CASE PRESENTATION

AUTISM SPECTRUM DISORDER AND DUCHENNE MUSCULAR DYSTROPHY
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ABSTRACT

Autistic spectrum disorder is a neurodevelopmental disorder characterized by difficulty with social relationships, difficulty understanding emotions, lack of visual contact, and expressive and / or receptive language impairment. Duchenne muscular dystrophy is an X-linked recessive degenerative neuromuscular disorder characterized by deficient dystrophin protein in the muscle. This paper presents the case of a two and half year old patient admitted to the Child and Adolescent Psychiatry Department of the “Prof. Dr. Al. Obregia” Psychiatry Hospital from Bucharest. The main symptoms the patient presented were delay in expressive and receptive language development, visual discontact, hyperkinetic behaviour, inability to initiate and maintain social contact with peers. The data obtained from the family, following clinical examination, laboratory investigation results and assessment of mental status were significant for the diagnosis of Autism Spectrum Disorder, hyperkinetic behaviour and Duchenne Muscular Dystrophy, according to ICD 10 and DSM 5 diagnostic criteria. In presenting this case, we are trying to highlight the importance of a multidisciplinary approach in this psychiatric and neurology comorbidity, and the challenging pharmacological treatment which required taking into account that many therapeutic factors can precipitate psychiatric symptoms.

Keywords: autism, Duchenne, delay language.

INTRODUCTION

The term of autism spectrum disorder describes a neurodevelopmental disorder characterized by impairments in social communication and a tendency to engage in repetitive behavioural stereotypes. Clinical presentation is diverse ranging from delay in learning how to speak, avoiding eye contact or using facial expressions that don’t match uttered messages, social difficulties, lack of imitative play, the presence of repetitive body movements (hand flapping, rocking, spinning), preoccupation with a narrow topic of interest, lack of empathy. These symptoms can change over time. Children with ASD develop at different rates in different areas. They may have delays in language, social, and learning skills, while their ability to walk and move around is almost the same as in other children their age.

In terms of cognitive difficulties, these are common but can vary greatly. Prognosis is reserved and acquisitions are slow if autistic symptoms are associated with mental retardation.

The incidence of autism is higher in boys than in girls, but girls are more severely affected and have lower intelligence scores [1].

The final diagnosis is established based on a clinical assessment doubled by instrumentation questionnaires like ADI-R (Autism Diagnostic Interview-Revised) (which is applied to the parents) and ADOS (Autism Diagnostic Observation Questionnaire-Revised) (which is applied to the child).

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Duchenne muscular dystrophy is an X-linked recessive degenerative neuromuscular disorder characterized by deficiencies in muscular dystrophin protein. Dystrophin is found in greater quantity in the skeletal and cardiac muscle and in small quantity in smooth muscles. It is essential in establishing a connection between intracellular contractile apparatus, sarcolemma membrane and extracellular matrix. Lack of protein and membrane fenestration can cause embrittlement during contraction or myofibrillar relaxation [2].

Duchenne muscular dystrophy has a prevalence of 1 in 35,000 males and is characterized by motor impairments, difficulties in walking, waddling, toe walking, difficulties in climbing stairs, running or lifting off the ground. Paraclinically, an increase in serum creatinine level can be observed. In two thirds of the cases, the final diagnosis is based on DNA analyses from peripheral blood samples by PCR, the result showing a deletion in Xp21.

The presence of cardiomyopathy can be considered a particular symptom in muscular dystrophy, so a neuromuscular evaluation is necessary in this case. It may be asymptomatic until the final stage of the disease. Scoliosis is a complication that develops especially after the loss of ambulation and should be regularly radiologically assessed [2].

There are few studies on the incidence of neuropsychiatric comorbidities in male populations with Duchenne muscular dystrophy [3]. In 2000, Mehler concluded in his review about the current studies of dystrophy that patients "have significant cognitive and behavioral abnormalities in their development, including increased frequency of autism and attention deficit disorder". [4]. In 2005, Wu JY and colleagues identified six male patients with autism and Duchenne muscular dystrophy using diagnostic criteria from the DSM IV in a sample of 158 individuals with Duchenne muscular dystrophy in Massachusetts, which accounts for a rate of frequency of 3.8% [5].

CASE PRESENTATION

We report the case of a 2 year and 4 month old boy, diagnosed with Autism Spectrum Disorder and Duchenne Muscular Dystrophy.

The patient, accompanied by his parents, referred to our clinic in March 2016, due to delays in expressive and receptive language development, failure to make eye contact to others, hyperkinetic behaviour, consistent irritation and general discomfort, failure to respond even when his name is called.

Physiological personal history:

Only child, pregnancy with pathological evolution, imminent risk of miscarriage at 3 months of pregnancy, recommendation for hormonal treatment and bed rest to maintain pregnancy, birth at 36 weeks (caesarean section), birth weight: 2800g, Apgar Score 9, difficult adjustment to extra uterine life. Delayed psychomotor development in different stages of age: he sits upright at 6 months of age, he walks unassisted at the age of one year and one month, after kinetotherapy intervention. The child had not acquired the ability to pronounce words or sentences by the time he was brought to hospital.

Pathological personal history:

1. Atopic Dermatitis
2. Muscular dystrophy
3. Left plagiocephaly
4. OS cyst surgery

Mental State Examination:

At the examination, the patient is uncooperative, failing to respond, even when his
name is called, he can’t establish eye contact with the examiner or other children. He can’t understand or execute simple or complex commands; absence of expressive language – he does not use words with meaning (babbling “ma” “da” “pa”). Cognitive: he can’t show his or somebody else’s body parts, he can’t recognize animals, colours or geometrical figures, can’t show objects around him by pointing them. Difficulties with basic skills such as dressing, eating, brushing teeth, and going to the bathroom. He depends on his parents in his daily routine. Socialization: he accepts the presence of other people/children, but he cannot initiate play with other children and we observed the absence of symbolic/imaginative/pretend play and preference for playing alone. We observed the absence of pointing, gesturing or waving goodbye. Motor behaviour: hyperkinetic behaviour, throws tantrums or cries for extended periods, often due to a lack of understanding or control. Fine motor skills are dominated by repetitive movements: spinning, rocking, or hand flapping.

Neurological examination: possible walking (postural with broader base of support), hyperlordosis, stepping only on a part of the foot; rise from the squat with support, specifically Gowers positive; proximal leg muscle deficiency; bilaterally slight limitation of ankle plantar flexion; humeral level without contractures; MS proximal muscle deficiency; genetic test for progressive Duchenne muscular dystrophy –positive.

EEG line performed while the patient was awake – without pathological graphic elements.

Psychological assessment – the diagnosis was instrument by assessing the child’s pathology with ADOS instrument, which revealed a score=12, score GAC-44.

The positive first axis diagnosis was set to Autism spectrum disorder and Hyperkinetic behaviour. Second axis of diagnosis: Delay in mental development and the third axis: Duchenne progressive muscular dystrophy.

The practitioner could potentially diagnose autism spectrum disorder as a somatic or neurological disorder (cerebral palsy, hydrocephalus, severe cortical agenesis, genetic disorders like phenylketonuria or metabolism disorders - metachromatic leukodystrophy, tuberous sclerosis) but laboratory tests were within normal limits and excluded these diagnoses. Autism spectrum disorder should be differentiated from disorders of language development (the child has impaired understanding and expression but the absence of stereotypes and autistic mentally discontact exclude such diagnosis), reactive attachment disorder (favourable evolution with care and emotional support), sensory disturbances (deafness, blindness). (6)

Psychopharmacological treatment:
During hospital admission, the child received low doses of atypical antipsychotic (risperidone) to reduce psychomotor agitation, stereotypes and improve relationships. After the psychological assessment, the family was trained in special educational skills and was informed about the future therapeutic plan.

Discharge recommendations consisted in continuing psychopharmacological treatment doubled by applied behavioural therapy and speech therapy. Simultaneously, the paediatric neurologist recommended glucocorticoid treatment and dietary supplements.

Prognosis and evolution:
The evolution of the patient may be favourable if the family complies with the recommendations. As a positive prognostic factor, we can mention family support and early intervention. But comorbidity with hyperkinetic behaviour, muscular dystrophy and different response to therapy are adverse prognostic factors.
Children with ASD may experience an improvement in social networking and appropriate expressive language development, but they never reach an optimal level of functioning, requiring constant surveillance from the family [6].

Despite modern advances in gene therapy and molecular biology, muscular dystrophy remains incurable. With proper care and attention, patients can have a better quality of life but around the age of 30 years die from cardiopulmonary complications [7].

CONCLUSIONS

In presenting this case, we want to highlight the importance of a multidisciplinary approach in this psychiatric and neurology comorbidity, and the challenging pharmacological treatment which required taking into account that many therapeutic factors can precipitate psychiatric symptoms.

The patient will require further recurrent evaluation (paediatric neurology and paediatric psychiatry) as well as paediatric examinations in order to keep under observation any potential cardiac or pulmonary complications.

REFERENCES

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