Autism Symptoms Related to Tyrosinemia Type III: A Case Report

Tirozinemi Tip-III ile İlişkili Otistik Belirtiler: Bir Olgu Sunumu

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Abstract

The published literature on tyrosinemia type III consists of only a few case reports. In this report, we present a patient with tyrosinemia type III, autism, and mental retardation. This patient's speech improved and his autistic symptoms lessened on a tyrosine-restricted diet, although his mental retardation remained unchanged. This is the first published report of a patient with tyrosinemia type III and autism. This observation is significant due to the paucity of published information about tyrosinemia type III. *Turk Jem 2008; 12: 55-6*
Key words: Tyrosinemia type III, autism

Özet

Tirozinemi Tip-III ile ilgili bilgilerimiz sadece bir kaç olgu sunumu ile sınırlıdır. Bu çalışmanın amacı Tirozinemi Tip-III, Otizm ve Mental Retardasyon tanıları konan bir olgu ile ilgili bilgi vermektir. Olgunun otistik belirtileri trozinden fakir diyetten sonra düzelme gösterirken mental retardasyon belirtilerinde düzelme gözlenmemiştir. Bu olgu, otistik belirtiler bildirilen ilk Tirozinemi Tip-III olgusudur. Bu bilgilendirme hakkında çok az şey bildiğimiz Tirozinemi Tip-III için önemlidir. *Turk Jem 2008; 12: 55-6*

Anahtar kelimeler: Tirozinemi tip-III, otizm

Introduction

Tyrosinemia type III, is an autosomal recessive disorder characterized by elevated tyrosine in the blood, and massive excretion of tyrosine metabolites in the urine. It is a rare disorder caused by a deficiency 4-hydroxyphenylpyruvate dehydrogenase (1,2). Tyrosinemia type III is represented in the literature by only a few case reports. We present a patient with tyrosinemia type III and autistic symptoms, who was diagnosed with tyrosinemia type III during clinical follow up. His autistic symptoms improved with dietary restriction for treatment of tyrosinemia.

Case Report

G.E.A. was a two-year-old boy whose parents reported head banging, flapping, and spinning, from the age of 15 months. During this time period, he did not respond to his own name. He knew 10-12 words and he had echolalia. He noticed people, but he did not communicate with them. His parents reported that he did not communicate well with other children his own age. When he became angry, he would fall down and hit his head on the ground or on other objects. Moreover, he habitually moved objects to revolve around himself, and exhibited stereotypical behaviors in manipulating those objects. He habitually grouped his toys and placed them in order in neat lines. He enjoyed TV commercials and cartoons, and especially, happy, upbeat songs on television. He was relatively insensitive to pain.

The Ankara Developmental Test (AGTE) was administered to G.E.A. when he was two years old. This test, whose validity has been verified, was developed for Turkish children. The AGTE assessment of his developmental age was 18 months. AGTE was administered again when he was 4 years and 3 months old, at which time his developmental age was 2 years and 8 months.

We found elevated levels of tyrosine both in serum and in urine by amino acid analysis. The patient was referred to the Child Endocrinology Department, and tyrosinemia type III was diagnosed by a pediatric endocrinologist. A special tyrosine-restricted diet, which included special infant foods, was recommended, and detailed dietary information was provided to the family. With regard to manifestations of autism, the whole family was given guidance on how best to care for the patient. In the 6-month follow-up evaluation, it was observed that his speech and autistic symptoms

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had improved on the tyrosine restricted diet, although his mental retardation remained unchanged. The family did not return to the Endocrinology Department for further evaluation and follow-up.
The family last visited our clinic when the patient was 9 years and 2 months old, at which time the autistic symptoms had entirely disappeared, and the patient was hyperactive. Wecshler (WISC-R) Intelligence Scale revealed an intelligence quotient of 65.

Discussion

To our knowledge, our patient is the first patient with tyrosinemia type III and autistic symptoms reported in the literature. In tyrosinemia type III, the most common symptoms are mental and motor developmental retardation, dyslexia, hyperactivity, and speech delay (1,3,4). Our patient had mental and motor retardation and hyperactivity symptoms as described in the literature; however, autistic symptoms are not mentioned in any other case report of tyrosinemia type III. This observation is significant, considering how little is known about tyrosinemia type III. The fact that a therapeutic low-tyrosine diet and special infant foods ameliorated the autistic symptoms in our patient suggests that these symptoms were related to his type III tyrosinemia. When evaluating children with autism, rare disorders such as tyrosinemia type III should be considered.

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