardation.

2. Method

2.1. Subject selection and evaluation of participants

Twenty-three consecutive patients with juvenile MD who were referred to the special clinic for myopathy at La Salpétrière in Paris were included in the study. The evaluation started with a clinical interview, including assessment of neurological status, and examination of medical records to retrospectively assess the subjects' development, education, social background, and genetic status (mutation size and parental transmission). All subjects were administered the Wechsler Intelligence Scale for Children-III (WISC-III) to assess their IQ scores (Wechsler, 1974), and a 2-h battery of tests (described below) administered by a reading specialist to assess the reading and spelling abilities.

2.2. Assessment of reading and spelling skills

To examine phonological processing, seven tasks were devised and administered orally. These tasks measure the child's understanding of the basic speech–sound structure of spoken language including sound discrimination, blending and segmentation skills (Chevrie-Muller & Plaza, 2001). These experimental tasks integrate different skills: (1) phonological similarities sensitivity involving rhyme identification; (2) phonetic discrimination and (3) metaphonological awareness, involving complex phoneme manipulation. Only task 1 (rhymes) is composed of real words. All the other tasks involve pseudowords, which do not have a lexical and semantic entry and then require the phoneme-by-phoneme (or syllable-by-syllable) assembling process. The experimenter gave two examples for each task. The child had to repeat each item of each task before processing it. No corrective feed-back was given. (1) Rhyme recognition (task 1): The child was asked to decide whether or not pairs of orally presented words rhyme (example: no for BOUT-BOL, yes for NOUS-SOUS). The list included 20 pairs of monosyllabic real words. (2) Identification of the initial phoneme (task 2): The child was asked to identify and to produce the initial phoneme of pseudowords (example: A for ALAC, M for MILU). The list included 10 bisyllabic pseudowords (with 5 vowels and 5 consonants in the initial position). (3) Syllabic inversion (task 3): The child was asked to reverse bisyllabic pseudowords, and to pronounce the resulting pseudowords (for example, PATI becomes TIPA). The list included 10 pseudowords. (4) Initial phoneme deletion (task 4): The child was asked to delete the initial phoneme and to pronounce the remaining pseudoword (for example POUK becomes OUK). The list included 20 mono and bisyllabic pseudowords with 14 consonants and 6 vowels in initial position. (5) Initial phoneme addition task (task 5): The child was asked to add phonemes to the beginning of pseudowords and to pronounce the resulting pseudowords (for example, P + AMO = PAMO). The list included 17 pseudowords. (6) Final phoneme deletion task (task 6): The child was asked to delete the final phoneme of pseudowords, and to pronounce the resulting pseudoword (for example, PAC without C = PA). The list included 20 pseudowords (10 monosyllabic and 10 bisyllabic). (7) Phonemic inversion task (task 7): The child was asked to reverse pseudowords and to pronounce the resulting pseudowords (for example, AL becomes LA). The list included 10 pseudowords. The seven phonological tasks are now validated in French with norms for children of 6, 7 and 8 years of age (Chevrie-Muller & Plaza, 2001). Two scores were generated: sensitivity score (tasks 1–3) and metaphonological awareness score (tasks 4–7).

Regarding global reading tasks, four tasks were proposed: word identification, syntax (sentence comprehension), narrative comprehension, and information seeking (for details, see Cohen et al., in press). Word identification was assessed with the *Mesure d'Identification de Mots* (MIM) [Word Identification Measure] (Mousty, Leybaert, Alegria, Content, & Morais, 1994). This task assesses oral reading. The subject is asked to read 48 words aloud which vary in terms of frequency, length, and complexity, and 24 non-words which vary in the same characteristics except for frequency. Word identification task is scored using the number of words correctly read by the subject. Reading time is also monitored. Sentence comprehension was assessed with the *Test de compréhension de phrase* [Sentence Comprehension Test] (Rivière, 1998). This task uses a picture/sentence matching task with five sentences in which one word is missing. The missing words are presented in various contexts, which differ only in terms of syntactic properties (negation, tense,

number, etc.). For each picture/sentence, the subject is asked to choose among four answer options. The task is scored by the percentage of correct answers. Text comprehension tasks included the evaluation of two types of texts that require high- and low-level processing (see Introduction). Narrative comprehension was assessed using two written prose texts: *Mortelle matinée*, a long but easy text (507 words, 7 multiple choice questions with four answer options) (Mesnager, Rivière, & Bentolila, 1994) and *Jacques Lentide*, a shorter but complex text (151 words, 4 multiple choice questions with four answer options) (Rivière, 1998). Global comprehension and comprehension of contradictions, actions, resolution, history, etc., in the narrative are assessed using a multiple-choice question paradigm. Both tasks are scored by the percentage of correct answers. Literacy is obtained for scores superior to 71%. Finally, the TV schedule test (Mesnager et al., 1994) is a task that requires a selective understanding of written information. In the task, the subject is asked to seek information in a document (TV schedule for 1 day). Several abilities are tested: scanning by using graphical features (bold, italic, etc.), understanding of the logic of a list or a table, and comparison of time slots. Seven multiple-choice questions are administered, and the task is scored by the percentage of correct answers.

Finally, we assessed spelling using two dictation tasks validated in French: "*Le Corbeau*", for children 11 years and younger; "*Tempête au Sahara*", for children 12 years and older (Chevrie-Muller, Simon, Fournier, & Brochet, 1997). Given the heterogeneity of scores and ages, a spelling score was calculated by comparing each subject's score with specific age-norms: -3 (non-readers); -2 (very low $\leqslant -2\sigma$); -1 (low $\leqslant -1\sigma$); 0 (normal); 1 (good $\geqslant 1\sigma$); 2 (very good $\geqslant 2\sigma$).

2.3. Data analysis

All statistics were performed with the SPSS package. Because of the heterogeneity in IQ scores, the data were shown for the total group and two subgroups (verbal IQ $<$ or >76) (see below). For a description of the study groups and comparison with French norms when available, we used parametric and non-parametric comparison tests as necessary. Effect of mutation size on severity was studied using Pearson correlation whereas effect of transmission (maternal versus paternal) was studied using a Spearman point biserial correlation.

3. Results

3.1. Characteristics of the experimental group

Twenty-three children and adolescents with juvenile MD, 14 boys and 9 girls, aged 8–18 years (mean age = 13.4 years; SD = 2.9), were included. General characteristics of the subjects are summarized in Table 1. IQ scores ranged between 42 and 91 with verbal IQ being significantly superior to performance IQ (Wilcoxon's test $Z = -2.37$; $p = 0.018$). We divided throughout the analysis the study sample into two subgroups: subjects with verbal IQ <76 ("low verbal") ($N = 12$) and subjects with verbal IQ >76 ("high verbal") ($N = 11$). Of note, the difference between verbal IQ and performance IQ was significant only in the high verbal IQ subgroup (Table 1 right column, Wilcoxon's test $Z = -2.7$; $p = 0.007$). The two subgroups had a similar mean age and sex ratio with more boys than girls.

Table 1
General characteristics of 23 children and adolescents with infantile dystrophic myotony

	Total ($N = 23$)	QIV < 76 ($N = 12$)	QIV > 76 ($N = 11$)
General characteristics			
Age	13.4 (±2.9) [8–18]	13 (±3) [8–18]	13 (±3) [8–16]
Gender	14 M, 9 F	7 M, 5 F	7 M, 4 F
Academic delay	16 (69%)	11 (91.6%)	5 (45.5%)
Special education	9 (39%)	8 (66.6%)	1 (9%)
Reading therapy	16 (69%)	9 (75%)	7 (63.6%)
Cognitive characteristics			
Global IQ	61 (±15) [42–91][a]	55 (±9) [42–70]	84 (±10) [72–91][a]
Verbal IQ	75 (±22) [46–129]	59 (±11) [46–75]	95 (±15) [77–129]
Performance IQ	68 (±14) [46–93]	59 (±10) [46–76]	79 (±11) [58–93]
Genetics			
Mutation size	567 (±282) [200–1066]	688 (±311) [300–1066]	466 (±198) [200–800]
Transmission	13 Pat (56%)	4 Pat (33%)	9 Pat (81%)
	10 Mat (44%)	8 Mat (67%)	2 Mat (19%)

M = male; F = female; Pat = paternal; Mat = maternal; IQ = intellectual quotient.
[a] $N = 14$, and 3 respectively (9 QIG scores are not valid because QIV ⩾ QIP).

Regarding education, 16 subjects (69%) were academically delayed, 9 were in a special education program (39%) and 16 (69%) had or were undergoing a reading therapy. Even in the high verbal subgroup, 5 (45.5%) and 7 (63.6%) subjects were academically delayed or benefited from reading therapy, respectively. As expected, mutation size was negatively correlated with both verbal and performance IQ (Pearson's $r = -0.581$, $p = 0.009$; $r = -0.527$, $p = 0.02$, respectively). Similarly, maternal transmission was negatively correlated with both verbal and performance IQ (Spearman's $\rho = -0.483$, $p = 0.023$; $\rho = -0.547$, $p = 0.008$, respectively).

3.2. Phonology and word identification

Phonology scores, and word identification scores and speeds are summarized in Table 2. For subjects in the low verbal IQ group, it appeared that all variables were significantly lower than scores obtained in regular French school samples, except for phonological sensitivity. Subjects in the high verbal IQ group did not exhibit lower scores for phonology nor word identification.

3.3. Global reading and spelling tasks (Table 3)

As expected, all subjects with verbal IQ < 76 were below the level of literacy for French persons, for both types of tasks (prose or TV schedule). Subjects with verbal IQ > 76 also had difficulties in global reading tasks. Although most had scores superior to the level of literacy for French persons in the short-prose text task, 5 (55%) of the subjects exhibited difficulties with the long-prose text task although this task was less complex in terms of linguistic characteristics. Similarly, 6 (66%) of the subjects had difficulties with extracting

Table 2
Phonology and word identification scores in 23 children and adolescents with infantile dystrophic myotony

	Total ($N = 23$)	QIV < 76 ($N = 12$)	QIV > 76 ($N = 11$)
Phonology			
Sensitivity			
Mean (±SD)	35 (±5)	33 (±6)	37 (±3)
Score < −2SD: *N* (%)	2 (8.7)	2 (16.7)	0 (0)
Metaphonology			
Mean (±SD)	36 (±11)	31 (±13)	41 (±4)
Score < −2SD: *N* (%)	8 (34.8)	7 (58.3)	1 (9.1)
Word identification scores			
Frequent words score			
Mean (±SD)	18 (±8)	14 (±10)	23 (±1)
Score < −2SD: *N* (%)	9 (39.1)	7 (58.3)	2 (18.2)
Non-words score			
Mean (±SD)	13 (±8)	8 (±8)	17 (±3)
Score < −2SD: *N* (%)	9 (39.1)	7 (58.3)	2 (18.2)
Total score			
Mean (±SD)	46 (±24)	31 (±25)	62 (±5)
Score < −2SD: *N* (%)	10 (43.5)	9 (75)	1 (9.1)
Word identification speed			
Frequent words speed			
Mean (±SD)	37.5 (±42.8)	60.4 (±59.5)	20.9 (±10.4)
Score < −2SD: *N* (%)	8 (34.8)	6 (50)	2 (18.2)
Non-words speed			
Mean (±SD)	61 (±37)	76.8 (±40.4)	49.6 (±31.5)
Score < −2SD: *N* (%)	8 (34.8)	6 (50)	2 (18.2)
Total speed			
Mean (±SD)	144.4 (±111)	199.4 (±142.5)	104.4 (±62.5)
Score < −2SD: *N* (%)	8 (34.8)	6 (50)	2 (18.2)

proper information in the TV schedule task. Finally, 9 (82%) had moderate to severe difficulties in spelling compared with a normative population of French students.

4. Discussion

4.1. Reading and spelling impairments in infantile myotonic dystrophy

To our knowledge, the current study is the first one to focus on reading and spelling acquisition in children and adolescents with juvenile MD although learning difficulties were highlighted since the identification of this form (Harley et al., 1993). Our results show that reading and spelling impairments are frequent, sometimes severe in children and adolescents with juvenile MD, even in those without mental retardation. The high frequency of academic delay and reading therapy in the study group further support the study results. This point needs to be highlighted as reading and spelling impairments may benefit from specific treatments.

Table 3
Reading ($N = 19$) and spelling ($N = 23$) scores in children and adolescents with infantile dystrophic myotony

	Total	QIV < 76	QIV > 76
Global reading tasks[a]	$N = 19$	$N = 10$	$N = 9$
Short text scores			
Mean (±SD)	0.4 (±0.4)	0.08 (±0.1)	0.78 (±0.2)
Score < 71%: *N* (%)	12 (63)	10 (100)	2 (22)
Long text scores			
Mean (±SD)	0.45 (±0.3)	0.26 (±0.3)	0.67 (±0.18)
Score < 71%: *N* (%)	15 (79)	10 (100)	5 (55)
TV program scores			
Mean (±SD)	0.40 (±0.3)	0.20 (±0.23)	0.63 (±0.23)
Score < 71%: *N* (%)	16 (84)	10 (100)	6 (66)
Spelling scores	$N = 23$	$N = 12$	$N = 11$
Spelling scores			
Mean (±SD)	−1.8 (±1)	−2 (±1)	−1 (±1)
Score ⩽ −1: *N* (%)	19 (82)	12 (100)	9 (82)

[a]For global reading tasks, only 19 children and adolescents were included as validation is available for children older than 10 years.

The analysis of the reading and spelling performances of the subjects showed that their profiles were not similar to those of children with dyslexia. The most atypical finding was the low proportion of subjects with phonological difficulties even in subjects with low IQ. This point is surprising since phonology is also associated with reading skills in subjects with mental retardation (Conners, Atwell, Rosenquist, & Sligh, 2001). Second, in subjects with normal IQ, the profile of narrative tasks was surprising as subjects had higher scores in the short but complex prose than in the long but easy one. We hypothesized that long prose would induce more cognitive cost in terms of working memory processing than the short text as the two sores appeared to be correlated (Pearson's $r = 0.594$; $p = 0.012$). Third, among global reading tasks, subjects with juvenile MD exhibited a specific difficulty in extracting information from a TV schedule with 84% of the subjects under the level of literacy.

In order to explain this particular cognitive cost, several hypotheses can be suggested. (i) Given the low word identification speed in some children with MD, the longer the prose the higher the cost. (ii) Attention and/or somnolence that are frequent in children with juvenile MD (Goossens et al., 2000; Harley et al., 1993; Steyaert et al., 1997, 2000) may also be involved. We choose to link attention and somnolence because the relation of the two dimensions in juvenile MD remains to be systematically explored. (iii) Working memory that is correlated with reading skills (Crain et al., 1990; Torgensen, 1985) may also be a major component in children with juvenile MD. (iv) Specific cognitive skills related to executive function, logic, visual–spatial orientation may also interfere and need to be explored in these subjects. We presumed these difficulties, because of the IQ subscores that showed a disharmonic profile with lower performance IQ in this series and the neuropsychological profile described by several groups in the adult form of MD after years of evolution (Angeard-Durand, Héron, Gargiulo, & Eymard, 2004).

Regarding the effect of mutation size and parental transmission on reading and spelling abilities, we found that more severe impairments were associated with longer mutation and maternal transmission. This point is not surprising as it has already been reported for global intelligence skills in both juvenile and adult forms of MD (for a review see Angeard-Durandet et al., 2004).

4.2. Limitation of the study

Although we report novel data, several limitations of the study should be kept in mind. (i) It is probable that our clinical series included selection bias despite all patients presented consecutively. Indeed boys were over-represented, which should not be the case in an autosomal dominant inherited disorder. (ii) The methodology was transversal, and not prospective, and included both children and adolescents. Although all our tasks had norms in French scholars, given the wide ranges in age, we had to approximate a new score for spelling that might be not valid. Furthermore, phonological tasks were not monitored for speed. Older subjects might have exhibited scores within the normal range but with low phonological treatment speed. (iii) Moreover, given the fact that 69% of the subjects had prior reading therapy, it is probable that our results underestimate reading and spelling difficulties as we may expect that basic reading skills were improved with such training.

4.3. Questions remaining and future research

The first point for future research is to confirm our results in another group of patients with the juvenile form of MD. Apart from this aspect, our results suggest that children with juvenile form of MD and reading and spelling impairments might have a particular cognitive profile that associates better verbal than performance IQ, but normal phonological sensitivity. Given the cognitive correlates of reading acquisition in children with normal intelligence (see Introduction), we can hypothesize that attention, working memory, naming speed and executive function may be correlates of reading and spelling impairments in children with juvenile form of MD. This point has major implications in terms of treatment. First, attention deficit may be improved by stimulants. Second, reading therapy should focus on core deficits but we need more comprehensive studies to recommend guidelines for training as phonological deficits does not appear to be the core deficit in the case of the juvenile form of MD. Our group is conducting a study challenging these issues.

5. Conclusion

Reading and spelling impairments appear to be typical of the phenotype of the juvenile form of myotonic dystrophy (MD) even in children with normal intelligence quotient (IQ). The severity of these learning difficulties is correlated with longer mutation size and maternal transmission. Children and adolescents with the juvenile form of MD should systematically be assessed for reading and spelling problems and treated where necessary. More generally, the study confirms that learning impairments should be explored more carefully in genetic diseases that impact brain development, maturation or functioning in particular when global intelligence is normal or subnormal.

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