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The BIMJ publishes articles ranging from original research papers, review articles, medical practice papers, special reports, audits, case reports, images of interest, education and technical/innovation papers, editorials, commentaries and letters to the Editor. Topics of interest include all subjects that relate to clinical practice and research in all branches of medicine, basic and clinical including topics related to allied health care fields. The BIMJ welcomes manuscripts from contributors, but usually solicits reviews articles and special reports. Proposals for review papers can be sent to the Managing Editor directly. Please refer to the contact information of the Editorial Office.

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Jeyasakthy SANIASIAYA

Figure 1

A 5-month-term neonate was referred to the Otorhinolaryngology Department for assessment. The parents noticed that child did not react to any form of sound. Both birth and antenatal history was unremarkable. In addition, there was no family history of deafness. Upon assessment, the child was noted to have different coloured irises with skin and hair pigmentation (Figure 1). Clinical examination revealed complete heterochromia and dystopia canthorum. No other dysmorphic or craniofacial abnormalites were noted. There was no white forelock. Auditory brainstem responses revealed bilateral profound sensorineural hearing loss.

What is the diagnosis?

Answer*: refer to page 7*

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DISCLOSURE: There is no conflict of interest and consent has been obtained from the child's parents for use of these images.

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(***Refer to page 6***)

ANSWER: KLEIN-WAARDENBURG SYNDROME

Figure 1: Asymmetry between iris colour with dystopia canthorum and skin hypopigmentation is noted.

Child was diagnosed with Waardenberg syndrome based on its consortium criteria. Bilateral cochlear implant was planned and referral was made to paediatrics team for futher assessment.

Heterochromia iridis (HI) can either be inherited or due to genetic mosaicism, chimerism, disease or injury.[1] Congenital heterochomia iridis has also been linked to intrauterine disease or trauma in addition to familial aetiology (usually autosomal-dominant trait).[2] Myriad congenital syndromes presenting with HI including Waardenburg syndrome, congenital Horner's syndrome, Sturge-Weber syndrome, Recklinghausen disease, Bourneville disease and Klippel-Trenaunay syndrome.[2]

Waardenburg syndrome is a mainly autosomal-dominant genetic condition, due to a PAX3 mutation on chromosome 2q37. Type IV is autosomal recessive. Sytemic and ocular findings include white forelock, skin pigmentation, telecanthus and heterochromia iridis.[3] We would like to highlight that all infants born with HI require a thorough assessment including hearing assessment as early detection of hearing loss and management is imperative for speech and language development.

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