Esquirol-Séguin-Down Syndrome Associated with Hepatic Hemangioma: An Association not Previously Reported in the Literature

Aamir Jalal Al-Mosawi

Advisor in Pediatrics and Pediatric Psychiatry, The National Training and Development Center and Baghdad Medical City

Corresponding Author: Aamir Jalal Al-Mosawi, e-mail: almosawiAJ@yahoo.com

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Abstract

Background: Hemangiomas including liver hemangiomas are benign vascular tumors that are generally observed during infancy, and can be congenital. Although Sepúlveda and colleagues emphasized that hepatic hemangioma are the most common tumor of the liver during childhood, the association with Esquirol-Séguin-Down syndrome has not been reported in the literature.

Patients and methods: A female infant with Esquirol-Séguin-Down syndrome and abnormal abdominal sound was studied.

Results: Dysmorphic features included oblique eye fissures, depressed nasal bridge, low set ears, small mouth, and protruded tongue. Ultrasound examination showed hepatic hemangioma.

Conclusion: In this paper, the first case of Esquirol-Séguin-Down syndrome associated with hepatic hemangiomas is reported.

Keywords: Esquirol-Séguin-Down syndrome, hepatic hemangiomas

Introduction

Hemangiomas including liver hemangiomas are benign vascular tumors that are generally observed during infancy, and can be congenital. Although Sepúlveda and colleagues emphasized that hepatic hemangioma are the most common tumor of the liver during childhood, the association with Esquirol-Séguin-Down syndrome has not been reported in the literature [1].

Patients and methods

A female infant with Esquirol-Séguin-Down syndrome and abnormal abdominal sound was studied.

Results

Dysmorphic features included oblique eye fissures,
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Depressed nasal bridge, low set ears, small mouth, and protruded tongue (Figure 1). Ultrasound examination showed hepatic hemangioma (Figure 2).

Discussion

Esquirol-Séguin-Down syndrome (Trisomy 21) was first described by Jean-Etienne Dominique Esquirol (Figure 3A) in 1838 and later by Edouard Séguin (Figure 3B) in 1846. Thereafter, in 1862, John Langdon Down (Figure 3C), 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 3D) in 1959, and the condition became known as trisomy 21 [2-4].

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

Figure 3B. 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 3C. John Langdon Down (18 November, 1828-7 October, 1896), a British physician

Figure 3D. 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
Although Sepúlveda and colleagues emphasized that hepatic hemangioma are the most common tumor of the liver during childhood, the association with Esquirol-Séguin-Down syndrome has not been reported in the literature [1].

Gourgiotis, et al. (2006) emphasized that hepatic hemangiomas which are vascular malformations that are observed at birth or during infancy, are the most common benign tumors of the liver and are frequently asymptomatic. Gourgiotis et al also emphasized that the management of hepatic hemangiomas is generally conservative, and surgery is indicated in cases complicated by spontaneous or traumatic rupture, intra-tumoral bleeding, consumption coagulopathy, and rapid growth [5].

### Conclusion

In this paper, the first case of Esquirol-Séguin-Down syndrome associated with hepatic hemangiomas is reported.

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### References


