Psychological aspects in children affected by Duchenne de Boulogne muscular dystrophy

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Abstract

Impairment of intelligence in Duchenne muscular dystrophy (DMD) patients was described by Duchenne de Boulogne himself in 1868. Further studies report intelligence disorders with mayor impairment of memory. The aim of the present study was to assess the presence of affective and personality disorders in a group of children affected by DMD. Twenty six male DMD patients, mean age eleven and four months years old, were assessed for their affective and personality disorder. Only eight subjects had a total IQ below average with major difficulties in verbal and visual-spatial memory, comprehension, arithmetic and vocabulary. All the subjects presented some disorders: tendency to marginalization and isolation, self-depreciation, sense of insecurity, hypochondriac thoughts and marked state of anxiety. These disorders are often a dynamic prolongation of a psychological process which starts when the diagnosis is made and continues, in a slow and latent fashion, throughout the evolution of the disease.

Introduction

Among the dystrophinopaties, which represent the first cause of myopathy in the children, Duchenne muscular dystrophy (DMD) is one of the most severe and frequent forms. DMD is due to an abnormality in a gene located on the short arm of chromosome X (Xp21 band), the mutation of which causes a severe alteration of gene and the loss of dystrophin production.[1-3]

DMD alters not only motor development, but also affective development, causing dramatic changes in the subject's life.[4-6]

Berlucchi in 1934, among 24 subjects affected with DMD (the article told about primitive muscular dystrophy), pointed out in 10 cases (42%) impairments of intelligence of variable degrees. In particular he stressed that mental disorders rose up in this patients, usually, at 14-15 years, when muscular disease was already very advanced.

Duchenne de Boulogne himself (1868), describing the clinical symptomatology of the dystrophinopathy which was to carry his name wrote about *intelligence souvent obtuse, de parole tardive*.[7]

Further studies reported intelligence disorder with major impairment of memory especially long term one.[8-9] Attention deficit was also reported with learning disabilities in arithmetic, writing and reading.[5,10] In children affected with DMD were also observed affective disorders, depressive issues and hypochondriac fears.[11,12]

In many dystrophic patients was noted that, beyond a certain stage of disease, appeared significant psychological and psychopathological problems in the boy and in the household. The family attitude predominantly observed is overprotective and anxious and had depressive reactions.

In this study we evaluated the affective and personality aspects in a group of children affected with DMD.

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Materials and Methods

Twenty six male subjects with DMD, mean age eleven and four months years old, submitted to a psychological evaluation to assess their emotional and relational states. The patients were selected on the basis of physical examination, muscle biopsy, histochemical analysis, Western blot analysis, genetic testing and on their regular and normal schooling. All the subjects enrolled in the study underwent a physical and functional examination (Vignos functional scale),[13] polysomnography in order to rule out the presence of severe respiratory failure, and neurodiagnostic examinations to rule out the presence of brain damage. The following psychodiagnostic tests were used: i) WISC-R intelligence scale: highlights harmonies and disharmonies between the different manifestations of intellectual development. ii) Draw-A-Person test: supplemented with tests on spatial orientation and laterality, it provides indications on how the child perceives his|her body schema by analyzing completeness|incompleteness and staticity or dynamism of the figures. iii) Spontaneous Drawing: gives children the opportunity to express themselves freely through graphic activity. iv) Family Drawing: this evaluates the position of the child in the family (and the degree of closeness with parental figures). v) Rorschach test: this test brings out possible affective disorders and problems in comprehension as well as their nature and origin, it provides a qualitative rather than a quantitative assessment of the individual's intelligence and it makes it possible to recognize if affective disorders have an impact on mental development and behavior. vi) Thematic Apperception test (TAT): this provides simultaneously information about the emotions, attitudes and cognitive processes of the subject.

Results

Genetic aspects

Molecular genetic testing with PCR (polymerase chain reaction) multiplex[14] detected a deletion in the central and proximal high frequency deletion regions (HFDR) in all subjects; no duplication was found.

Testing for dystrophin in muscles (immunohistochemistry, Western blot analysis) did not show immunoreactivity in 19 cases (73.1%) and very weak immunoreactivity was detected in 7 cases (26.9%).

Physical and functional assessment

According to Vignos functional classification[13] (a scale which mainly evaluates lower limb function and where higher scores correspond to more severe impairement), 5 cases (19.3%) had a disease stage between 7th and 8th, 14 cases (53.8 %) were between 4th and 5th and 7 cases (26.9%) where between 2nd and 3th. Pulmonary function was slightly reduced in 2 patients, where data showed a reduced efficiency of respiratory muscles. Polysomnography of other subjects did not reveal significant clinical signs.

The neuroradiological picture was within the norm in all subjects, with no sign of cerebral atrophy.

Psychological aspects

The average score at WISC-R in 24 subjects (84.6 %) was the following: verbal IQ 87; performance I.Q. 98,2; total IQ 94,8. Only 4 subjects (15.4%) had a total IQ below average. Spontaneous drawing was generally poorly developed and with scarce use of colors (73,1% of cases).

The children's drawings expressed depressive contents (a broken house with holes needing repair, night landscapes, etc.).

Some children made strong references to autobiographical episodes (a ship taking the child and his father to another town for a medical visit or the car rides for the physiotherapy) (Figure 1).

Human figures were present in only three cases, an aspect paralleled by the results of the Rorschach test (few H responses).

In the DAP test all figures were complete and mainly in static poses (in all cases), some children drew the picture starting from the lower limbs (Figures 2, 3). More than one child depicted a walking human figure in profile with the typical characteristics of a myodistrophic gait.

Although all subjects showed no difficulties during the orientation and laterality testing, a sense of staticity and movement inibition seemed to prevail.

The Family Drawing test mainly revealed self-depreciation and experiences of marginalization and isolation vis-à-vis the family, which was most often depicted as separated, on a different plane (85%) (Figure 4).

This proneness to isolation was even more apparent in the TAT results (*a boy is crying because the others don't want him to play with them*). Other recurrent stories made reference to death or accidents causing harm to the lower limbs of the *hero* (children or animals *falling down*). Along with depressive content (*a child is crying, he is sad because he is crying…he wants his mother…which is dead*; *there are a mother and a lily, they are very sad because they are thinking of certain things…*) some children, especially those who had not experienced death or disease within the family, developed defense mechanisms through which they removed these *disasters* by placing them far away in time (*in the ancient times this man went to war and was wounded*) or in the space (*there was an earthquake in London, a skyscraper collapsed and some people were killed. After many years of work the city has been built again*).

Other subjects resorted to miracles, thanks to which the stories have happy endings. In these stories we found recoveries after surgery (*a child is crying because he has grown a leg, then he has an operation and he is cured*; *a boy is a drug addict..he is brought to hospital for treatment and he is cured*) or rescue operations performed by imaginary characters (*this is*

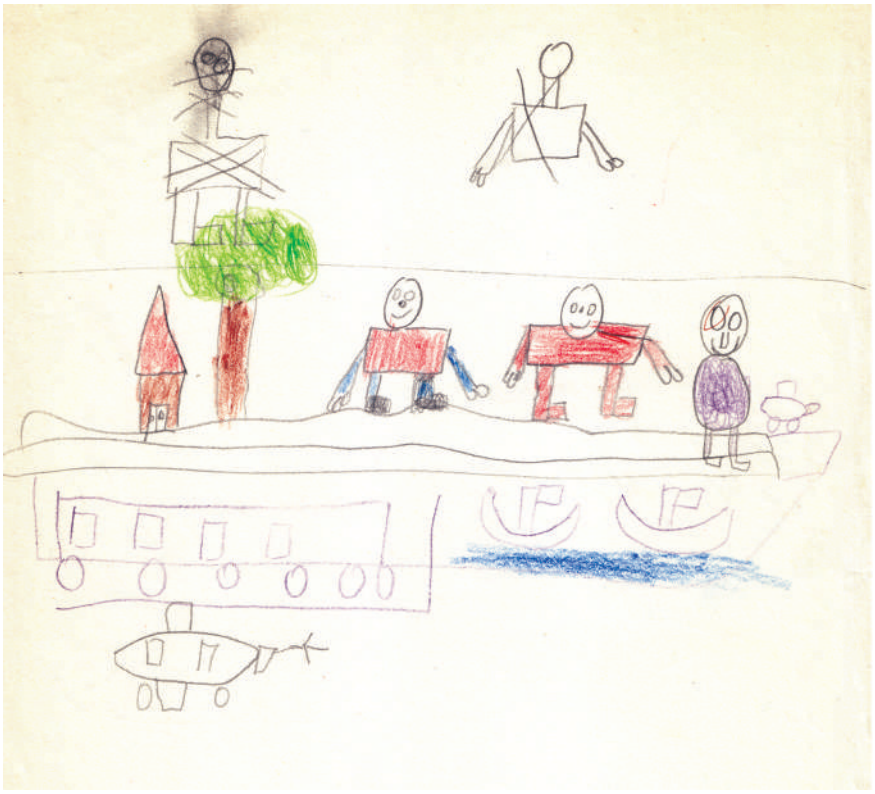

Figure 1. Drawing of a child with Duchenne muscular dystrophy.

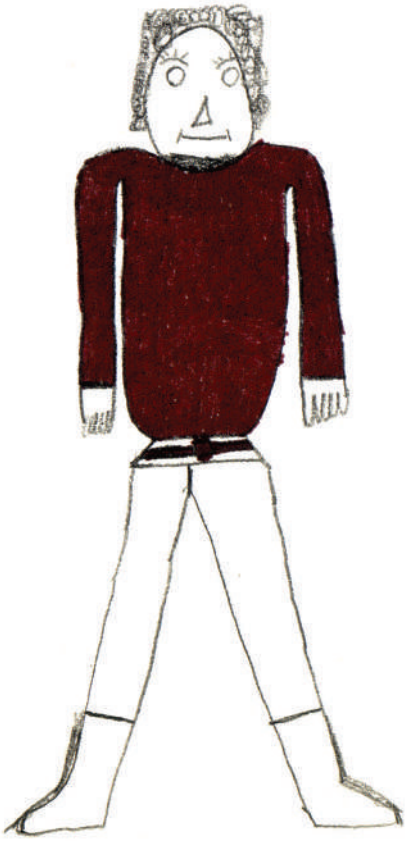

Figure 2. Drawing of a child with Duchenne muscular dystrophy (Draw-A-Person test, example 1).

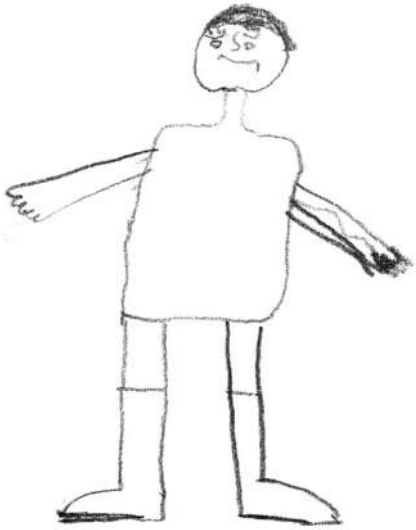

Figure 3. Drawing of a child with Duchenne muscular dystrophy (Draw-A-Person test, example 2).

Tarzan, he is climbing up the wires…he has to rescue someone who has fallen).

The stories often contained situations of loss (being deprived of something) and separation anxiety. At the Rorschach test the subjects displayed a significant sense of insecurity, which was mainly expressed by ideational perseveration, often dominated by hypochondriac thoughts (lacerations, breaks in 5 cases), with accentuation of symmetry (5 cases) and *chiaroscuro* responses (5 cases).

Finally, we didn't treat the correlation between IQ and psychopathological problems of study group, because it wasn't statistically significant. This analysis will be the subject of a subsequent study.

Discussion

Many studies have assessed the presence of psychic disorders in DMD patients. The literature shows that about 30-40 % of DMD subjects are mentally retarded.[4,14-19]

The non-progressive cognitive deficit mainly affects verbal abilities, therefore memory and language functions are the most severely impaired; the scales of patients with DMD show low scores in verbal and visuo-spatial memories, comprehension, arithmetic and vocabulary.[15-22]

Several hypotheses have been put forward to identify the cause of cognitive deficit in DMD, involving biological, emotional, familiar and social factors. Schooling and poor social relationships prevent them a normal social life. This, although while is not the cause of the intellectual deficit, affect the degree of the deficit itself.

Among the biological hypotheses mention should be made of the biochemical hypothesis, which attributes the neuropsychological disorder to dystrophin deficit which also occurs in the neurological structures involved in the regulation of emotions, memory and strategy processing[23-24] and the genetic hypothesis, which attempts to establish a correlation between the type of deletion/duplication and intelligence level;[25-27] some studies have tried to establish a correlation between the distal deletion of dystrophin gene and low intelligence levels.[25-28]

Studies on the personality of DMD children have pointed out some characteristics: dysphoric and disharmonic states, marked affective tension, feelings of self-compassion, hypochondriac fears, scepticism and pessimistic ideas, these affective disorders testify how personality is affected by the disease.[29-30]

Depressive issues and narcisistic fragility are often present in DMD subjects and seem to impair cognitive skills.[31-32]

The first symptoms of the disease affect the children's ability to project their life in space and time, as could be observed in the drawings and in the cognitive performance tests.[32,33]

Loss of ambulation is the most dramatic moment.[15]

At the Rorschach, children exhibited significant, original and creative responses (*a man who is climbing up*), thus becoming an autonomous human being.[15,34]

It is clear that in the Rorschach more than the other tests the personality of these children emerged, with their suffering and their death anxiety.

Their imaginary accounts of overcoming difficulties contrasts with their everyday life and with their families who, faced with the worsening of the disease, became increasingly withdrawn and take little advantage of the few therapeutic options available (physiotherapy, support psychotherapy). Moreover, this attitude exacerbates both the child's difficulties in relating with peers and his/her dependence on the maternal figure who, in turn, tends to overprotection.

Children and parents seem therefore to attend passively the progression of the disease, which its ultimate conclusion is the death.

Conclusions

The child may even have depression, especially to the seventh year of life when he becomes more conscious of being physically different from other children.

As reported in literature, since the motion is a mean to express themselves and then to communicate, it is possible that in these kids there are any deficiencies related not only to the movement but also to the motor expressiveness.[6,10,12] There is thus a tendency to increased aggression and refusal of contact with the outside.[12,22,29]

It is to remember also that many parents living disability as an injustice, as a drama that removes every other aspect of life and makes them victims.

At the first place there are certainly the problems associated with the disease: the need to provide uncommon cares and treatments; the recourse, but not always easy or satisfactory, to the health and rehabilitative services; the incessant research, almost endless, of specialized centers, sometimes very far from the residence; the uncertainty of diagnosis and prognosis made by different specialists, often in conflict with them.

The general psychological problems of developmental DMD patients are not very different from those of all children: they display marked problems regarding body schema, family adaptation, peer relationships, sexuality. DMD patients encounter more difficulty in establishing independence from their parents and if social contacts are limited they can sometimes use imagination and rejection as means of adaptation.[17,20-23,34]

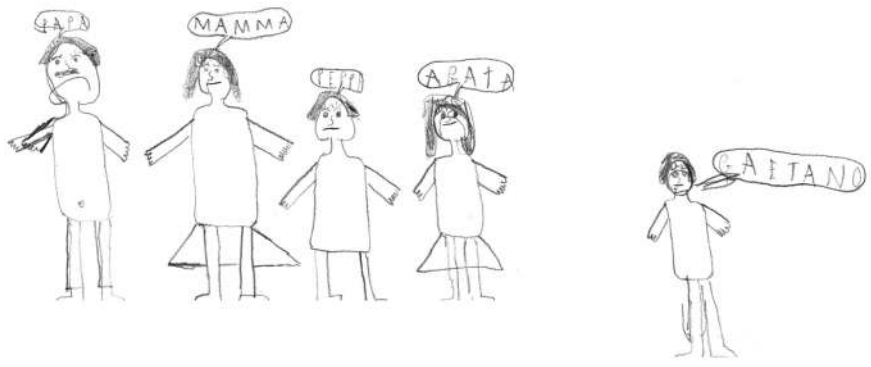

Figure 4. Drawing of child with Duchenne muscular dystrophy (Family Drawing Test).

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