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WAARDENBURG SYNDROME TYPE II : A CASE REPORT

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ABSTRACT Waardenburg syndrome is a rare genetically inherited syndrome affecting hearing, eyes, skin and hair. It shows heterogenous charracteristics both clinically & genetically. We present a 5years 4 months old female child with type II Waardenburg syndrome, one of its 4 types.

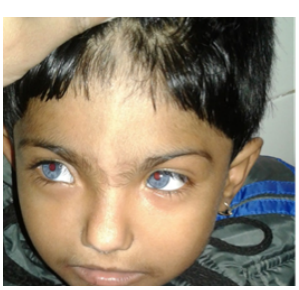
KEYWORDS : Wardenburg syndrome type II, blue iris, sensori-neural hearing loss.

INTRODUCTION :

Waardenburg syndrome is named after P.J. Waardenburg, an ophthalmologist and Dutch geneticist who first described the clinical charracteristics of the disease . It is a rare autosomally inherited disorder of neural crest development. He estimated the incidence of this syndrome to be 1.43% in congenitally deaf persons and 1:42,000 in general population (Waardenburg, P. J, 1951). It's most frequent clinical signs are sensorineural hearing loss, affecting about 60% of the patients; heterochromia of the iris; hypoplastic blue eyes; white streak; premature grey hair; leucoderma; high nasal root and hyperplasia of the medial portion of the eyebrows (synophrys) (DiGeorge et al, 1960). Waardenburg syndrome has 4 clinical types. Type I presents with a skin fold extending from the base of the nose to end of medial eyebrow region (epicanthus) increasing the distance of the internal medial corners of the eyes (canthorum dystopia), iris isochromia with bright blue color or heterochromia of iris, white hair streak (poliosis) that can appear at any age, confluent eyebrows (sinophrys) and changes in skin pigmentation. Type II differs from type I by not showing canthorum dystopia (Pardono E et al, 2003). Type III, also known as Klein Waardenburg Syndrome, has in addition to type I characteristics, microcephaly, malformation of the upper limbs and mental disabilities (Arnvig J, 1960) Type IV, also known as Waardenburg-Shah Syndrome presents Hirschsprung's disease in addition to Type II manifestations.

CASE REPORT: A 5years 4 months old female child born of parents with no history of consanguineous marriage presented with no speech development following congenital hearing loss. Child was born in hospital by normal vaginal delivery with adequate birth weight with no history of other diseases in neonatal period.Other milestones were normal. There is no history of constipation or persistent vomiting, limb abnormalities. Patient's father also has blue iris and synophrys but no hearing problem.

On examination , the patient has white forelock measuring 5x2cm ,bilateral blue iris, white hair streak, synophrys.



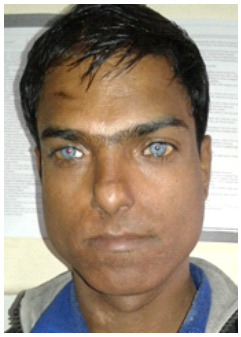


Figure 1. Clinical photograph of patient

Figure 2. Photograph Of Patient's Father..

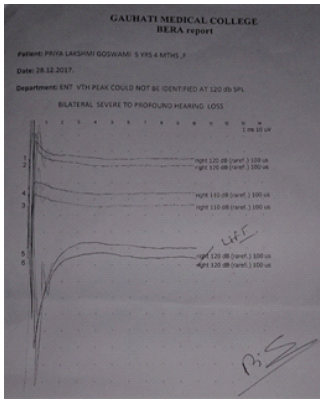


Figure 3. Patient's BERA report showing bilateral severe-profound sensorineural hearing loss.

DISCUSSION:

Waardenburg syndrome is inherited as autosomal dominant trait. WS type II occurs sporadically (few familial cases have been described in literature). No specific treatment is available. Presenting symptoms should be treated accordingly. Early detection of hearing loss and its management is beneficial for child's physical and mental development. For hearing deficit, patient may need hearing aids and proper schooling. Cochlear implants can be done successfully in such patients (Cullen RD et al, 2006). Prevention can be done by genetic counselling of parents.

So,with bilateral severe-profound sensorineural hearing loss, bilateral blue iris, white forelock, white streak of hair, synophrys and normal nasal root with absence of canthal dystopia, we can diagnose our case as Waardenburg syndrome type II.

REFERENCES:

1. Waardenburg, P. J. (1951). A new syndrome combining developmental anomalies of the eyelids, eyebrows and noseroot with pigmentary anomalies of the iris and head hair and with congenital deafness; Dystopia canthi medialis et punctorum lacrimalium lateroversa, hyperplasia supercilii medialis et radicis nasi, heterochromia iridum totaliis sive partialis, albinismus circumscriptus (leucismus, polioss) et surditas congenita (surdimutitas). American journal of human genetics, 3(3), 195.
2. DiGeorge, A. M., Olmsted, R. W., & Harley, R. D. (1960). Waardenburg's syndrome: A syndrome of heterochromia of the irides, lateral displacement of the medial canthi and lacrimal puncta, congenital deafness, and other characteristic associated defects. The Journal of pediatrics, 57(5), 649-669.
3. Pardono, E., van Bever, Y., van den Ende, J., Havrenne, P. C., Iughetti, P., Maestrelli, S. R., ... & Otto, P. A. (2003). Waardenburg syndrome: clinical differentiation between types I and II. American Journal of Medical Genetics Part A, 117(3), 223-235.
4. Arnvig, J. (1960). The Waardenburg-Klein syndrome. Nordisk medicin, 64, 953.
5. Cullen RD, Zdanski C, Roush P, Brown C, Teagle H, Pillsbury HC 3rd, Buchman C. Cochlear implant in waardenburg syndrome. Laryngoscope 2006 jul;116[7]; available at www. pubmed.gov ; accessed December 29, 2017.