ORIGINAL RESEARCH PAPER

Paediatrics

WAARDENBURG SYNDROME

KEY WORDS:

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INTRODUCTION

Waardenburg syndrome is a group of rare inherited disorder of neural crest cell development. It usually shows an autosomal dominant mode of inheritance, but autosomal recessive inheritance patterns are observed in some subtypes The prevalence varies from 1:42000 to 1:50,000. WS accounts for between 2% and 5% of cases of congenital deafness. It is characterised by at least some degree of congenital hearing loss and pigmentation deficiencies, which can include bright blue eyes (or one blue eye and one brown eye), a white forelock or patches of light skin.

The syndrome is caused by mutations in any of several genes that affect the division and migration of neural crest cells during embryonic development (though some of the genes involved also affect the neural tube). Neural crest cells are stem cells left over after the closing of the neural tube that go on to form diverse non-CNS cells in different parts of the body, including melanocytes, various bones and cartilage of the face and inner ear and the peripheral nerves of the intestines.

Clinical Profile

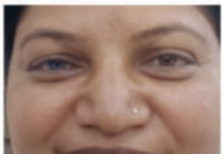
A 9-yr old boy came with complain of difficulty in speaking and hearing.

On Examination: Patient had heterochromia iridium with hearing difficulty intact Iq. And isolated speech delay with no white forelock of hair or any limb abnormalities or any cardiac anomaly facial anomaly or any neural tube closure defect or any bony deformity which is common in some types of Waardenburg Syndrome.

Patient had PTA report done suggestive of Right sided complete hearing loss with Left sided partial hearing loss.

Genetic testing was done which confirmed the diagnosis of Waardenburg Syndrome.

Patient had positive family history as patient's mother and maternal grandfather had similar complain with varying degrees of hearing loss.



RESULTS

VARIANT OF UNCERTAIN SIGNIFICANCE RELATED TO THE GIVEN PHENOTYPE WAS DETECTED

SNV(s)/INDELS

Gene* (Transcript)	Location	Variant	Zygosity	Disease (OMIM)	Inheritance	Classification§
SOX10 (-) (ENST00000396884.8)	Exon 3	c.449A>C (p.Lys150Thr)	Heterozygous	Waardenburg syndrome, type 2E with or without neurologic involvement (OMIM#611584); Waardenburg syndrome type 4C (OMIM#613266)	Autosomal dominant	Uncertain Significance (PM2,PP3)

DISCUSSION

Waardenburg syndrome is a group of rare inherited disorder of neural crest cell development. It usually shows an autosomal dominant mode of inheritance, but autosomal recessive inheritance patterns are observed in some subtypes.

Subtypes

1. Type 1 : Clinical features in this type include Telecanthus, Hypochromic Iridis, Partial albinism as hypopigmented ocular fundus and white eyelashes, eyebrows and forelock with hypopigmented skin lesions. Patient has associated hearing loss(53%), aplasia of posterior semicircular canal.

2. Type II. Similar to type I, except hearing loss is more common (92%), lateral displacement of the inner canthi is not present, and all other features occur less frequently than in type I.

3. Type III. Features of type I with the addition of upper limb defects, including hypoplasia of muscles, flexion contractures, carpal bone fusion, and syndactyly. Camptodactyly occurs occasionally. Hearing loss occurs in 57%.

4. Type IV. Features of type II with the addition of Hirschsprung disease. Hearing loss occurs in 84%

Hearing loss is most often associated with mutations in SOX10 (97%), MITF (90%), and SNAI2 (100%).

CONCLUSION

This case report illustrates the clinical presentation of Waardenburg syndrome Type II and emphasizes the importance of a thorough diagnostic workup, including genetic testing. Early diagnosis and comprehensive management can significantly improve the quality of life for individuals with Waardenburg syndrome.

- **Early Diagnosis** Early diagnosis of Waardenburg syndrome is crucial for timely intervention and optimal management of the condition.
- **Hearing Aid** Hearing aids are essential for improving hearing, facilitating speech development, and minimizing the impact of hearing loss.
- **Speech Therapy** Speech therapy is critical for supporting speech development in individuals with Waardenburg syndrome. It focuses on improving articulation, pronunciation, and language skills. Speech therapists work closely with the child and family to develop personalized treatment plans and provide ongoing support
- **Social Support** Social support from family, friends, and support groups is crucial for individuals with Waardenburg syndrome, providing understanding, encouragement, and access to resources.
- **Genetic counselling** Genetic counseling plays a critical role in supporting individuals and families affected by Waardenburg syndrome, providing accurate information, addressing concerns, and facilitating informed decision-making.