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Research Article

Esquirol-Séguin-Down Syndrome Associated with Inner Ear Impairment: An Education Article

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Abstract

Background: As early as 1990, A Maurel Ollivier emphasized that all doctors are familiar with Esquirol-Séguin-Down syndrome, but most of them are poorly educated about the associated disability. Ollivier emphasized the importance of early detection of hearing impairment because of its potential for worsening the manifestations of mental retardation. However, little emphasis has been made in the medical literature about the association of Esquirol-Séguin-Down syndrome with inner ear impairment.

Patients and methods: The case of Esquirol-Séguin-Down syndrome associated with inner ear impairment in Iraqi boy is described.

Results: The boy hearing was assessed using tympanogram, distortion product otoacoustic emissions (DPOAEs) which assesses outer hair cell and cochlear functions, transient evoked otoacoustic emission (TEOAE), auditory brainstem responses to a chirp stimulus (CHIRP signal ABR), and auditory steady state responses (ASSR).The tympanogram showed normal middle ear function, while the other tests showed unilateral right ear abnormally which included abnormal inner ear and defect of the outer hair cell on distortion product otoacoustic emissions (DPOAEs), abnormal inner ear and defect of the outer hair cell on transient evoked otoacoustic emission (TEOAE), Wave V detected at 35dB in CHIRP signal ABR, and mild hearing loss on auditory steady state responses (ASSR).

Conclusion: Little emphasis has been made in the medical literature about the association of Esquirol-Séguin-Down syndrome with inner ear impairment. In this paper, the association of Esquirol-Séguin-Down syndrome with inner ear impairment is reported.

Keywords: Esquirol-Séguin-Down syndrome; Inner Ear Impairment; Iraq

Introduction

Esquirol-Séguin-Down syndrome (Trisomy 21) was described for the first time by Jean-Etienne Dominique Esquirol (Figure 1A) in 1838 and was later described by Edouard Séguin (Figure 1B) in 1846. John Langdon Down (Figure 1C), a British physician, emphasized that the syndrome is a distinct form of mental retardation in 1862. In 1959 Jérôme Lejeune (Figure 1D) reported that the syndrome is caused by a trisomy of chromosome 21 [1-5]. As early as 1990, A Maurel Ollivier emphasized that all doctors are familiar with Esquirol-Séguin-Down syndrome, but most of them are poorly educated about the associated disability. Ollivier emphasized the importance of early detection of hearing impairment because of its potential for worsening the manifestations of mental retardation [6]. However, little emphasis has been made in the medical literature about the association of Esquirol-Séguin-Down syndrome with inner ear impairment.

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

Figure 1B: 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.

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

Figure 1D: Jérôme Jean Louis Marie Lejeune (13 June 13, 1926- April 3, 1994), a French pediatrician and geneticist.

Patients and Methods

The case of Esquirol-Séguin-Down syndrome associated with inner ear impairment is described.

Results

During early childhood the boy was hypotonic and had very slow development of motor skills. After the age of two years, he received courses of cerebrolysin, piracetam, citicoline, and other therapies to make him able to stand and walk. He received these medications based on the evidence presented in several publications [7-10]. At the age of seven years, the boy had evidence of significant mental retardation as he had poor fine motor skills and was unable to use a pen to scribble. He was able to understand only few simple commands and was not saying any word. The boy had dysmorphic features consistent with Esquirol-Séguin-Down syndrome, including oblique eye fissures, depressed nasal bridge, low set ears, small mouth, and protruded tongue (Figure 2). The boy hearing was assessed using tympanogram, distortion product otoacoustic emissions (DPOAEs) which assesses outer hair cell and cochlear functions, transient evoked otoacoustic emission (TEOAE), auditory brainstem responses to a chirp stimulus (CHIRP signal ABR), and auditory steady state responses (ASSR). The tympanogram showed normal middle ear function, while the other tests showed unilateral right ear abnormally which included abnormal inner ear and defect of the outer hair cell on distortion product otoacoustic emissions (DPOAEs), abnormal inner ear and defect of the outer hair cell on transient evoked otoacoustic emission (TEOAE), Wave V detected at 35dB in CHIRP signal ABR, and mild hearing loss on auditory steady state responses (ASSR).

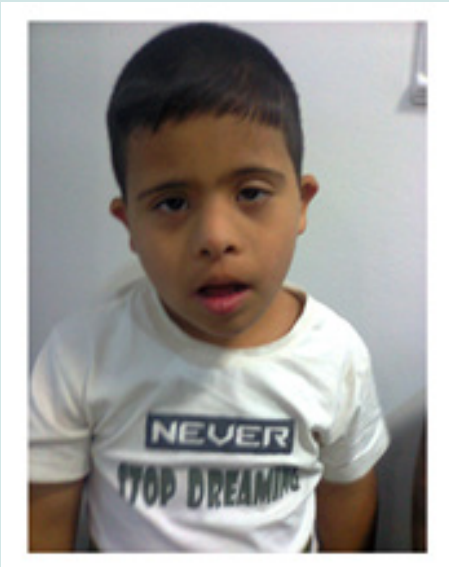

Figure 2A: A boy with Esquirol-Séguin-Down syndrome: Oblique eye fissures, depressed nasal bridge, low set ears, small mouth, and protruded tongue.

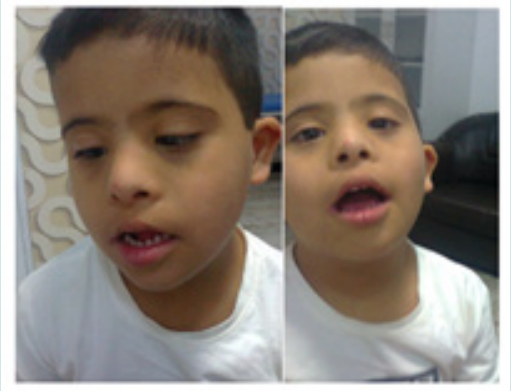

Figure 2B: A boy with Esquirol-Séguin-Down syndrome: Oblique eye fissures, depressed nasal bridge, low set ears, small mouth, and protruded tongue.

Discussion

As early as 1998, Hassmann et al emphasized that DPOAE distortion product otoacoustic emissions examination performed in patients with Esquirol-Séguin-Down syndrome with non-conductive hearing impairment reveal inner ear impairment. They performed audiological assessment on 47 children and 14 adult patients with Esquirol-Séguin-Down syndrome. Assessment included tympanometry, auditory brain response (ABR) and distortion products otoacoustic emissions (DPOAE). The study showed that patients with Esquirol-Séguin-Down syndrome frequently had conductive hearing impairment with tympanometry of B and C type detected in 56% of ears examined. The amplitude of DPOAE was lower in children with Esquirol-Séguin-Down syndrome [11].

Conclusion

Little emphasis has been made in the medical literature about the association of Esquirol-Séguin-Down syndrome with inner ear impairment. In this paper, the association of Esquirol-Séguin-Down syndrome with inner ear impairment is reported.

Acknowledgement

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Conflict of Interest: None.

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