

Syndromic Deafness-Variant of Waardenburg Syndrome

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Background

Deafness in children is alarming and a cause of worry to near and dear ones. EBM documents that serious hearing impairment is found in one in 800 newborns. Amongst the 50 percent of permanent childhood deafness, 30 percent is syndromic and is thought to be because of abnormal genetic makeup. Syndromic cases of deafness are more accurately diagnosed by the associated additional features of the syndrome. Waardenburg syndrome is a rare, autosomally inherited disorder with distinct clinical manifestations of dystopia canthorum, white forelock, congenital hearing loss and heterochromia iridis.

Aims & Objectives

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syndrome.

Materials & Methods

A study of deaf mutism in 2 siblings, who presented with Waardenburg syndrome.

Results & Conclusions

Herewith, we are reporting 2 siblings who presented with deaf mutism and with clinically significant notable variations suggestive of a rare presentation of type 1 and type 2 Waardenburg syndrome in the same family.

Keywords: Waardenburg syndrome; Deaf mutism; Syndromic deafness.