Introduction

The differential diagnosis of Autism Spectrum Disorder (ASD) is a complex process, and it is important to consider that there are many neurological, metabolic or genetic disorders that share autistic symptoms. Metachromatic leukodystrophy (MLD) is a neurodegenerative lysosome storage disorder with an autosomal recessive pattern, reported in 1 in 40,000–160,000 individuals worldwide.[1] The juvenile form of MLD shares common symptoms with a great number of psychiatric disorders, making the path to diagnosis quite challenging.

Case description

A 14 year old boy was referred to our clinic for the following problems:

• impaired social interaction with peers
• language problems, monotonous speech, inability to control voice volume
• falling grades and difficulties in usual self-care activities

He was examined by a child psychiatrist at the age of 12 for the same difficulties, and received a diagnosis of Asperger syndrome. The other psychiatric findings, beside social impairment seemed to develop over the last 2 years, with important impact on global functioning. These impairments became more severe over time, despite therapeutic intervention.

Diagnostic assessment

Clinical interview:

No significant personal or family medical history. Psychomotor development was normal, except for social impairments which seemed to be present from an early developmental stage.

The clinical examination revealed ligamentous laxity of the hands with ulnar deviation of both hands and fingers, important dyspraxia, bilateral intention tremor slightly dysmetria and scoliosis.

Psychiatric examination:

At present time, the psychiatric evaluation showed:

• temporo-spatial disorientation
• rare eye contact (he was gazing around the room most of the time), no coordination between the eyes and the action he was performing (he was looking on the walls while trying to unbutton his shirt)
• hypoprosxia (one needed to repeat the questions multiple times), bradydylia and bradypsychia
• monotonous speech and inability to control voice volume, rare spontaneous speech, poor vocabulary and echolalia
• no interest in examining the environment or talking to other people
• no interest in playing with toys or doing age-related activities
• cognitive performance under his age, with important deterioration in the past 2 years

Psychometric tests

The psychological examination revealed:

• an IQ of 78 at the age of 12
• an IQ of 60 at the age of 14, the difference being suggestive of a cognitive decline over time.

Paraclinical investigations

Brain MRI showed extensive bilateral and symmetrical T2 hyperintense changes in the periventricular white matter and centrum semiovale, and atrophic corpus callosum suggestive for a leukoencephalopathy, most likely metabolic. A brain MRI was performed two years prior the current evaluation, which revealed no significant changes, so the present modifications are indicative for a progressive disorder.

The MRI findings correlated with the clinical presentation, was indicative of metachromatic leukodystrophy – juvenile form. In order to confirm the diagnosis, a dosage of arylsulfatase A in the blood, urine and leukocytes was performed. The deficiency of arylsulfatase A was identified in urine and also leukocyte arylsulfatase A activity was low.

The clinical picture together with the MRI abnormalities and lab findings are highly suggestive of metachromatic leukodystrophy – juvenile form.

Discussions

This clinical case outlines the importance of the differential diagnosis of neurodevelopmental disorders, mostly because they are associated with different outcomes. Thus, in atypical psychiatric presentations with poor improvement of symptoms after therapeutic intervention, neuroimaging assessment should be considered. Early detection of associated neurological disorders may be important for treatment and outcome.