(***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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