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Waardenburg Syndrome Associated With Nephroticsyndrome and Hypothyroidism – A Rare Case Scenario

Dr.Sravya Ravi[1], Dr.P.V.VSatyanarayana[2].
1(Post Graduate, Department of General Medicine, Rangaraya Medical College, Kakinada)
2(Professor, Department of General Medicine, Rangaraya Medical College, Kakinada).
Corresponding author: Dr.P.V.VSatyanarayana

Abstract: *Waardenburg syndrome is a group of rare genetic conditions that include: pigmentary disturbances of skin, eyes and nails, hearing loss, wide bridge of the nose etc. Symptoms vary from one type of the syndrome to another and from one patient to another. A 19 year old female came with the chief complaints of generalized swelling of the body associated with shortness of breath since 15 days. Similar episode is present in the past, 1 year before. She has Hypothyroidism since 2years, under medication.It is disease of improper migration of cells during embryogenesis that is inherited in autosomal dominant pattern. It is due to the defect in genes encoding the regulatory proteins of cell migration*

Keywords: *Nephrotic syndrome, Waardenburg syndrome.*

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I. Introduction

Waardenburg syndrome is named after a Dutch ophthalmologist Petrus Johannes Waaredenburg who described the syndrome in detail in 1951. The condition he described now categorized as WS1. It is a group of rare genetic conditions that include: pigmentary disturbances of skin, eyes and nails, hearing loss , wide bridge of the nose etc. Symptoms vary from one type of the syndrome to another and from one patient to another [1,2].

Waardenburg syndrome is of 4 types, presents with varying clinical features. Usually it is an autosomal dominant inheritance, but some types of type II and type IV are autosomal recessive pattern[3]. The overall incidence is ~1/42,000 to 1/50,000 people. 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 [4].

There is currently no treatment or cure for Waardenburg syndrome. The symptom most likely to be of practical importance is deafness, and this is treated as any other irreversible deafness would be. In marked cases there may be cosmetic issues. Other abnormalities (neurological, structural, Hirschsprung disease) associated with the syndrome are treated symptomatically.

II. Case Report

A 19 year old female came with the chief complaints of generalized swelling of the body associated with shortness of breath since 15 days. No history of fever, decreased urine output, jaundice. Similar episode is present in the past, 1 year before. She has Hypothyroidism since 2years, under medication. No history suggestive of Cirrhosis of liver, congestive cardiac failure, chronic kidney disease hypertension, diabetes and asthma. Positive family history in sister, father and paternal grandfather

General examination revealed anasarca with wide bridge of nose, heterochromiairides,white forelock. Vitals are normal. Normal cardiovascular examination Respiratory examination: decreased breath sounds bilaterally in basal areas. GIT examination: shifting dullness present. CNS examination: bilaterally, Rinnes test is negative.Schwabach's test- bone conduction of the patient is less than that of the examiner, patient is mute [Fig 1].

Fig 1. Showing facial features of Waardenburg syndrome patient

Urine analysis showednephrotic range proteinuria, lipid profile was abnormal, kidney biopsy suggestive of minimal change disease.Thyroid profile is within normal limits. Audiometry observation was profound sensorineural deafness bilaterally. Rheumatoid factor,nRNP/Smare positive. 2D echo is normal, chest X ray revealed bilateral pleural effusion, ultrasound abdomen showed ascites.

III. Discussion

It is disease of improper migration of cells during embryogenesis that is inherited in autosomal dominant pattern. It is due to the defect in genes encoding the regulatory proteins of cell migration

So far, 4 types are identified: type1- PAX3 gene mutations, type-2- MITF gene mutations-profound hearing loss,type-3/Klein-Waardenburg-PAX-3, type-4 /Shah-Waardenburg –EDN3/EDNRB gene mutations-associated with Hirschsprung disease. Most of the WS1 and WS3 patients were caused any abnormalities in the PAX3 gene [5].

MCD is an important cause of nephrotic syndrome and is characterized by massive proteinuria and hypoalbuminemia, resulting in edema and hypercholesterolemia. This case is a rare syndromic presentation [6].

In 1988 Tiger and de Chanderevian reported a case of WS associated with duplication of renal collecting system [7]. In 1997Jankauskiene et al observed a WS1 patientwas associated with Multicystic dysplastic kidney [8].In 2015 Pembegul et al documented the first association of WS1 with nephrotic syndrome [9]. In 2018, Anvesh G et al [6]reported a case ofnephrotic syndrome in a female of WS with autosomal dominant inheritance with variable penetrance. The genetic analysis could not be performed in our case. The probable genetic association between WS and nephrotic syndrome needs further evaluation.

IV. Conclusion

This is a rare case of Waardenburg syndrome in association with nephrotic syndrome and hypothyroidism.

References

[1]. "Waardenburg syndrome. Genetic and Rare Diseases Information Center (GARD) – an NCATS Program". rarediseases.info.nih.gov. Retrieved 2018-04-17.
[2]. "Waardenburg syndrome". Genetics Home Reference. October 2012
[3]. Dourmishev AL, Dourmishev LA, Schwartz RA, Janniger CK. Waardenburg syndrome. Int J Dermatol 1999;38:656-63.
[4]. Egbalian F (2008). "Waardenburg-Shah Syndrome; A Case Report and Review of the Literature" (PDF). Iranian Journal of Pediatrics. 18 (1): 71–74
[5]. Sheffer R, Zlotogora J. Autosomal dominant inheritance of Klein-Waardenburg syndrome. Am J Med Genet 1992;42:320-2.
[6]. Anvesh G, Raju SB, Prasad K, Sharma A, Surendra M. Rare association of waardenburg syndrome with minimal change disease. 2018;28:226-228.
[7]. Tiger P, de Chanderevian JP. Piebaldism-Waardenburg syndrome: Histopathologic evidence for a neural crest syndrome. Am J Med Genet. 1988;31:679-688

[8]. Jankauskiene A, Dodat H, Deiber M, Rosenberg D, Cochat P. Multicystic dysplastic kidney associated with Waardenburg syndrome type 1. PediatrNephrol. 1997;11:744-5.
[9]. Pembegul IY, Gurel A, Ulu R. Waardenburg syndrome and nephrotic syndrome coexistene SDU J Health Sci. 2015;6:41-42.

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