



Waardenburg Syndrome Type I: An Otorhinolaryngologist's Perspective

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Abstract

Keywords: Waardenburg Syndrome Type I; Severe hearing loss; Bilateral; Deafness; Deafness

Introduction

A 11-year-old female patient presented with bilateral congenitally deafness. She had a family history of deafness. On physical examination, she had a white forelock and heterochromia. Audiogram showed bilateral profound hearing loss. Genetic testing confirmed Waardenburg Syndrome Type I. She underwent cochlear implantation and is now using hearing aids. This case highlights the importance of genetic testing in the management of congenitally deaf children.

Case Report

An 11-year-old female child with bilateral congenitally deafness was referred to the Department of Otorhinolaryngology, Gandhi Medical College, Hyderabad. She had a family history of deafness. On physical examination, she had a white forelock and heterochromia. Audiogram showed bilateral profound hearing loss. Genetic testing confirmed Waardenburg Syndrome Type I. She underwent cochlear implantation and is now using hearing aids. This case highlights the importance of genetic testing in the management of congenitally deaf children.

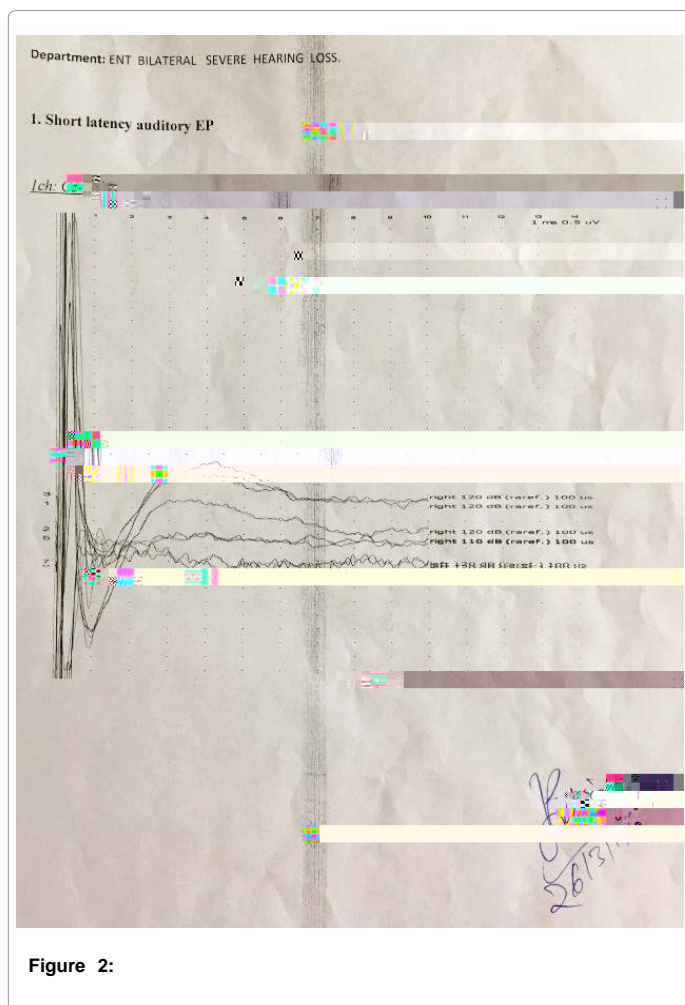


Figure 2:

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Figure 1:

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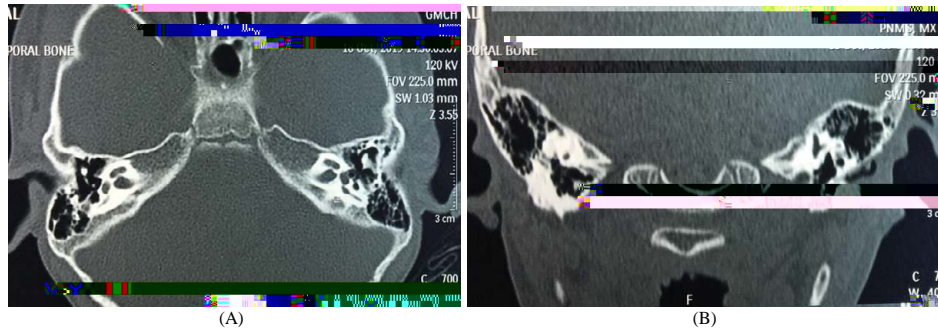


Figure 3:

- Affected first degree relative

Citation:

References

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