Research Article

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The Association of Esquirol-Séguin-Down Syndrome with Bilateral Cryptorchidism: Educational Report and Ultrasound Image

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Abstract

Background: *Esquirol-Séguin-Down syndrome (Trisomy 21) was first described by Jean-Etienne Dominique Esquirol in 1838 and later by Edouard Séguin in 1846. Thereafter, in 1862, John Langdon Down, a British physician emphasized that the syndrome is a distinct form of mental retardation.*

Patients and methods: *A four-year boy old with Esquirol-Séguin-Down syndrome, developmental delay, and bilateral undescended testes is presented, and the recent relevant literatures were reviewed.*

Results: *Dysmorphic facial features included low set ears, depressed nasal bridge and oblique eye fissures. Ultrasound showed that both testicles were of normal size, but they were located in the pelvis.*

Conclusion: *In this paper, the association of Esquirol-Séguin-Down syndrome with bilateral undescended testes is highlighted, and an education ultrasound is presented.*

Keywords: Esquirol-Séguin-Down syndrome, bilateral cryptorchidism.

Introduction

Esquirol-Séguin-Down syndrome (Trisomy 21) was first described by Jean-Etienne Dominique Esquirol (Figure-1A) in 1838 and later by Edouard Séguin (Figure-1B) in 1846. Thereafter, in 1862, John Langdon Down (Figure-1C), a British physician, emphasized that the syndrome is a distinct form of mental retardation. The syndrome was recognized as a chromosome 21 trisomy by Dr Jérôme Lejeune (Figure-1D) in 1959, and the condition became known as trisomy 21[1, 2, 3].

Figure 1A: Jean-Etienne Dominique Esquirol (3 February 1772 -12 December, 1840), a French psychiatrist

Figure 1B: Edouard Séguin (January 20, 1812-October 28, 1880), a physician and educationist born in Clamecy, Nièvre, France. He was best known for his work with children with cognitive impairments in France and the United States

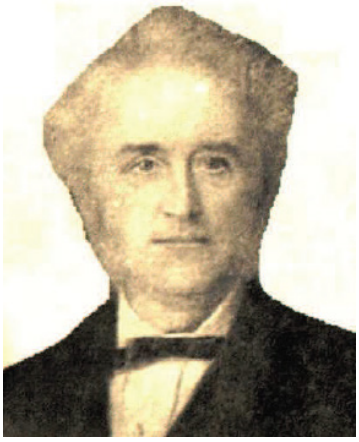

Figure 1C: John Langdon Down (18 November, 1828-7 October, 1896), a British physician

Figure 1D: Jérôme Jean Louis Marie Lejeune (13 June, 1926-3 April, 1994) was a French pediatrician and geneticist, best known for discovering the link of diseases to chromosome abnormalities and for his subsequent opposition to prenatal diagnosis and abortion

Patients and methods

A four-year boy old with Esquirol-Séguin-Down syndrome, developmental delay, and bilateral undescended testes is presented, and the recent relevant literatures were reviewed.

Results

Dysmorphic facial features included low set ears, depressed nasal bridge and oblique eye fissures (Fifgure-2A). The boy had poor speech development, and when the boy was convinced to take a pen to draw something: He couldn't copy a line or a circle (Figure-2B) suggesting a mental age under the three years. Ultrasound showed that both testicles were of normal size, but they were located in the pelvis (Figure-3).

Figure 2A: A four-year boy old with Esquirol-Séguin-Down syndrome and undescended testes: Depressed nasal bridge and oblique eye fissures

Figure 2B: When the boy was convinced to take a pen to draw something: He couldn't copy a line or a circle

Figure 3: Ultrasound of a four-year old boy with Esquirol-Séguin-Down syndrome showed that both testicles were of normal size, but they were located in the pelvis

Discussion

Salemi et al (2012) emphasized that cryptorchidism is the most common congenital abnormality of the urogenital in males, and patients with Esquirol-Séguin-Down syndrome have a higher risk of cryptorchidism [4].

Satgé et al (1997) and Papatsoris et al (2003) emphasized that the association of Esquirol-Séguin-Down syndrome and undescended testes [5, 6].

Ebert et al (2008) reported the urological abnormalities in 24 patients with Esquirol-Séguin-Down syndrome (23 males and female) with a mean age of 79.4 months. Thirteen male patients had uni- or bilateral cryptorchidism. Two patients had posterior urethral valves, two patients had bladder exstrophy, and two patients had hypospadias. Three patients had neurogenic and eight non-neurogenic functional bladder dysfunction [7].

Miki et al (1999) reported the association of typical testicular seminoma with bilateral undescended testes in Esquirol-Séguin-Down syndrome [8].

Conclusion

In this paper, the association of Esquirol-Séguin-Down syndrome with bilateral undescended testes is highlighted, and an education ultrasound is presented.

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Some of the figures in this paper were included in author's previous publication, but the author has their copyright.

Conflict of interest: None.

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