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Waardenburg Syndrome-A Rare entity

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I. Introduction

Waardenburg syndrome is characterised by a pigmentary auditory genetic disorder including white forelock and heterochromia of the iris . It may also manifest as a congenital condition. The range and severity of associated symptoms and findings may vary greatly in presentation. It affects an estimated incidence of 1 in 40,000 of General population .There are 4 types documented in literature. Types I and II are the most common forms of Waardenburg syndrome, while types III and IV are rare[1]. Waardenburg syndrome is usually inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder. In most cases, an affected person has one parent with the condition. A small percentage of cases result from new mutations in the gene; these cases occur in people with no history of the disorder in their family.[2] Some cases of Waardenburg syndrome type II and type IV appear to have an autosomal recessive pattern of inheritance, which means both copies of the gene in each cell have mutations. Most often, the parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but do not show signs and symptoms of the condition genetic analysis have documented role of genes such as PAX3 ,WS2D-SNAI2(snail homolog2,Drosophilia),WS2A-MITF(micropthalia associated transcription factor) etc in the WS[3].

II. Case Report

A nine month old male infant was referred to the department of paediatric medicine for complaint of abdominal distention and inability to pass stools.The parents were illiterate and from a rural background and belonged to a interior village in Latur ,Maharashtra ,India.Due to lack of facilities a,proper diagnosis and care was not done.The child was born full term by normal delivery without any complications as such. Birth weight was 200gms as told by parents .Shortly after birth the baby was kept in Neonatal intensive care for 2 weeks .After four weeks the baby developed abdominal distention with irregular passage of stools.

Provisional diagnosis of Waardenburg Syndrome with Hirshprung disease was made and was confirmed with ultrasound of abdomen and other associated features. The patient reported this hospital at 9 months of age .On clinical examination the child appeared dull but alert to the surroundings.Body weight was 4000gms and head circumference was 40.5cms. Physical findings included blepharomosis with broad nasal root,white forelock and Heterochromia iris.there was a hypodermic patch on left forearm.(fig).Patient was showing poor hearing function for which on investigation , audiometry revealed severe SN deafness.

Rectal manometry showed Aganglionic segment and rectisigmoidal (+) based on which it further reconfirmed the diagnosis.

III. Discussion

Waardenburg syndrome was diagnosed on the basis of classic features such as Heterochromia Iris,hypodermic patch,white forelock ,sensorinueral loss and abdominal distention .The genetic cause in this case was not revealed due to familial poor socioeconomic limitations.None of the parents exhibited any of the syndromic features and neither any such familial history was revealed by parents in previous generations.Thus we believe that this case was a new mutation.the association between Ws and ganglionic megacolon is well known .Atretic GI abnormalities have been described in association with WS but never been considered part of the syndromic earlier[4.] current literature search describes workup on genetic evaluation with systematic classification. Clinically it is divided into 4 types based on the following clinical criteria .Presence (type I)and absence (type II) of dystropia canthorum.

IV. Management

The management depends on the severity and variability of the manifestations.hearing loss may be treated for cohlear implants in severe SNL cases.GI manifestations may be treated as per the requirement .The length of the aganglionic segment involved affects the clinical course of the disease and is also very important in surgical treatment planning.Enterostomy methods such as the Soave endorectal pull -through for short segments,the modified extended Duhamel method for long segments,the Swenson pull -through and the Kimura -Stringel operation are some of the series tried for the surgical treatment[5]. Post operative issues such fluid electrolyte balance maintenance,bacterial overgrowth and catheter infections are common which should be tackled with utmost care. Other manifestations such as white forelock and heterochromia Iris may be treated for cosmetic reasons.

V. Summary and conclusion.

This is an uncommon condition and proper documentation of cases with variable presentations with or without hereditary findings will definitely add to the incidence related and other diagnostic resources in the literature.

Acknowledgements

1. We thank the patient's family members for their consent in publication.. We also thank Dr. Suresh Narayan Singh for his help in manuscript writing.
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Table 1

	clinical manifestation	**incidence rate of Sensory Neural Hearing Loss**	**Genetic mutation**
Type 1	Dystopia canthorum, white forelock, white eyelashes, leukoderma, heterochromia iridis	60%	*PAX3*
Type 2	The absence of dystopia canthorum	90%	*MITF, WS2B, WS3B EDNRB, EDN3, SOX10 SNAI2*
Type 3	Type 1 + upper limb abnormalities	60%	*PAX3*
Type 4	Type 2 + Hirschsprung's disease	90%	*EDNRB, EDN3, SOX10*

Abdominal distention prominent in the patient of WS

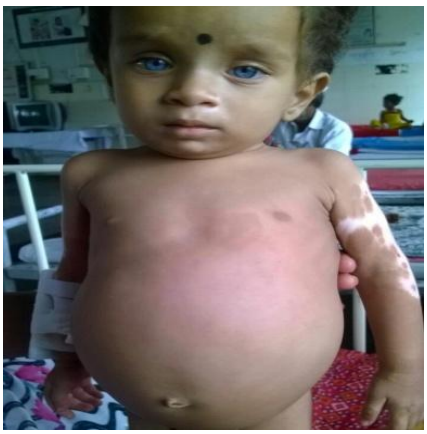

Fig 2-Hypopimented patch on forearm

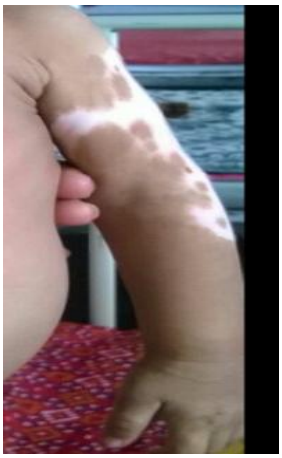

Fig 3-Heterochromia Iris

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