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Dyschromatosis universalis hereditaria with STRC- related sensorineural hearing loss in a child: A dual genetic diagnosis

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Description

8-year-male child, born of a second- degree consanguineous marriage, a known case of moderate intellectual disability was referred from the paediatric outpatient department with complaints of bilateral hearing loss of insidious onset, gradually progressive initially noticed at the age of four years by his parents along with defective speech development. The hearing loss had become noticeable over time and is now significantly affecting his routine communication and academic activities. The child was born of a full-term pregnancy via caesarean section, with an unremarkable antenatal and perinatal history. There were no complications at birth and the neonatal period was uneventful. Developmentally, the child had delayed milestones and showed poor scholastic performance. Formal cognitive assessment revealed moderate intellectual disability associated with behavioural disturbances. There was no history of seizures or developmental regression. At around 01 year of age, parents noticed mottled skin pigmentation, which gradually spread to involve the face, trunk and extremities. The lesions were non-pruritic and consisted of interspersed hyperpigmented and hypopigmented macules with sparing of palms, soles and mucosa.

There is a negative family history of similar cutaneous, auditory or neurodevelopmental disorders. On examination, the child had diffuse, irregularly shaped, mottled pigmented macules over lower face, trunk and extremities (Figure 1). Systemic and neurological examinations were unremarkable. Otoscopic findings were normal. Pure tone audiometry (PTA) demonstrated bilateral moderate sensorineural hearing loss (Figure 2) and Brainstem Evoked Response Audiometry (BERA) showed wave V at 60 dBnHL on Right and at 50 dBnHL on left side (Figure 3). Punch biopsy from a hypopigmented lesion over back revealed basal layer pigment incontinence with dermal melanophages, consistent with Dyschromatosis Universalis Hereditaria (DUH). Genetic testing identified a homozygous a pathogenic mutation in the STRC gene (stereocilin), confirming a diagnosis of autosomal recessive non-syndromic sensorineural hearing loss (DFNB16 related). Though Genetic analysis for ABCB6 (ATP Binding Casette Subfamily B Member 6) was not done due to unavailability and high cost; nevertheless, the diagnosis of DUH was made clinically and histopathologically.

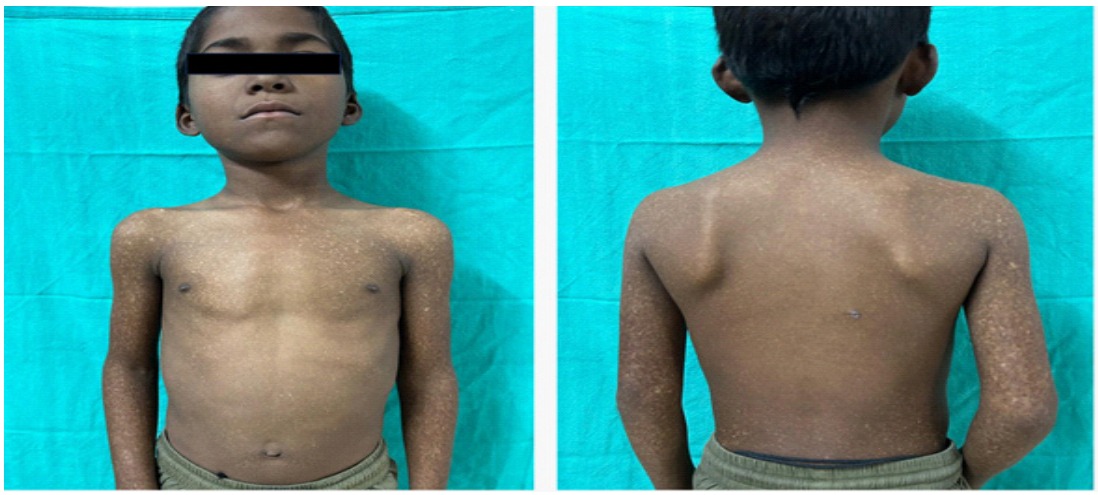

Figure 1: Cutaneous examination revealed generalized symmetrically distributed hyperpigmented and hypopigmented lesions on the trunk, lower face and upper extremities.

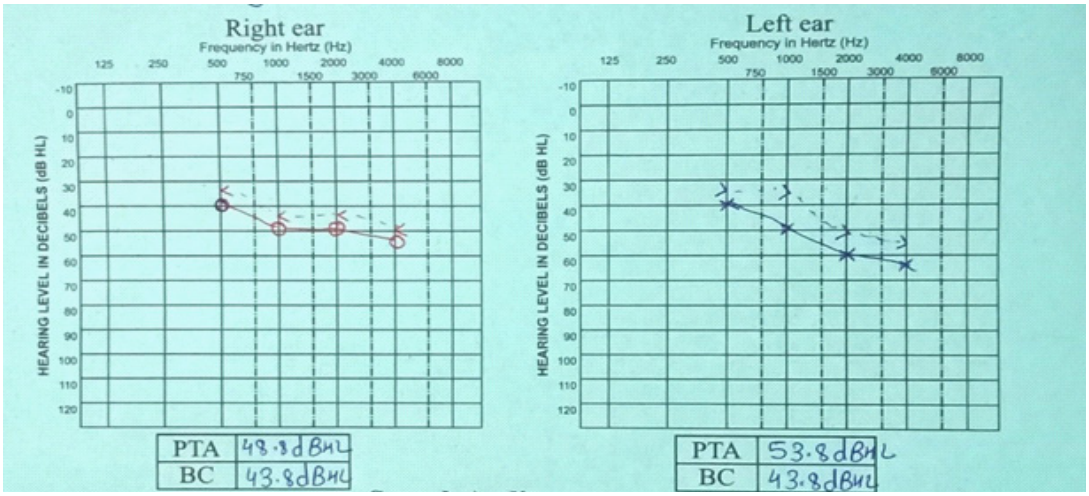

Figure 2: Pure tone audiometry (PTA) showing bilateral moderate degree of Sensorineural hearing loss.

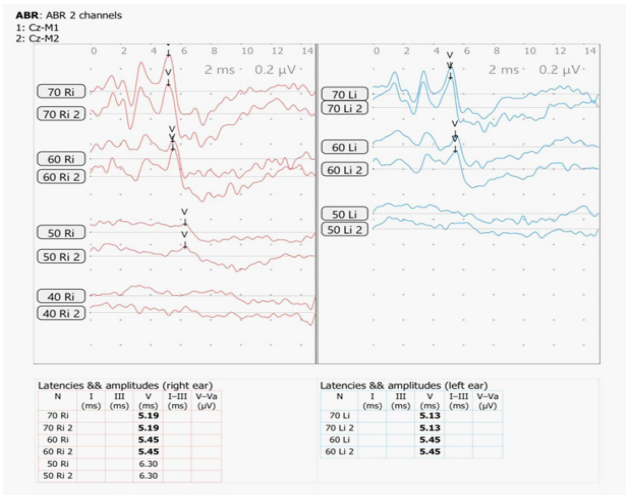

Figure 3: Brainstem evoked response audiometry (BERA) showing wave V at 60 dBnHL on Right and at 50 dBnHL on left side.

Discussion

DUH is a rare genodermatosis with autosomal dominant inheritance, rare reports have suggested potential systemic involvement, including neurological and auditory abnormalities. ABCB6 is the most commonly implicated gene in DUH and mutations in the transporter protein can also affect inner ear development, leading to hearing deficits [1]. STRC gene is associated with autosomal recessive non- syndromic hearing loss, specifically DFNB16 and its pathogenic variants are among the more common genetic causes of mild to moderate hearing loss [2]. Genetic studies are necessary to elucidate the various associations of DUH like sensorineural hearing loss [3].

This case illustrates a dual genetic diagnosis in a child involving clinically and histopathologically confirmed DUH and genetically confirmed STRC related non-syndromic hearing loss [4]. The co-existence of two genetically distinct conditions is likely attributable to consanguinity and highlights the importance of not assuming syndromic overlap when multiple symptoms are present. This case demonstrates the value of targeted genetic testing in establishing a precise diagnosis and guiding counselling.

Declarations

Conflicts of interest: Nil,

Informed consent: Informed consent was taken from the patient for this publication.

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