

Autosomal Recessive Autism: Cure of the Major Autistic Features

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Abstract

Background: We have previously reported our extensive experiences with autism disorders and their treatments, and we showed the possibility of curing the major autistic features with a new therapeutic approach which included individualized courses of intramuscular cerebrollysin as the main therapy for the main autistic features. Our previously published experiences included observing the occurrence of atypical autism associated with evidence of mental retardation in four brothers from outside Baghdad, each two of them from two unrelated families. The four patients were treated based on our extensive published experiences with the pharmacological treatments of autism disorders; however, the follow-up and the outcome of treatments of these four children were not described in the previous publications. The aim of this paper is to report the cure of the major autistic features in 2 brothers with autosomal recessive autism.

Patients and methods: It was possible to follow the first two brothers for only few weeks and to see them after the first course of treatment, however, the second brothers were followed for more than one year, and it was possible to achieve cure of the main autistic features. **Results:** After one short course, the first two brothers showed slight but noticeable improvement. The younger brother showed some reduction in autistic features as he developed rather an acceptable eye contact. However, the two brothers were still not responding to name and didn't say any word. After, treatment, the second two brothers showed no autistic features, and experienced significant improvements in speech and cognitive functions. The older brother was not having autistic features after eighth months, and was obeying commands, but was still saying only few words. The younger brother joined primary school and was doing well apart from some behavioral problems especially running away from school and sometimes breaks things. **Conclusion:** In this paper, cure of autism has been achieved in two boys with autosomal recessive disorder. However, cure of autism in older children has never been expected to totally normalizes them, as the patients have already lost several years of learning, social adaptation, and maturation of personality and behavior.

Keywords: Autism, Major, Autistic, treatments.

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INTRODUCTION

Autistic disorders which are also called "Pervasive developmental disorders" are a very complex and heterogeneous group of disorders characterized by early impairments in social interaction and communication and behavioral problems which are clinically dominated by the main autistic feature which include poor or no eye contact, poor or no response to name, and poor or no speech development. However, patients with Asperger syndrome have good speech development. A child with acceptable eye contact and acceptable response to name can not receive the diagnosis of an autism disorder [1-10].

We have previously reported our extensive experiences with autism disorders and their treatment, and we showed the possibility of curing the major

autistic features with a new therapeutic approach which included individualized courses of intramuscular cerebrollysin as the main therapy for the main autistic features. Courses of intramuscular cerebrollysin were individualized according to the age and severity of the illness, and with aim of improving social interactions including response to name, looking at faces, and eye contact.

It was speculated that improvements in social interaction can contribute to improving other features of autism, especially verbal communication and speech.

Many patients with autism disorders need neuroleptics to control hyperactivity, anxiety and other abnormal behaviors. Trifluoperazine and prochlorperazine were used frequently. Risperidone was

also used in patients with severe disorder. Some patients also receive citicoline as an adjunctive therapy to improve speech development and cognitive functions, and piracetam can also be given to improve cognitive functions in atypical autism with mental retardation [11-13].

Our previously published experiences included observing the occurrence of atypical autism associated with evidence of mental retardation in four brothers from outside Baghdad, each two of them from two unrelated families. The diagnosis of atypical autism was based on evidence of mental retardation as indicated by poor and late development of adaptive behaviors including bowel control and spoon feeding [12, 13]. The four patients were treated based on our extensive published experiences with the pharmacological treatments of autism disorders; however, the follow-up and the outcome of treatment of these four children were not described in the previous publications [12, 13].

The aim of this paper is to report the cure of the major autistic features in two brothers with autosomal recessive autism.

PATIENTS AND METHODS

It was possible to follow the first brothers for only few weeks and to see them after the first course of treatment, however, the second brothers were followed

for more than one year, and it was possible to achieve cure of the main autistic features.

The first two brothers (Figure-1) whom were first seen on the 25th of July, 2019, aged ten and seven years respectively.

The older brother (Figure-1A) had severe autistic feature and had no eye contact and was not responding to name, and displayed abnormal behaviors with sitting in an abnormal attitude on the chair at the clinic. He had evidence of mental retardation as he had no bowel control, and was still unable to eat with spoon.

The younger brother had less severe condition, but he was also not responding to name, but he had some eye contact. He also had evidence of mental retardation as he was still unable to eat with spoon, but he was able control bowel. The younger brother was rather anxious at the clinic.

The two brothers were not saying any word, and were not responding to commands nor understanding speech. Both brothers received intramuscular cerebrolysin 5ml daily for 20 days plus oral trifluoperazine 1mg daily in the morning. Additionally, the older brother received oral risperidone 1mg at night, and the younger brother received oral citicoline 300 mg daily in the morning.



Fig-1: The first two brothers were first seen on the 25th of July, 2019

The second two brothers whom were first seen on the 22nd of August, 2019, aged eleven and eight years respectively. The older brother had severe disorder with no eye contact and was not responding to name, and had displayed significant repetitive movements at the clinic (Figure-2). He was not saying any word, was still unable to control bowel nor was able to eat with a spoon.

The younger brother had less severe disease as he had some eye contact, but was not responding to name. He had poor speech development which started at the age of six years. He achieved bowel control late at the age of five years, and he was able to eat with a spoon. Table-1 shows the treatments of the second two brothers.



Fig-2: The older brother had severe disorder with no eye contact and was not responding to name, and had displayed significant repetitive movements at the clinic

Table-1: The treatments of the second two brothers

THE OLDER BROTHER
First month
Intramuscular cerebrolysin 5 ml daily for 10 days. Intramuscular cerebrolysin 5 ml every other day, 10 doses (20 days). Oral trifluoperazine 1mg daily in the morning. Oral prochlorperazine 5 mg in the afternoon.
Second and third months
Intramuscular cerebrolysin 5 ml every third day, 20 doses (60 days). Oral trifluoperazine 1mg daily in the morning. Oral prochlorperazine 5 mg in the afternoon. Risperidone 1 mg at night.
Fourth , fifth and sixth months
Intramuscular cerebrolysin 5 ml every third day, 30 doses (90 days). Oral trifluoperazine 1mg daily in the morning. Oral prochlorperazine 5 mg in the afternoon.
Seventh month
Intramuscular cerebrolysin 5 ml every third day, 20 doses (60 days). Oral trifluoperazine 1mg daily in the morning. Oral prochlorperazine 5 mg in the afternoon. Oral piracetam 800 mg once daily in the morning.
Eighth month
Intramuscular cerebrolysin 5 ml every third day, 20 doses (60 days). Oral trifluoperazine 1mg daily in the morning. Oral prochlorperazine 5 mg in the afternoon. Oral piracetam 800 mg once daily in the morning. Citicoline 300 mg once daily in the morning.
THE YOUNGER BROTHER
First month
Intramuscular cerebrolysin every other day, 15 doses (One month). Prochlorperazine 2.5 mg in the morning and 2.5 mg in the afternoon.
Second month
Intramuscular cerebrolysin 5 ml every third day, 10 doses (One month). Prochlorperazine 2.5 mg in the morning and 2.5 mg in the afternoon.
Third month
Intramuscular cerebrolysin 5 ml every third day, 10 doses (One month). Citicoline 300 mg once daily in the morning.
Fourth and fifth months
Intramuscular cerebrolysin 5 ml every third day, 20 doses (Two month).

RESULTS

After one short course, the first two brothers showed slight but noticeable improvement. The older brother experienced improvement in behavior and at the clinic he sat on the chair in a normal manner (Figure-3), and showed some anxiety instead of the indifference

observed at the clinic before treatment. The younger brother showed some reduction in autistic features as he developed rather an acceptable eye contact. However, the two brothers were still not responding to name and didn't say any word.



Fig-3: The older brother experienced improvement in behavior and at the clinic he sat on the chair in a normal manner, and showed some anxiety instead of the indifference before treatment



Figure-4: After eighth months of treatment, the older brother experienced significant improvements in cognitive functions, as he was understanding and responding to simple commands well. He sat when the mother asked him to sit

After the treatments in table-1, the second two brothers showed neither autistic features nor repetitive behaviors, and experienced significant improvements cognitive functions.

The older brother was not having autistic features or repetitive movements after eighth months of treatment, and was obeying commands and accepting shaking hands (Figure-4), but was still saying only few words, and was unable to answer when asked “What is

your name”. He also achieved bowel control and spoon feeding.

Figure-5 shows the younger brother after five month of treatment trying to copy a line, a circle, and a square. Thereafter, the younger brother joined primary school and was doing well apart from some behavioral problems especially running away from school and sometimes breaks things.



Fig-5: The younger brother after five month of treatment after disappearance of the autistic features. He was trying to copy a line, a circle, and a square

However, cure of autism at his age has never been expected to totally normalizes him, as he has already lost several years of learning, social adaptation, and maturation of personality and behavior.

DISCUSSION

In this paper, cure of autism was achieved in two brothers with autosomal recessive autism treated with courses of intramuscular cerebrolysin which was used with other adjunctive therapies including neuroleptics (Trifluoperazine, prochlorperazine, and risperidone), citicoline, and piracetam.

Cerebrolysin solution contains free amino acids (85%) and 15% biologically active low molecular weight amino acids including neuro-peptides (Brain-derived neurotrophic factor, glial cell line-derived neurotrophic factor, nerve growth factor, ciliary neurotrophic factor [14]. Cerebrolysin has been used safely with benefit in a variety of neuro-psychiatric disorders including idiopathic mental retardation [15, 16], cerebral palsy [17, 18], myelomeningocele [19], pediatric juvenile spinal muscular atrophy [20, 21], pediatric Charcot Marie Tooth disease [22, 23], kernicterus [24, 25], agenesis of corpus callosum with colpocephaly [26, 27].

Citicoline, which has been increasingly grouped with the water soluble B vitamins, and is regarded as a form of the essential nutrient choline. It has been increasingly used with noticeable benefits in the treatment of several pediatric neuro-psychiatric disorders including, pervasive developmental disorders including Rett syndrome, and kernicterus [28, 29].

Piracetam beneficial effects on impaired cerebral functions include improving neuronal and cognitive functions, increasing cerebral blood flow and oxygen consumption, improving neurotransmitters function and brain neurotransmission. Piracetam is not associated with important side effect nor has acute toxicity at the therapeutic doses. Piracetam has been used with important benefits in the treatment of cerebral palsy and other childhood neuro-psychiatric disorders [30, 31].

The occurrence of autosomal recessive cases of autism has been reported as early as 1985[32]. In 1988, Smalley et al. emphasized the genetic heterogeneity of autism disorders and their association with mental retardation, and the occurrence of autosomal recessive inheritance [33].

CONCLUSION

In this paper, cure of autism has been achieved in two boys with autosomal recessive disorder. However, cure of autism in older children has never been expected to totally normalizes them, as the patients have already lost several years of learning, social adaptation, and maturation of personality and behavior.

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