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Case Report

Waardenburg Syndrome

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Introduction

Waardenburg syndrome is a rare genetic disorder with at least 1,400 cases has been reported in medical lecture.In waardenburg syndrome a patient presents with hypopigmented areas of skin,white forelock of hairs,difference in the colour of both iris and some associated conditions like hirschsprung disease,malformation of upper limbs as well as certain abnormalities of CNS. [1]

It may be evident at birth (congenital).Primary features often include distinctive facial abnormalities diminished pigmentation of the hair, the skin, and/or the iris of both eyes (irides); and/or congenital deafness. In addition, pigmentary abnormalities may include a white lock of hair growing above the forehead (white forelock); premature graying or whitening of the hair; differences in the coloration of the two irides or in different regions of the same iris (heterochromia irides); and/or patchy, abnormally light (depigmented) regions of skin (leukoderma). Some affected individuals may also have hearing impairment due to abnormalities of the inner ear (sensorineural deafness). [2-4]

Case Report

A 3 months old female child,resident of Rawalpindi presented in Paediatric emergency with complaints of fever,cough, breathing difficulty and reluctance to feed.She presented with same complaints two times in the past.She had been diagnosed with complex congenital heart disease at 5th day of life and since then she was on antifailure medications.There was no history of loose stools,vomiting,abdominal distension and fits or any other complaints.On physical examianation she was febrile, with tachycardia and tachypenia with lower chest indrawing having HR 164/min, RR 74/min and temp 100F with failure to thrive with frontal bossing,coarse facial features,depressed nasal bridge,white forelockof hair,vitiligo on forehead and both lower limbs, and pansystolic murmur on left lower sternal border.Laboratory evaluation revealed complex congenital heart disease on Echocardiography(Large PDA ,ASD , Severe Tricuspid Regurgitation and Severe pulmonary HTN).Chest Xray showed increased pulmonary vascularity.On slit lamp examination there was brownish discolouration of bilateral iris. Rest of the investigations were unremarkable. She was given antibiotics(ceftriaxone and amikacin) and improved.

Figure 1: Waardenburg Syndrome- frontal bossing, coarse facial features, depressed nasal bridge, white forelock of hair,vitiligo on forehead

Discussion

Waardenburg syndrome (WS) is named after the investigator (PJ Waardenburg) who first precisely described the disorder in 1951. At least 1,400 cases have since been recorded in the medical literature.The disorder appears to affect males and females relatively equally. Waardenburg syndrome is a congenital disorder with different types. Waardenburg syndrome type I (WS1) and type II (WS2) are inherited as autosomal dominant traits with variable penetrance and expressivity. Some cases of Waardenburg syndrome type III (WS3) and type IV (WS4) appear to have an autosomal recessive pattern of inheritance. Mutations in the EDN3, EDNRB, MITF, PAX3 and SOX10 genes cause Waardenburg syndrome. [5]The risk of transmitting the disease gene from parent to offspring is 50 percent for each pregnancy regardless of the sex of the resulting child. In autosomal dominant disorders with variable penetrance and expressivity, manifestations of the disorder may not be present in all those who inherit the altered (mutated) gene for the disease. In those who do develop symptoms, the specific characteristics that are

manifested may vary greatly in range and severity from case to case. In some individuals with WS1 or WS2, there may be no apparent family history of the disorder. In such cases, researchers indicate that the disorder may sometimes result from new genetic changes (mutations) that occur spontaneously (sporadically) for unknown reasons. [6,7] In other instances, an apparent lack of a positive family history may be due to incomplete penetrance and/or variable expressivity. Evidence suggests that new (sporadic) mutations for WS1 may be associated with advanced age of the father (advanced paternal age).

Waardenburg syndrome (WS) may be diagnosed at birth or early childhood (or, in some cases, at a later age) based upon a thorough clinical evaluation, identification of characteristic physical findings, a complete patient and family history, and various specialized studies. Additional studies may include examination with an illuminated microscope to visualize internal structures of the eyes (slit-lamp examination); specialized hearing (auditory) tests; and/or advanced imaging techniques, such as to evaluate inner ear abnormalities. [8]

Such treatment may require the coordinated efforts of a team of medical professionals, such as physicians who specialize in skin disorders (dermatologists); eye specialists (ophthalmologists); hearing specialists. Early recognition of sensorineural deafness may play an important role in ensuring prompt intervention and appropriate supportive management. Because individuals with pigmentary abnormalities of the skin may be prone to sunburns and a risk of skin cancer, physicians may recommend avoiding direct sunlight, using sunscreen with a high sun protection factor (SPF), wearing sunglasses and coverings that help to protect against the sun (e.g., hats, long sleeves, pants, etc.), and following other appropriate measures. [9,10]

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