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Autism: Facts and a Hope

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Introduction

When the editorial board invited me to write this article, I remembered my autistic patients and their families. I remember how much the parents suffered to help their autistic kids. I always have a dream, are we able to cure these lovely children?

To write an article about this interesting topic many questions have to be answered, the first one is; *what is Autism?*

Autism is a neurodevelopmental disorder characterized by impaired social interaction, verbal and non-verbal communication, as well as restricted and repetitive behavior [1].

The prevalence of autism spectrum disorder (ASD) differ from country to other one. According to estimates from CDC's Autism and Developmental Disabilities Monitoring (ADDM) Network, about 1 in 68 children has been identified with ASD [2]. Studies published from other countries worldwide give an estimate of 1-2.6% [3].

Another question is; *what is the etiology of autism?*

Autism is considered a genetically determined disease and the exposure to environmental modifiers contribute to the variable clinical presentations of the disease [4].

Evidences of genetic etiology for autism come from its association with some genetic disorders. Autism symptoms are present among some patients who have tuberous sclerosis [5], fragile X chromosome [6], 15q chromosome duplications [5], and 15q chromosome deletions which is known as Angelman syndrome [7].

On the other hand, environmental factors may have a role acting as a "second-hit," modifying the effect of existing genetic factors leading to ASD [8,9]. The effects of environmental factors depend on many factors including; how do this factor work and how much is its concentration? [10].

Environmental insults may include infections, teratogens and toxins but some authors believe that environmental insults have a minor role as etiological factor for autism [11].

For some time MMR was blamed as etiological agent causing autism but well-designed studies have found MMR is innocent in this regards [12,13].

How these genetic and environmental factors interact, is not clear. But there are evidences of decreased numbers of Purkinje cells in the cerebellum and abnormal structure in the prefrontal and temporal cortex. These areas have a role in language processing, as well as social, emotional and communication skills [14,15].

When Do We have to Screen for Autism?

According to American academy of pediatrics, all children at 18 and 24 months of age should be screened for autism, the age of early social and language development.

But we should conducted screening for ASD for all children presented with delayed language, communication milestones or in children with a regression in social or language skills [16].

Autism clinical features

Among clinical spectrum for autism, impaired social communication is a characteristic sign for ASD [17] as well as delays language development [18].

Autistic child has a reduced spontaneous desire to share enjoyment and interests with other people as well as deficits in social or emotional reciprocity [19,20].

One important stigmata of ASD is failure to develop and maintain peer relationships [20].

Restricted, repetitive and stereotyped patterns of behavior [17], as well as impaired ability to use and interpret nonverbal behaviors such as facial expression and eye-to-eye gaze are other features of autism [20].

Another stereotyped behavior linked to autism is repetitive complex movements like toe walking, as well as repetition of daily activities in the same pattern such as using same way to go from one place to another, and eating same foods without any desire to change [21].

Impaired Cognitive skills is another hallmark for ASD with variable presentation reflecting poor reasoning and interpretation of higher mental centers [22].

Autistic child has different degrees of impaired spoken language and misdiagnosed as hearing defect [23].

The association between autism and macrocephaly is interesting subject. Macrocephaly is present in about a quarter

of autistic patients but, it is not one of diagnostic features [24]. Another important point in this regards is the presence of mutations in the *PTEN* gene in some autistic patients who have macrocephaly. This gene mutation is a risk for hamartomatous tumor syndromes [25].

In spite of all poor stigmata reflecting impaired poor skills, autistic patients may have some well-developed skills like arts and music [26].

Prognosis

It is very difficult to predict outcome for autistic patient. But some factors have been associated with positive outcomes. Among these predictors are; early identification, decreased severity of ASD symptoms and presence of joint attention. Higher cognitive abilities and functional play skills are other positive predictors [27,28].

Management

Improving the quality of life for autistic child is an ultimate goal in the management plan. Multidisciplinary team and approach are required to improve the defective behavior, cognitive and communication skills as well as social adaptation [29].

Although no defined standard approach exists for autism, available treatments can be divided into behavioral, nutritional and medical approaches [30].

Every day, some new approaches are advocated to help autistic patients and the treatment for autism is active interest in medical researches

Medical teams who are caring autistic children know well that there is no absolute cure, but their aim is to decrease the symptoms over time and if possible to minimize the patient symptoms and decrease his disability [31].

But is there a hope to cure autistic child in the future?

Children with autism have been consistently shown to have impaired or subnormal CNS circulation as well as resulting hypoxia. Defects include basal hypo perfusion and decreased perfusion in response to stimuli that under normal circumstances upregulates perfusion. In numerous studies the areas affected by hypo perfusion seem to correlate with regions of the brain that are responsible for functionalities that are abnormal in autism [32].

Theoretically, it is conceivable that reversing hypoxia may lead to activation of self-repair mechanisms. The concept of increasing oxygen to the autistic brain through various means such as hyperbaric medicine is currently being tested in 2 independent clinical trials in the US [33,34].

Nowadays, stem cell therapy represents the great promise for the future of molecular medicine. Among the stem cell population, mesenchymal stem cells show probably best potential good results in medical research. Due to the

particular immune and neural dysregulation observed in ASD, mesenchymal stem cell transplantation could offer a unique tool to provide better resolution for this disease. However, personalized stem cell therapy will be the most effective treatment for a specific autistic child, opening a new era in autism management in the next future [35].

There are some trial for stem cell therapy for autism that have generated anecdotal evidence of stem cell therapy for autism, and through development of a potent clinical study with appropriate endpoints, much will be learned about the pathophysiology of autism regardless of trial outcome [32].

At the end, in the future we have a lot of hopes and a lot of questions.

Do we have the ability to understand in depth the genetic background of ASD?

Do we have the ability in the near future to cure our children who have ASD?

Finally I still have a dream, which is an old one, and the answer will come in the near future with the help of the valuable and enthusiastic researches in the field of ASD

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