ISSN 2582 – 6050

Available Online at www.ijpba.info
International Journal of Pharmaceutical & Biological Archives 2026; 17(1):1-3

CASE STUDY

Recognizing and Avoiding the Misdiagnosis of Esquirol-Séguin-Down Syndrome in Clinical Practice: A Clinical Imperative

Aamir Jalal Al-Mosawi

Baghdad Medical City and the National Training and Development Center, Baghdad, Iraq

Received: 15-01-2026; Revised: 13-02-2026; Accepted: 21-03-2026

ABSTRACT

Esquirol-Séguin-Down syndrome (Trisomy 21) is the most common chromosomal disorder with well-defined clinical features, but misdiagnosis is still common in clinical practice. This leads to unnecessary distress, delayed interventions, and inappropriate management. This article explores common reasons for misdiagnosis, using a clinical case example, and discusses strategies for improving diagnostic accuracy. A focused educational approach can enhance awareness and prevent misdiagnosis, ultimately improving patient outcomes.

Keywords: Misdiagnosis, Clinical features, Developmental delay

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.

John Langdon Down [Figure 1c] further defined the syndrome in 1862 as a distinct form of intellectual disability.

In 1959, Jérôme Lejeune [Figure 1d] reported that the syndrome is caused by a trisomy of chromosome 21.

Esquirol-Séguin-Down syndrome is the most common chromosomal disorder, with well-defined clinical features and a straightforward diagnostic approach, and the diagnosis is often clinical. However, misdiagnosis still occurs in clinical practice, leading to unnecessary distress, delayed interventions, and inappropriate management. This article presents an example and highlights the key reasons for misdiagnosis and outlines best practices for accurate identification.

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

The condition presents with hypotonia during infancy, developmental delay, and characteristic dysmorphic features, including upslanting palpebral fissures, flat nasal bridge, and flat occiput).

Sandal gap (Separation of the big toe has) may occur in 45% of patients with Esquirol-Séguin-Down syndrome.[1-7]

PATIENTS AND METHODS

A 6-month-old girl was referred for evaluation after being diagnosed with brain atrophy by multiple physicians. On examination, the patient presented with classic features of Esquirol-Séguin-Down syndrome [Figure 2]. The child was alert, smiling, and engaged, demonstrating good eye contact and a willingness to interact. There was no history of birth asphyxia, significant neonatal hyperbilirubinemia, or serious neonatal infections. Family history was unremarkable for any similar condition. Brain ultrasound findings were normal. Based on the clinical features, a diagnosis of Esquirol-Séguin-Down syndrome was confidently made.

Based on the clinical features including hypotonia, flat occiput, facial characteristics (round face, epicanthic folds, flat nasal bridge), and the sandal gap deformity, a clinical diagnosis of Esquirol-Séguin-Down syndrome was confidently made.

Figure 1: (a) Jean-Etienne Dominique Esquirol (February 3, 1772–December 12, 1840), a French psychiatrist. (b) Edouard Séguin (January 20, 1812–October 28, 1880), a physician born in Clamecy, Nièvre, France. He was best known for his work with children with cognitive impairments in France and the United States. (c) John Langdon Down (18 November, 1828–7 October, 1896), a British physician. (d) Jérôme Jean Louis Marie Lejeune (13 June 13, 1926–April 3, 1994), a French pediatrician and geneticist

DISCUSSION

Esquirol-Séguin-Down syndrome, while the most common chromosomal disorder, is often misdiagnosed due to its wide clinical variability and the subtlety of its presentation in some cases. As noted by A. Maurel Ollivier in 1990, while most physicians are familiar with Esquirol-Séguin-Down syndrome, many lack sufficient knowledge about the associated disabilities, leading to potential misdiagnoses.[7] Common mistaken diagnoses include brain atrophy, other neurological conditions, and even non-specific developmental delays. The absence or subtle expression of classic dysmorphic features, such as epicanthic folds or a flat nasal bridge, may lead clinicians to overlook the possibility of a chromosomal disorder.[1-7]

Further, physicians' limited exposure to genetic and dysmorphic conditions and a lack of awareness regarding the full range of presentations contribute to diagnostic uncertainty. It is crucial to recognize that the presence of features like hypotonia, flat occiput, round face, epicanthic folds, and the sandal gap sign should immediately raise suspicion for Esquirol-Séguin-Down syndrome, even if other symptoms are less obvious. Educating healthcare providers about these characteristic signs, possibly

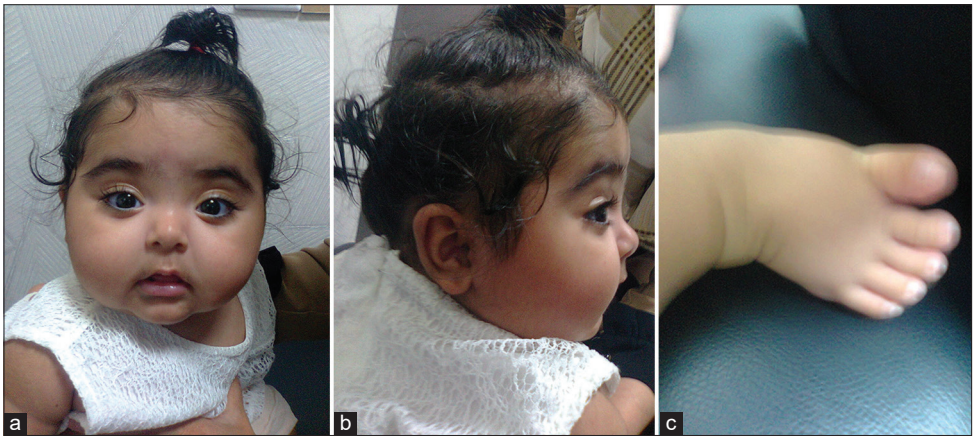

Figure 2: (a) An infant with round facial features and a mildly flattened nasal bridge. (b) Side facial profile. (c) Sandal gap sign

through atlases and hands-on training, is key to improving diagnostic accuracy.[1-7]

CONCLUSION

Esquirol-Séguin-Down syndrome misdiagnosis remains a significant yet preventable issue in neonatal and pediatric practice. A cautious approach balancing clinical expertise with confirmatory genetic testing, ensures accurate diagnosis and appropriate early intervention. Increased awareness among healthcare providers is crucial to minimizing diagnostic errors and improving patient outcomes.

ACKNOWLEDGMENTS

The parents of the patient kindly consented to and encouraged the publication of their child's photo to contribute to advancing the understanding of the condition.

Some of the sketches (Figures) were included in the previous author's publications, but the author has their copyright.

CONFLICTS OF INTEREST

None.

REFERENCES

1. Al-Mosawi AJ. Down Syndrome Atlas. 1st ed. Saarbrücken: LAP Lambert Academic Publishing: 2018.
2. Al-Mosawi AJ. Atlas of esquirol-séguin-Down Syndrome in Iraq. Saarbrücken: LAP LAMBERT Academic Publishing: 2021.
3. Al-Mosawi AJ. Chronic cutaneous disorders in down syndrome patients. J Health Care Res 2020;1:65-71.
4. Al-Mosawi AJ. Esquirol-séguin-down syndrome associated with atrial septal defect plus tricuspid regurgitation. Cardiol Res Rep 2022;4:1-5.
5. Al-Mosawi AJ. The association of Esquirol-Séguin-down syndrome with bilateral cryptorchidism: Educational report and ultrasound image. Int J Endocrinol Res Rev 2022;2:1-3.
6. Al-Mosawi AJ. Esquirol-séguin-down syndrome associated with hepatic hemangioma: An association not previously reported in the literature. Case Rep Res Pract Med 2022;2:34-6.
7. Al-Mosawi AJ. Esquirol-séguin-down syndrome associated with inner ear impairment: An education article. Online J Neurol Brain Disord 2023;7:635-8.