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Research Article

Goldberg Shprintzen Syndrome: A Novel Therapeutic Approach

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Abstract

Background: Goldberg Shprintzen syndrome is a very rare autosomal recessive syndrome associated with mental retardation, a distinctive facial abnormalities, Hirschsprung disease and neurological abnormalities. In a previous publication, we described the occurrence of the thirty fourth and thirty fifth cases of the syndrome in two Iraqi brothers. There are no known therapies that are associated with improvement in the neurologic disability and mental retardation in Goldberg Shprintzen syndrome. The aim of this paper is to describe the successful treatment of one of the Iraqi brothers with a novel therapeutic approach.

Patients and Methods: The younger brother was treated with aim of improving neurological abnormalities which included spasticity and inability to walk without support and also to improve mental disabilities which included poor cognitive abilities, and the lack of speech development. The younger brother was treated with an evidence-based therapeutic approach based on our extensive experiences with treatment of brain damage and mental retardation.

Results: After about six months, the older brother who received only nutritional support and muscle relaxants showed slight improvement in spasticity and general health, but he didn't experience any improvement in motor and mental-cognitive disabilities, and he remained unable to stand and didn't show any evidence of speech development. Treatment of the younger was associated with significant improvement in cognitive abilities and his speech development was initiated and improved. He was able to walk alone, and his fine motor skills showed marked improvement and was able to drink from cup and feed himself with as spoon. He was trying to copy a line and a circle. Treatment was not associated with any side effects.

Conclusion: There are very limited experiences with treatment of very rare disorders like Goldberg-Shprintzen syndrome. However, we have shown in this research paper that the use of evidence-based multi-factorial therapies was very beneficial in this condition.

Keywords: *Goldberg-Shprintzen Syndrome; Novel Therapies*

Introduction

Goldberg Shprintzen syndrome is a very rare autosomal recessive syndrome associated with mental retardation, a distinctive facial abnormalities, Hirschsprung disease, neurological abnormalities, and cerebral abnormalities on imaging studies. In 1981, Goldberg and

Shprintzen reported a brother and his sister with a mental-growth retardation syndrome associated with hypertelorism, submucous cleft palate, and aganglionic megacolon (Hirschsprung disease). In a previous publication, we described the occurrence of the thirty fourth and thirty fifth cases of the syndrome in two Iraqi brothers. There are no known therapies that are associated with improvement in the neurologic disability and mental retardation in Goldberg Shprintzen syndrome [1].

Aim of the Study

The aim of this paper is to describe the successful treatment of one of the Iraqi brothers with a new therapeutic approach.

Patients and Methods

The younger brother was treated with aim of improving neurological abnormalities which included spasticity and inability to walk without support, and also to improve mental disabilities which included poor cognitive abilities, and the lack of speech development. The younger brother was treated with an evidence-based therapeutic approach based on our extensive experiences with treatment of brain damage and mental retardation.

Treatment of the younger brother with started at about the age of five years. Before the initial treatment started, he had right hemiparesis and could not walk alone. The boy didn't have any speech development and had distinctive facial dysmorphism characterized by narrow palpebral fissures, hypertelorism open mouth, and laterally lifted ear (Figure 1). The boy also had submucous cleft palate.

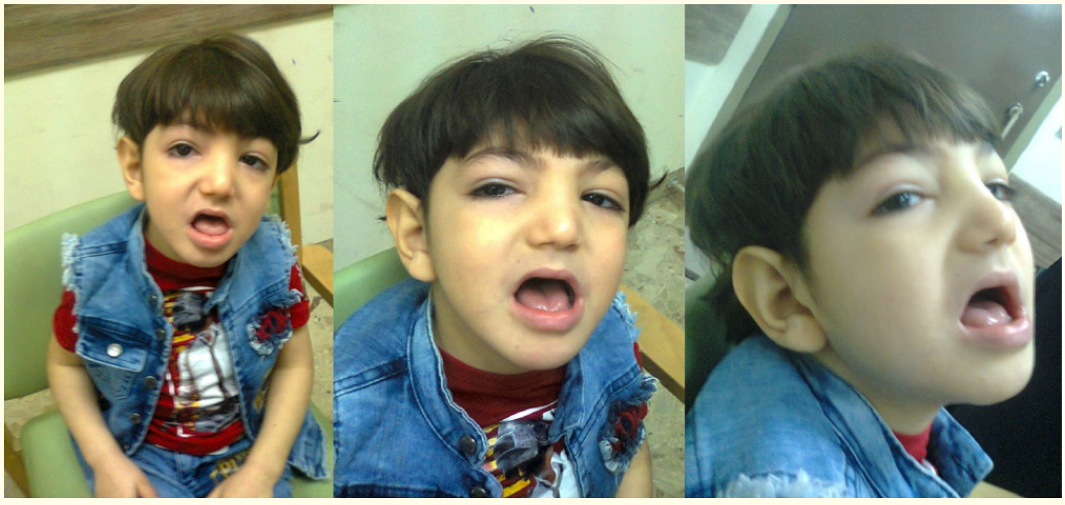

Figure 1: *The younger boy had distinctive facial features characterized by hypertelorism, open mouth, and laterally ear.*

Before treatment, the boy had right hemiparesis with spasticity and was not walking alone, but he could stand for a moment without support and was walking supporting himself to the wall and furniture (Figure 2). The had poor cognitive abilities and was not understanding nor responding to simple commands. The boy also had impairment in his fine motor skills that prevented him from feeding himself with a spoon and was unable to drink with a cup appropriately. MRI revealed some atrophic changes at the left parietal region of the brain.

Figure 2: *The boy was unable to walk alone, but he could stand momentarily unaided and walk holding the wall and furniture.*

The boy didn't have clear history of birth asphyxia nor CNS injury or infection during infancy that can be blamed for the right hemiplegia. However, he had Hirschsprung disease presented with intestinal obstruction during the neonatal period and required surgical resection and colostomy at about one month of age.

The parents were healthy and close relatives who also had a 17-year girl and a six-year boy that were both healthy.

The older Iraqi brother with Goldberg Shprintzen syndrome aged about 14 years and had significant mental and physical disability and had the same facial features of the younger brother (Figure 3).

Figure 3: *The two Iraqi brothers with Goldberg Shprintzen syndrome.*

Table 1 summarizes the evidence-based treatment courses received by the younger brother which was based on our extensive experiences with treatment of brain damage including brain atrophy and cerebral palsy, and mental retardation [2-11]. The older brother received only nutritional support and muscle relaxants.

First treatment course (One month)
Oral baclofen 10 mg twice daily
Oral citicoline 200 mg daily in the morning
Intramuscular nandrolone decanoate 25 mg once
Nutritional supplement mainly in the form of Royal jelly capsules once daily.
Second treatment course (One month)
Oral baclofen 10 mg twice daily
Oral citicoline 200 mg daily in the morning
Intramuscular cerebrolysin 5 ml every third day in the morning (10 doses).
Third treatment course (One month)
Oral baclofen 10 mg twice daily
Oral citicoline 300 mg daily in the morning
Intramuscular nandrolone decanoate 25 mg once
Nutritional supplement mainly in the form of Royal jelly capsules once daily.
Fourth treatment course (One month)
Oral baclofen 10 mg three times daily
Oral citicoline 300 mg daily in the morning
Intramuscular nandrolone decanoate 25 mg once
Nutritional supplement mainly in the form of Royal jelly capsules twice daily.
Fifth treatment course (One month)
Oral baclofen 10 mg three times daily
Oral citicoline 300 mg daily in the morning
Intramuscular piracetam 800 mg every third day in the morning (10 doses)
Amino acid supplementation.

Table 1: *The evidence-based treatment courses received by the younger brother.*

Results

After about six months, the older brother (Figure 4) who received only nutritional support and muscle relaxants showed slight improvement in spasticity and general health, but he didn't experience any improvement in motor and mental-cognitive disabilities, and he remained unable to stand and didn't show any evidence of speech development.

Figure 4: *After about six months, the older brother showed slight improvement in spasticity and general health, but he didn't experience any improvement in motor and mental-cognitive disabilities, and he remained unable to stand.*

The younger brother showed obvious improvement in cognitive abilities after the first course of treatment and was understanding simple commands like wave goodbye to the doctor (Figure 5). After the third course of treatment, he showed significant improvement in cognitive abilities and his speech development was initiated. He liked to talk with the doctor asking "How are you" and also liked shaking hand with the doctor. He also showed marked reduction in spasticity and improved mobility and ability to stand, but he was frightened when he was under pressure to walk without support and was still unable to walk alone (Figure 6). After the fifth course, he was able to walk alone (Figure 7), and his fine motor skills showed marked improvement and was able to drink from cup and feed himself with as spoon. He was trying to copy a line and a circle (Figure 8).

Figure 5: *After the first course of treatment, the younger brother showed obvious improvement in cognitive abilities and was understanding simple commands like wave goodbye to the doctor.*

Figure 6: *After the third course of treatment, he showed significant improvement in cognitive abilities and his speech development was initiated. He liked to talk with the doctor and shaking hand with the doctor. He also showed marked reduction in spasticity and improved mobility and ability to stand, but he was frightened when he was under pressure to walk without support and was still unable to walk alone.*

Figure 7: *After the fifth course, he was able to walk alone.*

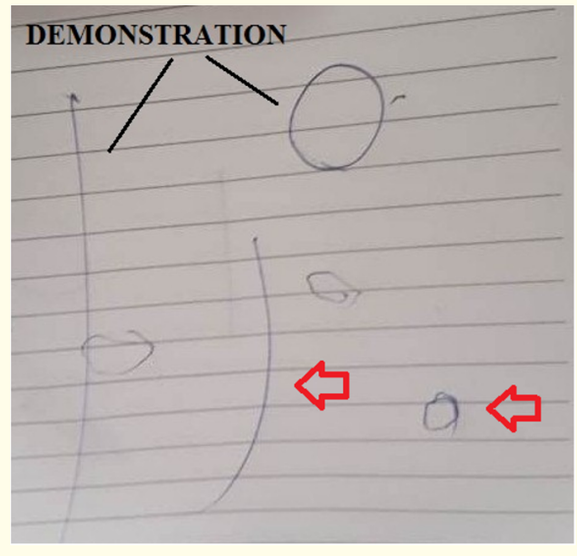

Figure 8: *After the fifth course, the boy's fine motor skills showed marked improvement and was trying to copy a line and a circle.*

Treatment was not associated with any side effects.

Discussion

The evidence-based multi-factorial therapies used in this boy with Goldberg-Shprintzen syndrome included citicoline, cerebrolysin and piracetam with aim of repairing brain damage and improving brain functions. Nandrolone was used with aim of strengthening muscles.

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 [4,5,12-18].

Cerebrolysin intramuscular solution contains free amino acids (85%) and 15% biologically active low molecular weight amino acids. It contains neuro-peptides (Brain-derived neurotrophic factor, glial cell line-derived neurotrophic factor, nerve growth factor, ciliary neurotrophic factor [19]. It has been used safely with benefit in a variety of neuro-psychiatric disorders including idiopathic mental retardation [7,10], cerebral palsy [2,6], myelomeningocele [20], pediatric juvenile spinal muscular atrophy [21,22], pediatric Charcot Marie Tooth disease [23,24], kernicterus [4,5], agenesis of corpus callosum with colpocephaly [25,26].

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 [27].

Nandrolone decanoate has been used with noticeable benefit in the treatment of cerebral palsy [9,28], refractory vitamin D-resistant rickets [29] and achondroplasia [30]. Nandrolone decanoate is not 17-testosterone derivatives, and therefore nandrolone esters are not associated with sodium sulfobromophthalein retention; therefore liver complications are infrequent when used for short periods. Nandrolones is known to have a useful muscle strengthening effects [9,28-30].

Conclusion

There are very limited experiences with treatment of very rare disorders like Goldberg-Shprintzen syndrome. However, we have shown in this research paper that the use of evidence-based multi-factorial therapies was very beneficial in this condition.

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Conflict of Interests

None.

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