Clinical Quiz

What is the Diagnosis?

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The proband is a 15 month old girl. She was born full term in Hong Kong to a non-consanguineous Chinese couple. Antenatal checkup was normal. Postnatally, she had bilateral profound hearing loss detected at newborn and ear anomalies were noted. She was referred to genetic clinic for the above. Physical examination at 15 month old of age revealed short stature (69 cm <3rd) and failure to thrive (7.845 kg <3rd centile), subtle facial dysmorphism (mild hypertelorism, depressed nasal bridge and facial asymmetry), bilateral pre-auricular pits and bilateral neck sinus with clear discharge. She was on hearing aids. Her vision and cardiovascular examination was unremarkable. She had normal renal function and structure by ultrasound. She had borderline developmental delay. No significant family history of hearing impairment or renal problem was noted. The body weight and body height centiles were all below 3rd centile in subsequent follow up. Subsequent MRI temporal bone showed bilateral cochlear hypoplasia, type II incomplete partition anomaly with deficient cochlear nerve. She had right cochlear implant surgery subsequently.

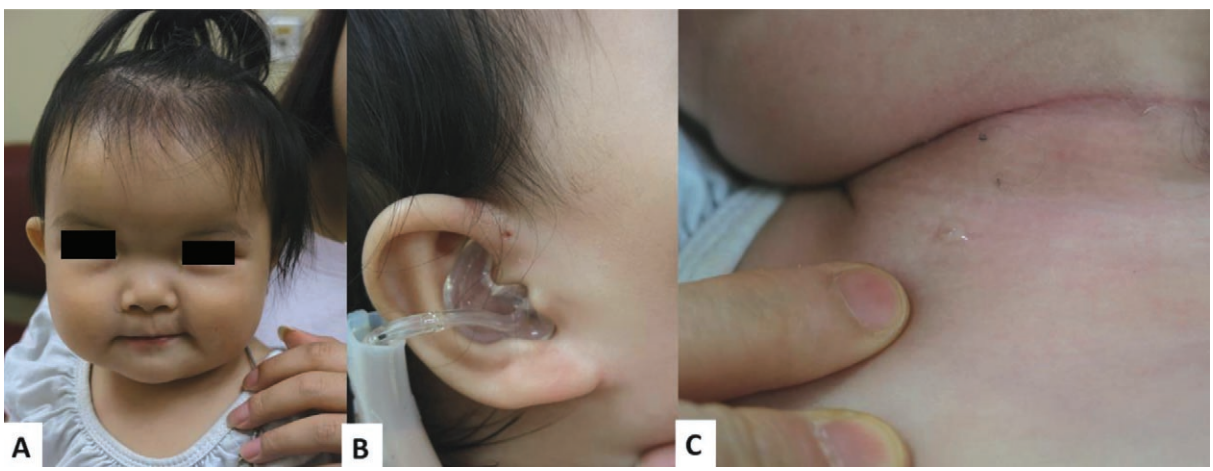

Figure A. Facial profile; B. Right preauricular pit and with hearing aids; C. Neck sinus with clear discharge (with consents for publication by parents).

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N.B. The Editors invite contributions of illustrative clinical cases or materials to this section of the journal.