
CASE REPORT

Esquirol-seguin-down syndrome associated with hepatic hemangioma: An association not previously reported in the literature

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ABSTRACT

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

PATIENTS AND METHODS: A female infant with Esquirol-Seguín-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.

DISCUSSION: 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. The syndrome was recognized as a chromosome 21 trisomy by Dr Jérôme Lejeune in 1959, and the condition became known as trisomy.

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

Key Words: *Esquirol-Seguín-Down syndrome; Hepatic hemangiomas; Iraq*

INTRODUCTION

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

PATIENTS AND METHODS

A female infant with Esquirol-Seguín-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 (Figure 1). Ultrasound examination showed hepatic hemangioma (Figure 2).



Figure 1) A female infant with Esquirol-Séguin-Down syndrome: Oblique eye fissures, depressed nasal bridge, low set ears, small mouth, and protruded tongue

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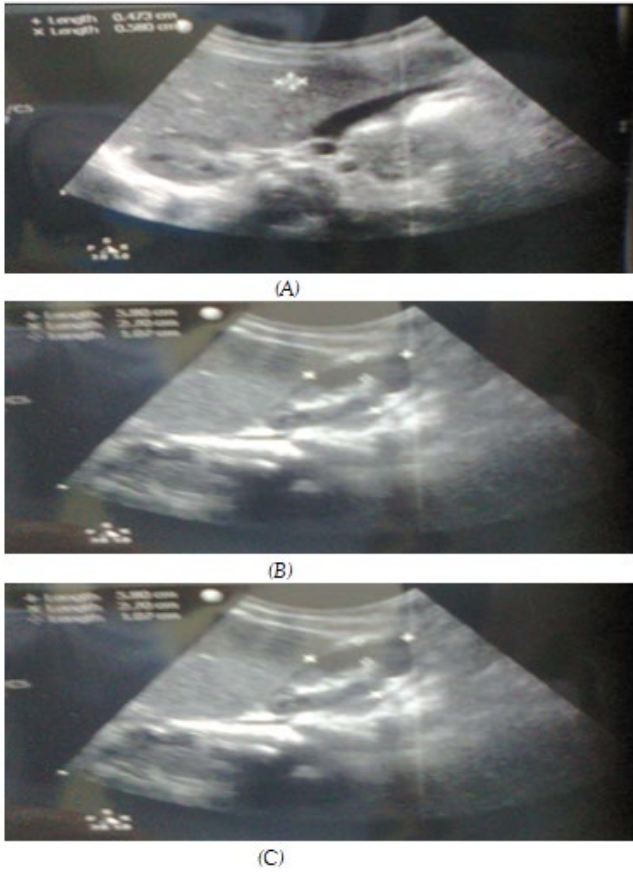


Figure 2) A) An ultrasound imaging of a female child with Esquirol-Séguin-Down syndrome suggestive of hepatic hemangioma B) An ultrasound imaging of a female child with Esquirol-Séguin-Down syndrome suggestive of hepatic hemangioma C) An ultrasound imaging of a female child with Esquirol-Séguin-Down syndrome suggestive of hepatic hemangioma

DISCUSSION

Esquirol-Séguin-Down syndrome (Trisomy 21) was first described by Jean-Etienne Dominique Esquirol (Figure 3) in 1838 and later by Edouard Séguin (Figure 4) in 1846. Thereafter, in 1862, John Langdon Down (Figure 3), 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 5) in 1959, and the condition became known as trisomy 21 [2-4].

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 [5].



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



Figure 4) 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 5) John Langdon Down (18 November, 1828-7 October, 1896), a British physician



Figure 6) 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

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] (Figure 6).

CONCLUSION

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

DECLARATIONS

The author would like to express his gratitude for the parents of the child who willingly accepted publishing his photos. Some of the figures in this paper were included in author's previous publication, but the author has their copyright.

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