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A CASE REPORT ON WAARDENBURG SYNDROME : A RARE GENETIC DISORDER

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ABSTRACT Waardenburg syndrome (WS) is a uncommon autosomal inherited and genetically heterogenous disorder .Patients have heterochromia or eyes with iris of different colour,increases intercanthal distance,distopia canthorum ,pigmentation anomalies,Hirschprung's disease and varying degree of deafness.In this report,one case has been discussed with no family history of WS.Parents have been counseled regarding the disease and its prognosis and Hirschprung's disease is managed with colostomy.

KEYWORDS : Autosomal dominant,deafness,heterochromia,pigmentation anomalies,Hirschprung's disease,white forlock,Waardenburg syndrome

INTRODUCTION:

Waardenburg syndrome is a rare genetic disorder most often characterized by varying degrees of deafness, minor defects in structures arising from the neural crest, and pigmentation changes. It was first described in 1951.[1] The syndrome was later found to have four types.

Types I and II are the most common types of the syndrome, whereas types III and IV are rare. Type 4 is also known as Waardenburg-Shah syndrome (association of Waardenburg syndrome with Hirschsprung disease).

Type 4 is rare with only 48 cases reported up to 2002.[3][4]

CLASSIFICATION

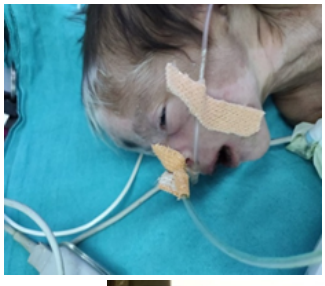
Type	Gene	Locus	Features
Type I	*PAX3*	2q36.1	• Almost always have eyes that appear widely spaced[2] • Hearing loss more common than in type II[2] • Hearing loss is congenital.[2]
Type II	*MITF*	3p14.1-p12.3	• Most common • Permanent hearing loss • Heterochromia of the irises
Type III	*PAX3*	2q36.1	• Abnormalities of the upper limbs • Hearing loss over time • Changes in skin pigmentation[2]
Type IV	*EDNRB*	13q22	• Characteristics of Waardenburg syndrome, in addition to Hirschsprung disease which can be life-threatening and requires surgery if the colon is enlarged.[2]

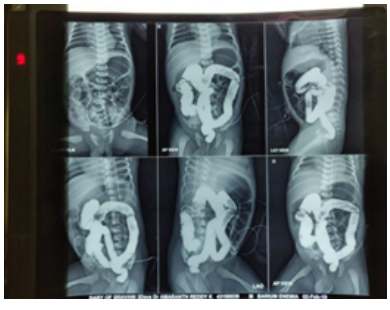
Type IV (WS4) can also affect portions of nerve cell development that potentially can lead to intestinal issues. Most commonly presents at neonatal age group.Type III is also known as Klein-Waardenburg syndrome, and type IV is also known as Waardenburg-Shah syndrome.[2]

Type I and II are most common types of WS.There is no cure for the syndrome till now.The symptoms most likely to be of practical significance include Deafness,Heterochromia and Intestinal obstruction(Hirschprung's disease) which are being treated symptomatically.

CASE REPORT

A 16 days old male babypresented with complains of not passing stools and bilious vomiting since birth,abdominal distension since 10 days.He was having bilateral pale colored iris ,white forelock ,depigmentation of upper part of trunk and both arms.He also had left anotia and abdominal distension with absent bowel sounds.On further evaluation,XRay Abdomen revealed multiple air fluid levels,USG Abdomen shows sluggish bowel peristalsis with echogenic content and Barium enema shows long segment aganglionic colon . All these features go in favour of Type IV WS ileostostomy was done for Hirschprung's disease.





DISCUSSION

This syndrome is autosomal dominant for most persons with Type I and III .Some cases of Type II and IV inherited as autosomal recessive with variable penetrance.A small percentage of cases result from new mutations in gene;these occur in people with no history of disorder in their family.

In our case report,baby had no family history of this syndrome.Early diagnosis and surgical management of Hirschprung's disease with colostomy is important in prevention of neonatal septic shock(DIC) and early death.

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