Abstract: Waardenburg syndrome is a rare disease characterized by sensorineural deafness associated with pigmentary anomalies and defects of neural crest derivative tissues. Waardenburg syndrome also known as Waardenburg Shah Syndrome, Waardenburg Klein syndrome, Mendes syndrome II, Van der Hoeve Halbertsma- Waardenburg syndrome. The frequency of Waardenburg syndrome is estimated to be 1 case per 212000 persons in general population but owing to low penetrance of about 20 the frequency of entire syndrome is approximately 1 in 42000. Children with Waardenburg syndrome have normal life expectancy. Morbidity is due to eye disorders, deafness, mental retardation skeletal anomalies. The following article is a case report of Waardenburg syndrome which presented to our opd with eye manifestations.

Keyword: Waardenburg syndrome, heterochromia iridium, sensorineural deafness.

On examination: She had hypertelorism and flat high nasal bridge. Right eye showed heterochromia iridium. Left eye anterior segment was normal. In both eyes pupils were round, regular, 3mm equally reacting to light with a vision of 6/6. She had an inter lateral canthus distance of 7.5 cms, inter medial canthus distance of 4.1 cms and inter pupillary distance of 5.3 cms. Ocular tension and corneal diameter of both eyes were normal. Right eye lacrimal passage was partially obstructed. On slit lamp examination there was no keratic precipitates or transillumination defects or vitreous disturbances.

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RIGHT EYE FUNDUS : ALBINOTIC
LEFT EYE FUNDUS : NORMAL
Patient was subjected to audiometry
RIGHT EAR : SENSORINEURAL DEAFNESS

LEFT EYE : NORMAL
STUDY
With the above findings we made the diagnosis of WAARDENBURG SYNDROME.

DISCUSSION
Waardenburg syndrome was first described in 1951. They are of 4 types with autosomal dominant inheritance. The gene mainly responsible for this syndrome is PAX 3 gene. DIAGNOSTIC CRITERIA

MAJOR
1. Hypopigmentation of hair
2. Pigmentary disturbance of iris
3. Sensoneural hearing loss
4. Affected first degree relatives
5. Dystrophia canthorum

MINOR
1. Broad high nasal bridge root
2. Hypoplasia of alae
3. Synorphrys
4. Congenital leucoderma
5. Premature greying of hair

2 Major or 1 major and 2 minor criteria are required to make the diagnosis. Our case had 3 major and 1 minor criteria. Since there was no family history this could be a sporadic case due to incomplete penetrance of the gene and variable expressivity. The diagnosis of this syndrome is essentially clinical.

CONCLUSION: There is currently no cure for Waardenburg syndrome. The symptom most likely to be of practical importance is deafness and is treated as any other hearing loss. Folic acid supplementation in pregnancy has been recommended for women at risk of having child with Waardenburg syndrome. Proper audiological assessment at birth and at periodic intervals can detect hearing impairment. This syndrome usually remains undiagnosed until another family member presents with features of this syndrome. This calls for examination of the entire family members of the diagnosed. Thus the early diagnosis of Waardenburg syndrome from cutaneous and ocular hypopigmentation may aid in the initiation of early treatment, social and vocational training and rehabilitation of these patients.

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