Original Research Paper

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Otolaryngology

A CASE REPORT OF WAARDENBERG SYNDROME

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KEYWORDS :

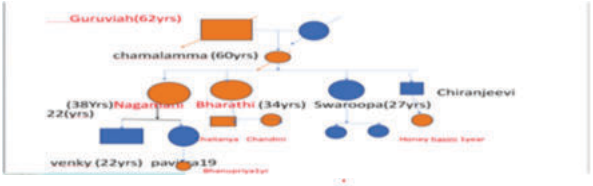
INTRODUCTION:

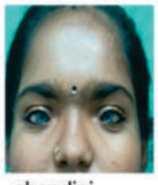
Waardenburg syndrome (ws) is a group of genetic conditions inherited in an autosomal dominant fashion. During embryogenesis, there is an abnormal distribution of melanocytes, which results in patchy areas of depigmentation. It is a rare disease, caused by loss of pigmentary cells in eyes, skin, stria vascularis of the cochlea, and hair. Based on genetic and clinical criteria, four types of WS are identified.

CASE REPORT:

Descriptive observational study A Case report: A 19-year-old female Chandini presented to OPD with hearing defect in both ears (deaf) and mute, whitish forelock, bluish discolouration of eyes, lateral displacement of inner canthi, and a first-degree relative with Ws

• At our ENT department, an ear examination performed by an otoscope showed normal findings. A tuning fork test and a pure-tone audiogram (PTA) revealed bilateral sensorineural hearing loss (SNHL).
• On PTA she has 100dbhl in both ears. For clinical diagnosis of Type 1 WS, two major or one minor and two minor criteria are needed.





chandini

mother,brother and niece of chandini

TYPES

Type 1 is due to mutations in heterozygous PAX3 gene And is characterized by
1) Broad and high nasal root
2) Hypertrichosis of the medial part of the eyebrows
3) Partial or total heterochromia iridis
4) White forelock
5) Congenital deaf mutism.
6) Lateral displacement of the medial canthi and lacrimal punctae.

Type 2 Waardenburg syndrome is due to MIFT gene mutations(AD)
• Patients have normally located canthi, profound sensorineural deafness, and different coloured irises.

Type 3 WS (also called Klein-Waardenburg syndrome) Due to homozygous PAX3 gene mutations(AR)

• Patients have the same features as type-1, but musculoskeletal abnormalities are more prominent .

Type 4 WS (also called Shah-Waardenburg syndrome) Due to multiple genes SOX10, EDNRB, EDRB(AR) Patients have the same features as type 1 WS, but it is associated with congenital megacolon (Hirschsprung disease).

Management

There is no definitive treatment for waardenburg syndrome, but supportive treatment with cochlear implants and surgery in case of association with hirschsprung syndrome can be done.

- genetic counselling
- Prenatal testing to determine gene pathology
- Interprofessional approach

CONCLUSION

Life expectancy is normal in children with Waardenburg syndrome. Genetic counselling is necessary because one affected gene can pass the syndrome to the next generation. The family and the patient's awareness regarding symptomatic treatment is also very important. Morbidity is related only to neural crest defects

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