

Goldberg Shprintzen syndrome: The novel association with congenital unilateral anorchia (monorchism)

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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 (monorchism) has not been reported before. We have previously reported the thirty fourth and thirty fifth cases of the syndrome which occurred in Iraq, and described a novel therapeutic approach which was used to treat the younger boy. This paper reports the novel association of Goldberg Shprintzen syndrome with congenital right monorchism.

Patients and methods: T.A.S, the younger of two boys with Goldberg Shprintzen syndrome was first seen at the age of four years and 10 months at the pediatric 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 found 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 (monorchism).

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

Keywords: Goldberg-Shprintzen syndrome, absent testis (monorchism).

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 (monorchism) has not been reported before [1, 2, 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 monorchism.

Patients 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 pediatric 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 himself 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

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(Figure1B).

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 a second testis. An ultrasound was performed and showed normal left testis in the scrotum (Figure 2) with a dimension of 11 by 7 mm. However, the right testis could not found 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 (monorchism).

Discussion

In 1981, Goldberg and Shprintzen described siblings with mental and growth retardation, characteristic facial dysmorphism, short-segment Hirschsprung disease, and cleft palate



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



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

[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 (monorchism) has not been reported before [1, 2, 3].

Unilateral anorchia (monorchism), congenital absence of one testis is an extremely rare condition, and this syndrome was reported only in two patients [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 [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 monorchism who had normal number of spermatogonia per seminiferous tubule [12].

The first case of syndromic congenital monorchism 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].

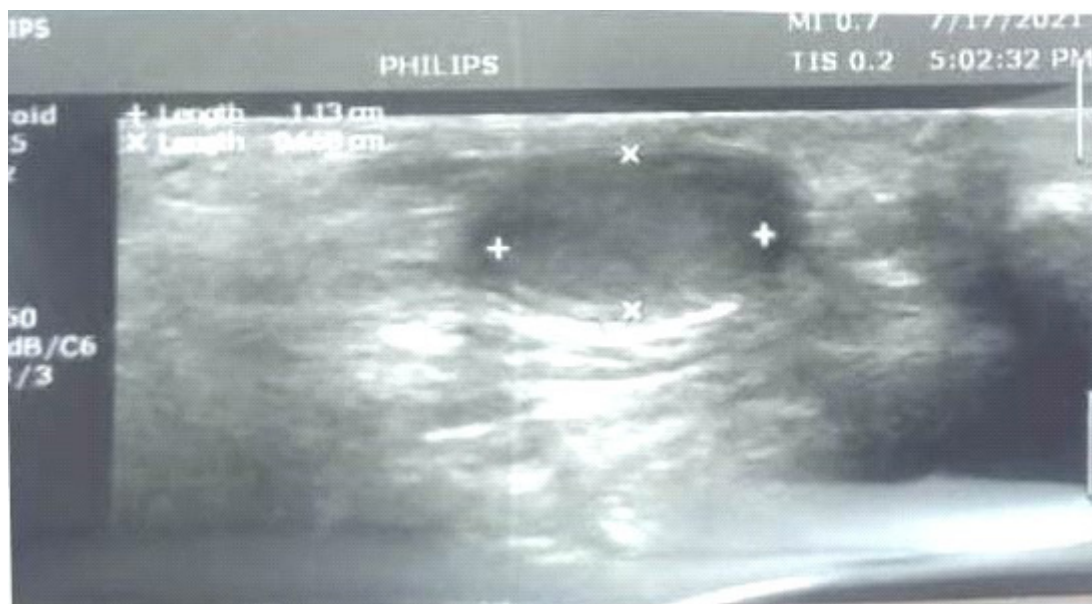


Figure 2: An ultrasound was performed and showed the presence of normal left testis in the scrotum.

The second case of congenital syndromic monorchism 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 a no-rectal malformation which was associated with agenesis of the left kidney and right-side anorchia. Laparotomy showed congenital pouch colon [14].

Conclusion

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

Conflict of interest: None.

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