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A rare case report on unusual presentation of waardenburg syndrome auto-somal genetic disorder

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Waardenburg syndrome is a rare hereditary disorder associated with higher incidence of Hirschsprung disease and total colonic or intestinal aganglionosis among neonates with clinical presentations i.e. bilious vomiting, gastrointestinal disorders, gene mutations, failure to feed, congenital deafness, pigmentation abnormalities (eye, forehead, hair and skin); thus, timely diagnosis, genetic counseling and improvements in hearing loss plays substantial effort in psychological wellbeing of neonates with this syndrome.

Keywords: Waardenburg Syndrome, Hirshsprung disease, Hypo-pigmentation, Gene Mutations, Hearing Loss

INTRODUCTION

Waardenburg Syndrome is a rare auto-somal genetic disorder had observed highly detected cases in Kenyan Africans with incidence approximately ranged from 1:20000 to 1:40000 (Rawlani et al. 2018) and 2 per 100000 globally (Kassem et al. 2018) with 2% to 5% cases of congenital deafness; caused due to absence of melanocytes in skin, stria vascularis of cochlea, eyes and hair; results in congenital hearing loss (Rawlani et al. 2018), hyppigmentation of skin and bluish discoloration of eyes among children; thus timely diagnosis of the syndrome through clinical investigations at birth or in early childhood or at later age plays a substantial role in rehabilitation of these children (Kassem et al. 2018; Pattebahadur et al. 2016).

CASE REPORT

The case report aims to highlight an unusual presentation of Waardenburg Syndrome in a 3 months old infant, known case of Hirshsprung disease and ileostomy was done for it (Figure 1), presented with Ileostomy diarrhea in Department of Pediatrics, KEMU/Mayo Hospital Lahore, Pakistan. Patient was admitted in the hospital with bilious vomiting and inability to pass meconium for 3 days after birth and exploratory laparotomy was done which showed total colonic aganglionosis. Patient is preterm male infant weighing 2.5kg at 34 weeks gestation was born via caesarean section to 33 years old mother, was on mother feed; can't respond to voices and developmentally delayed with no neck holding at 3 months of age. His mother had cousin marriage and elder brother has sensineural hearing loss but had no pigmentation. Antenatal history was unremarkable.

Histopathological findings revealed that stoma site consists of soft tissue piece measuring 0.8x0.7cm with origin of ganglionic cells and partial thickness on ileal mucosa. Appendix showed thin tubular structure of appendicular mucosa measuring 2.3x0.2cm with ganglionic cells. Transverse colon showed soft tissue piece of intestinal mucosa measuring 0.6x0.4cm with no ganglionic cells observed. Sigmoid colon was observed with the piece of soft tissue measuring

0.5x0.5cm; which on examination showed intact surface epithelium and inflammation in submucosa. 1-2 ganglion cells were also present in muscularis propria. Thus, revealed hypoganglionosis (hirschsprung disease) among patient. But no hypertrophic nerve fibre and granuloma or malignancy was seen. Immunohistochemistry is advised for confirmation of ganglion cells and also correlate clinically.

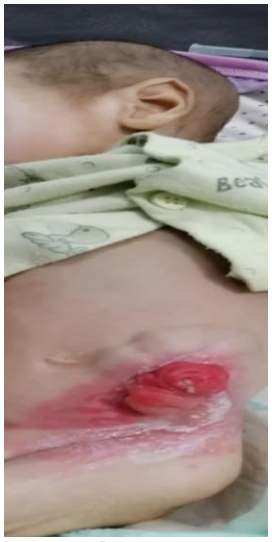

Figure 1

Figure 2

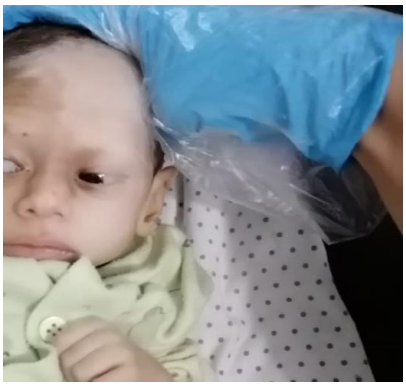

Figure 3

When the patient was 1 month old, hearing confirmatory test was performed where no response was observed uptil 110dBA which concluded profound degree of hearing loss among the patient and cannot be benefitted by the use of Hearing Aids at these levels. However, if parents showed willingness towards the trial with the use of Hearing Aids for a period of 3 to 4 months, only then the patient may be little benefitted with Super Power behind the Ear (BTE) hearing Aids with soft ear moulds in both ears.

When the patient was 6 month old, Ileostomy reversal was planned. He was not gaining weight with his growing age i.e. failure to thrive, has white patch on his left eye and forehead progresses to involve hair as well (Figure 2, 3), whereas, other areas did not show evidence of pigment changes.

DISCUSSION

A rare case of Waardenburg Syndrome was identified with unpredictable clinical presentations and symptoms i.e. bluish eyes, white forelock, bilious vomiting, deafness, denied mother feed and had intestinal obstruction i.e. failure to pass meconium among 3 months old infant in Pakistan (Pattebahadur et al. 2016; Khan et al. 2020). Waardenburg syndrome Type-IV had shown association with Hirschsprung disease; was an intestinal disorder that causes severe abdominal distention since birth and was observed through several ways such as physical examination, clinical investigations i.e. abdominal X-ray and rectal biopsy thus identified aganglionosis among patients (Mahmoudi et al. 2013).

Hypo-pigmentation is a pigmentary system disorder; causes defects in melanocytes number or function. The study findings had found association with Soni and his co-associates (2017) and Engin and Cayir (2015) that hypo-pigmentation refers to reduced pigmentation; causes change in skin color, hair and eyes due to

change in levels and locations of melanin producing melanocytes and genetic heterogeneity. Bluish eyes and hearing loss was also observed in Waardenburg Syndrome. Study findings found co-related with the Song and his co-authors (2016) research concluded that prevalence of hearing loss in Waardenburg Syndrome type 1 observed was 52.3% among individuals whereas, hearing loss in Waardenburg Syndrome type 2, type 3 and type 4 observed was 91.6%, 57.1% and 83.5% respectively.

Varied gene expressions attributed to the implications of Waardenburg Syndrome. Waardenburg Syndrome type 1 and type 2 were autosomal dominant inherited syndrome and was prevailed in most of the patients, Waardenburg Syndrome type 3 may be sporadic or autosomal dominant syndrome whereas; Waardenburg Syndrome type 4 is an autosomal recessive inherited syndrome. Waardenburg Syndrome type 1 and type 3 showed mutations on paired box gene-3 (PAX3) located on chromosome 2 and controls development of face and inner ear, whereas, Waardenburg Syndrome type 2 showed microphthalmia-associated transcription factor (MITF) also located on chromosome 2 and controls development of ear and hearing whereas, Waardenburg Syndrome type 4 showed Sex determining region Y-box-10 (SOX10) or endothelin-B receptor (EDNRB) gene mutations (Garg and Surana, 2012; Kumar and Rao, 2012).

CONCLUSION

The case report concluded that Waardenburg syndrome (WS) is a rare disorder associated with Hirschsprung disease and total colonic aganglionosis; was diagnosed through clinical and physical evaluations at birth or early childhood; thus reported to cause high occurrence of aliments and deaths among neonates. Therefore, early interventions, appropriate prognosis and parents counseling should be intervened to prevent inevitable aganglionosis and hearing loss and helps to improve psychological health of newborns who were suffering from Waardenburg syndrome.

CONFLICT OF INTEREST

The authors declared that present study was performed in absence of any conflict of interest.

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AUTHOR CONTRIBUTIONS

Dr. Maryam Shahid (corresponding author): initiated the concept, designed the study, contributed to data acquisition, interpretation of findings and case report writing.

Rukiya tariq: interpretation of findings and case report writing. Dr. Abdullah hussain: revised the case report, prepared figures and references.

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REFERENCES

Engin RI, Cayir Y, 2015. Pigmentation Disorders: A Short Review. Pigmentary Disorders 2: 189.

Garg U, Surana R, 2012. Waardenburg syndrome 2. Indian J Otol 18: 220-222.

Kassem LH, Ahmado MF, Alganameh MS, 2018. A rare case of seven siblings with Waardenburg syndrome: a case report. J Med Case Reports 12: 192.

Khan TA, Safdar CA, Zameer S, Khushdil A, 2020. Waardenburg-Shah syndrome (WS type IV): a rare case from Pakistan. Perioper Med 9: 4.

Kumar S, Rao K, 2012. Waardenburgsyndrome: A rare genetic disorder, a report of two cases. Indian J Hum Genet 18: 254–255.

Mahmoudi A, Rami M, Khattala K, Elmadi A, Afifi MA, Youssef B, 2013. Shah-Waardenburg syndrome. Pan Afr Med J 14: 60.

Pattebahadur R, Singhi S, Maharana PK, 2016. Waardenburg-Shah Syndrome: a rare case in an Indian child. BMJ case reports 2016: bcr2016216366.

Rawlani, S. M., Ramtake, R., Dhabarde, A., & Rawlani, S. S. (2018). Waardenburg syndrome: A rare case. *Oman journal of ophthalmology*, 11(2), 158–160.

Song J, Feng Y, Acke FR, Coucke P, Vleminckx K, Dhooge IJ, 2016. Hearing loss in Waardenburg syndrome: a systematic review. Clin Genet 89: 416-425.

Soni B, Raghavendra KR, Yadav DK, Kumawat P,

Singhal A, 2017. A clinico-epidemiological study of hypopigmented and depigmented lesions in children and adolescent age group in Hadoti region (South East Rajasthan). Indian J Paediatr Dermatol 18:9-13.