CASE REPORT
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Autism in a Child with Common Variable Immunodeficiency

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ABSTRACT

Autism is a neurodevelopmental disorder, characterized by poor social interaction and communication impairment and repetitive behavior. Autism is considered as a genetic and multifactorial disorder, with diverse risk factors involved.

Herein, we report a 13-year-old male with common variable immunodeficiency (CVID), who was diagnosed with autism at the age of 3 years old.

As there are some evidences about the role of the immune system defects in the pathogenesis of autism, specific primary antibody deficiency diseases such as CVID might predispose some affected cases to this neurodevelopmental disorders.

Keywords: Antibody Deficiency Syndrome; Autism; Common variable immunodeficiency

INTRODUCTION

Autism is a neurodevelopmental disorder, characterized by weak social interaction, verbal and non-verbal communication impairment and restricted and repetitive pattern of behavior; the symptoms are usually presented by the age of 3 years. The screening and surveillance policies have led to early detection and a better outcome.1 Autism is considered as a multifactorial disorder, with diverse risk factors. Defects in immunity are a recently discovered entity with increasing evidences in the literature.2-5

Common variable immunodeficiency (CVID) is the most common symptomatic primary immunodeficiency, characterized by hypogammaglobulinemia and impaired antibody responses,6-9 which may associate with some neurological disorders.10,11 Herein, we report a CVID patient who has been suffered from autism since the age of 3 years.

CASE REPORT

A 13-year-old male with CVID from non-consanguine parents is presented here, who had history
of recurrent infections since early childhood. He had a history of acute otitis media, diarrhea and vomiting since 10 months old. He was admitted at 2 years of age due to pneumonia. When the child was 3-years old, the diagnosis of autism was established based upon DSM-IV criteria including the decline in social interaction, language, symbolic and imaginative play. The next admission was due to septic arthritis in left knee 2 years later. The first episode of seizure started at age 4.5 years, accompanied with a decrease in level of consciousness. The seizure was tonic contracture of upper limbs. Brain CT scan revealed mild brain atrophy. Since then, he was on the maintenance therapy of seizure. The diagnosis of CVID was established in 5th year of his life based on the immunological studies (IgG <100 mg/dL, IgA <10 mg/dL, IgM =20 mg/dL, and CD3+ lymphocytes =65.14%, CD3+CD4+ lymphocytes =43.86%, CD3+CD8+ lymphocytes =32.01%, CD19+lymphocytes = 5.18%). Although the treatment with intravenous immunoglobulin was started, the patient suffered from some episodes of cellulitis, diarrhea, conjunctivitis, oral thrush and sinobronchitis after the time of diagnosis.

**DISCUSSION**

Autism is one of the neurodevelopmental disorders, characterized with defects in three developmental fields: socialization, communication and behavior. Although exact etiologies of autism are unclear, both genetic and environmental factors seem to be involved.1

There is increasing evidence in the literature indicating the role of immune system defects in the pathogenesis of autism. Humoral and cellular immunity defects have been shown in small studies among autistic children, revealing decreased immunoglobulin production or B- and T- cell dysfunction.12 Heuer et al in 2008 showed that children with autistic disorder had significantly reduced levels of plasma IgG and IgM, compared to normally developed children.3 Immunoglobulin levels were negatively correlated with Aberrant Behavior Checklist scores for all children, meaning the lower Ig level could be associated with more severe symptoms.3

The association between autism and some primary immunodeficiency disorders has previously been reported. This association with hyper-IgE syndrome was discussed by Grimbacher et al.13 who showed that proximal chromosome 4q is a candidate region for genes involved in both disorders.12 Chromosome 4q is also considered as a possible loci for dominant CVID genes.14 In the study of IgA deficient patients, Santealla et al showed that there is a higher occurrence of autism in the IgA deficient group, compared to control group.5 Even intravenous immunoglobulin has been proven to improve autistic behavior.5 More epidemiologic and genetic studies are required to establish the association between autism and CVID.

**REFERENCES**

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