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CASE REPORT

Goldberg Shprintzen Syndrome: The Novel Association with Congenital Unilateral Anorchia (Monarchism)

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ABSTRACT

BACKGROUND

Goldberg Shprintzen syndrome is a very rare autosomal recessive mental-growth retardation syndrome associated with characteristic facial dysmorphism, Hirschsprung disease, and a variety of neurological abnormalities, and abnormalities on brain imaging studies. However, the association of the syndrome with congenital unilateral absence of the testis (monarchism) has not been reported before. We have previously reported the thirty fourth and thirty fifth cases of the syndrome which occurred in Iraqi brothers and described a novel therapeutic approach which was used to treat the younger brother. The aim of this paper is to report the novel association of Goldberg Shprintzen syndrome with congenital right monarchism.

PATIENT AND METHODS

T.A.S, the younger of two brothers with Goldberg Shprintzen syndrome was first seen at the age of four years and 10 months at the paediatric neuro-psychiatric clinic on the 29th of August, 2019. He had spastic right hemiparesis and was unable to walk alone and was not saying any word and had characteristic facial features including hypertelorism, narrow palpebral fissures, open mouth, and laterally lifted ear. He also had neonatal intestinal obstruction which was attributed to Hirschsprung disease and was treated surgically with resection and colostomy. The boy was treated successfully with novel therapeutic approach and experienced improvement in cognitive abilities, speech, and motor function, and after treatment was able to walk alone.

RESULTS

During July 2021, the family reminded us that the child had single testis in the scrotum, and during early infancy an MRI study failed to find any second testis anywhere. An ultrasound was performed and showed normal left testis. However, the right testis could not find in the right hemi-scrotal sac nor with the right inguinal canal or

within the abdomen. Thus, the ultrasound confirmed the earlier MRI findings which suggested congenital absence of the right testis (monarchism).

CONCLUSION

This paper reported the novel association of Goldberg Shprintzen syndrome with monarchism, and this case represented the third case of congenital syndromic monarchism in the world.

KEYWORDS

Goldberg-shprintzen syndrome; Absent testis (monarchism)

INTRODUCTION

Goldberg Shprintzen syndrome is a very rare autosomal recessive mental-growth retardation syndrome associated with characteristic facial dysmorphism, Hirschsprung disease, and a variety of neurological abnormalities, and abnormalities on brain imaging studies. However, the association of the syndrome with congenital unilateral absence of the testis (monarchism) has not been reported before [1-3].

We have previously reported the thirty fourth and thirty fifth cases of the syndrome which occurred in Iraqi brothers and described a novel therapeutic approach which was used to treat the younger brother [2,3].

The aim of this paper is to report the novel association of Goldberg Shprintzen syndrome with congenital right monarchism.

PATIENT AND METHODS

T.A.S, the younger of two Iraqi brothers with Goldberg Shprintzen syndrome was first seen at the age of four years and 10 months at the paediatric neuro-psychiatric clinic on the 29th of August 2019. He had spastic right hemiparesis and was unable to walk alone, and was not saying any word, and had characteristic facial features including hypertelorism, narrow palpebral fissures, open mouth, and laterally lifted ear (Figure 1A). He also had submucous cleft palate. The boy also had impaired fine motor skills and was unable to neither feed self with spoon nor drink with a cup appropriately.

There was no clear history of asphyxia at birth nor CNS injury or infection during infancy that can be blamed for the spastic hemiplegia. However, the boy had neonatal intestinal obstruction which was attributed to Hirschsprung disease and was treated surgically with resection and colostomy at about thirty days of age.

Brain MRI showed some atrophic changes at the left parietal region. The boy was treated successfully with novel therapeutic approach and experienced improvement in cognitive abilities, speech, and motor function, and was able to walk alone (Figure 1B).

RESULTS

During July 2021, the family reminded us that the boy had single testis in the scrotum, and during early infancy an MRI study failed to find any second testis anywhere. An ultrasound was performed and showed normal left testis in the scrotum (Figure 2) with a dimensions of 11 mm × 7 mm. However, the right testis could not find in

the right hemi-scrotal sac nor with the right inguinal canal or within the abdomen. Thus, the ultrasound confirmed the earlier MRI findings which suggested congenital absence of the right testis (monarchism).

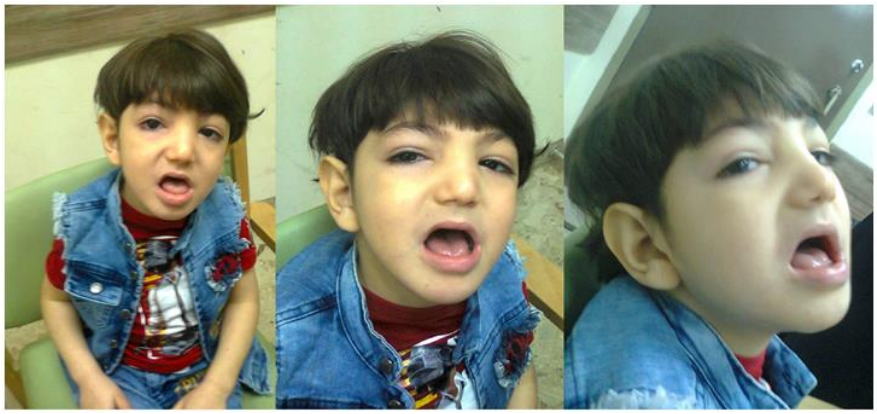

Figure 1A: The boy had characteristic facial features including by hypertelorism, open mouth, and laterally ear.

Figure 1B: The boy was treated successfully with novel therapeutic approach, and experienced improvement in cognitive abilities, speech, and motor function, and was able to walk alone (July 2021).

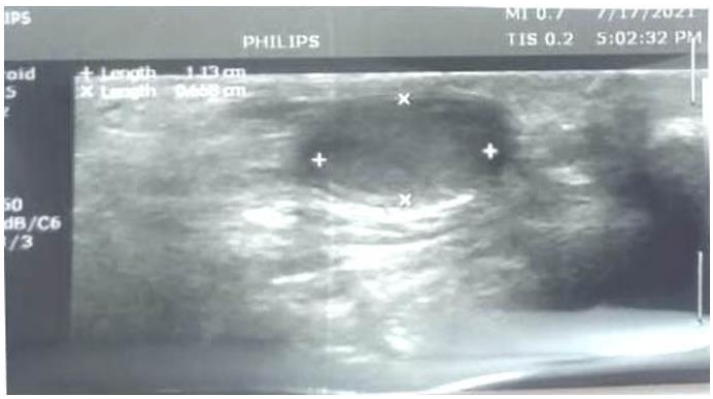

Figure 2: An ultrasound was performed and showed normal left testis in the scrotum.

DISCUSSION

In 1981, Goldberg and Shprintzen described siblings with mental and growth retardation, characteristic facial dysmorphism, short-segment Hirschsprung disease, and cleft palate [1]. We have previously reported the thirty fourth and thirty fifth cases of the syndrome which occurred in Iraqi brothers [2], and we described a novel therapeutic approach which was used to treat the younger brother [3]. The association of the syndrome with congenital absence of the testis (monarchism) has not been reported before [1-3].

Unilateral anorchia (monarchism), congenital absence of one testis is an extremely rare condition, and the syndromic form was reported only in two patients previously [4-14]. The condition was known as early as the late 1950s [4], and few cases were reported during the 1960s and 1970s [5-9] including eight cases reported by TIBBS (1961) [6].

In 1984, Hamidinia and colleagues reported one case of unilateral anorchia [10], while Schindler et al (1987) from Switzerland reported 512 boys who had an empty scrotum. 495 (96.7%) of them had cryptorchidism, 4 of them had ectopia and 13 patients unilateral anorchia [11].

Saito and Kumamoto (1989) from Japan reported seven cases of congenital monarchism who had normal number of spermatogonia per seminiferous tubule [12].

The first case of syndromic congenital monarchism was most probably reported by Cremades Mira et al. (1991) from Spain who reported a neonate with Prune Belly syndrome who had urethral obstruction, unilateral anorchia and hyaline membrane disease [13].

The second case of congenital syndromic monarchism was most probably reported by Chaudhury et al. (2010) from India who reported a neonate who had abdominal distension with no passage of meconium caused by ano-rectal malformation which was associated with agenesis of the left kidney and right-side anorchia. Laparotomy showed congenital pouch colon [14].

CONCLUSION

Unilateral anorchia (monarchism), congenital absence of one testis is an extremely rare condition, and the syndromic form was reported only in two patients. This paper reported the novel association of Goldberg Shprintzen syndrome with monarchism, and this case represented the third case of congenital syndromic monarchism in the world.

ACKNOWLEDGEMENT

The author would like to express his gratitude for the parents who accepted the publication of the photos of their child.

Figure 1A was included in author's previous publication, but the author has its copyright.

CONFLICTS OF INTEREST

None.

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