



A 3 Years Old Girl with Autism Spectrum Disorder and Positive Thioglycolic Acid in Urine

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Abstract

Autism Spectrum Disorder (ASD) is a neurodevelopment disorder that must be present from birth or early childhood according to DSM-V classification.

Behavior problem is common in ASD patients. Trichotillomania, is also a very important complain in this disorder.

Thioglycolic acid (TGA) is used as hair fixer. Is an active ingredient of permanent-waving solution (PWS). Hui-Fang Gan., *et al.* in 2003, studied the effect of TGA-containing PWS on the health of a human population. He concluded that the reproductive function of hairdressers may be affected by long-term exposure to PWS, probably due to the presence of TGA, and more attention should be paid to its potential carcinogenic effects.

Our index case is a 3 years old boy with ASD, that has trichotillomania and we found in his urine high amounts of TGA secondary to the persistent swallow of her mother hair, that contains this product as part of the composition of hair fixer.

In this situation, we recommend the mother to avoid use hair fixer and also try to change the behavior of the child with this tendency to swallow her hair daily. This component can affect the reproductive function by long-term exposure to PWS, and even can have potential carcinogenic effects.

Keywords: Autism Spectrum Disorder; Trichotillomania; Thioglycolic Acid

Introduction

Autism Spectrum Disorder (ASD) is a neurodevelopment disorder that must be present from birth or early childhood according to DSM-V classification.

The ASD according to the new definition, include autistic disorder (autism), Asperger disorder, childhood disintegrative disorder and pervasive developmental disturbance without further specification. In 75 - 80% of cases, the etiology is unknown.

Cytogenetic examinations, metabolic study, neuroimaging and electroencephalogram (EEG) are the exams that we need to do in a patient with criteria of ASD.

Inborn error of metabolism are rare causes of ASD with Mitochondrial disease responsible for around 5% of cases [1].

Thioglycolic acid (TGA) is used as hair fixer. Is an active ingredient of permanent-waving solution (PWS). TGA has been shown

to be a chemical of high toxicity, which can be absorbed through intact skin and cause damage to organs or systems in animals. Hui-Fang Gan., *et al.* in 2003, studied the effect of TGA-containing PWS on the health of a human population. The results suggest that the reproductive function of hairdressers showed risk to be affected by long-term exposure to PWS, due to the presence of TGA, even can have potential carcinogenic effects [2].

Case Report

A 3 years old boy with ASD according to DSM-V classification was sent to the Genetic consultation to evaluate a possible etiology of the disorder. The patient showed a normal phenotype, with no significant changes on his physical examination.

The mother complained that he has an abnormal behavior, the so called trichotillomania, (can't resist the urge to pull out the hair on their or other people head, eyebrows or eyelashes).

The study for the ASD etiology consisted of determination of fragile X, microarray, amino acid in blood and urine and organic acid in urine.

The results were all normal, with the exception of organic acid in urine that showed large amount of TGA.

Discussion

The identification of ASD is of great importance for the early onset of the intervention and consequently, a more favorable prognosis.

In the last 25 years, the numbers of cases of ASD increased from 0.5/1000 to 9/1000 under the age of 8 years. The prevalence is 4(M)/1(F).

In some cases, patients with ASD also can have mental retardation, epilepsy, structural cerebral malformations, dysmorphism, microcephaly and macrocephaly [1].

In personnel data study of one year review of ASD cases, we detected 9 patients out of 22 with an etiology for ASD, 8 with chromosome disorders (deletions or duplications) and only 1 with metabolic disorder (Hunter disease). The percentage of cases with etiology was 40.9%, more than we expected if we compared with the average prevalence (25 - 30%) [3].

Behavior problem is common in ASD patients. Trichotillomania, is also a very important complain in this disorder.

In our index case, the finding of TGA in urine was secondary to the persistent swallow of her mother hair that contains this product as part of the composition of hair fixer.

Is this situation, we recommend the mother to avoid use hair fixer and also try to change the behavior of the child with this tendency to swallow her hair daily. We can see in the study of Hui-Fang Gan., *et al.* in 2003 that this component can affect the reproductive function by long-term exposure to PWS, and even can have potential carcinogenic effects [2].

Conclusions

In our index case, we didn't found an etiology for ASD, but the result of organic acid in urine, showed high amount of TGA secondary to trichollomania. This product is use in hair fixer and may cause potential carcinogenic effects and we need to avoid it.

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