Te whakamātau i te rongo a te pēpi hou | Universal hearing screening for newborns (UNBHS): Much more than meets the ear.

# Early Ears, Early Action: The Vital Role Of Newborn Hearing Screening

## A whānau's journey navigating their pēpi's incidental diagnosis of mitochondrial short-chain enoyl-CoA hydratase 1 deficiency (ECHS1) via the Newborn Hearing Screening.

Clinical presentation of ECHS1 includes cardiomyopathy, developmental delays, neuroregression, dystonia, visual impairment, seizures, and brain MRI abnormalities consistent with Leigh syndrome (LS).

There are less than **60 cases** worldwide with around **46%** of cases presented with some degree of SNHL.

19 & 28 days old (corrected)

Listeninglab.my (2025)

### Initial Screening

Pēpi was born at 42+3 gestation via c-section with no risk factors. UNBHS was attempted twice: INC (interference) from the right ear and "refer" from the left ear.

1 month + 2 & 9 days old (corrected)

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### Bilateral normal steeply sloping to severe mixed HL confirmed

- Conductive overlay without any evidence of ME dysfunction in either ear
- No evidence of ANSD
- Referred to ENT

2 month 3 days (corrected)

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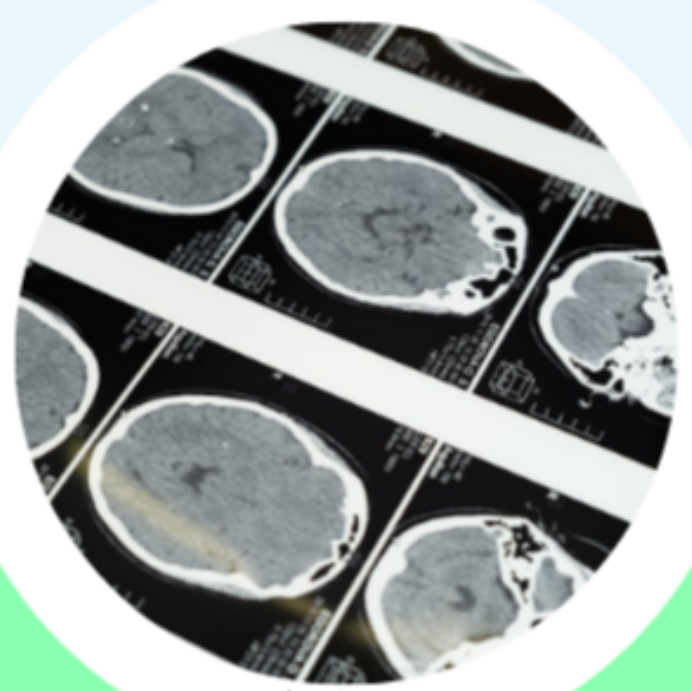
### CMV -ve confirmed

Cytomegalovirus (CMV) was not present in urine sample taken.

2 months 22 days (corrected)



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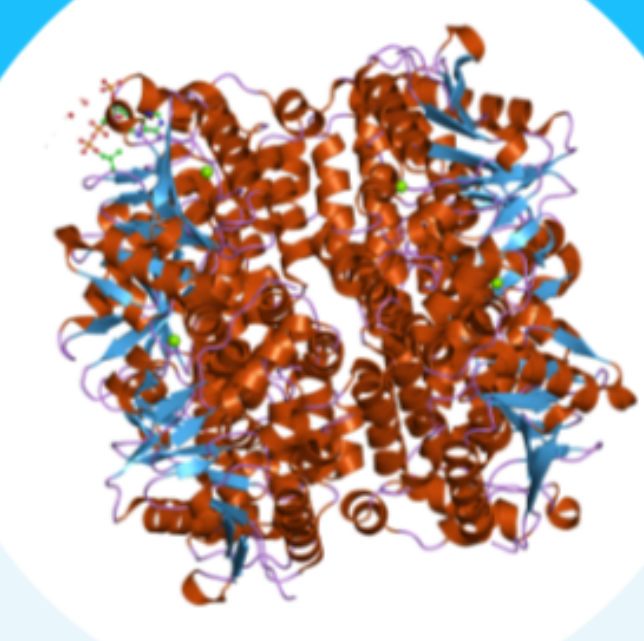
### Abnormality in putamen confirmed bilaterally

- MRI confirmed:
  - Normal cochlear nerves and inner ear structures; and
  - Abnormal MRI with T2 hyperintensity in the basal ganglia (putamen) bilaterally.
- Referred to Paediatrician and Metabolic Consultant.

2 months 28 days (corrected)



wikimedia.org

### ECHS1 diagnosis confirmed

Established management plan of pēpi's hauora, medical needs and overall development. Referred for further genetic counselling, ophthalmology assessment, and support from Child Development Services and Dietetics.

4 months (corrected)

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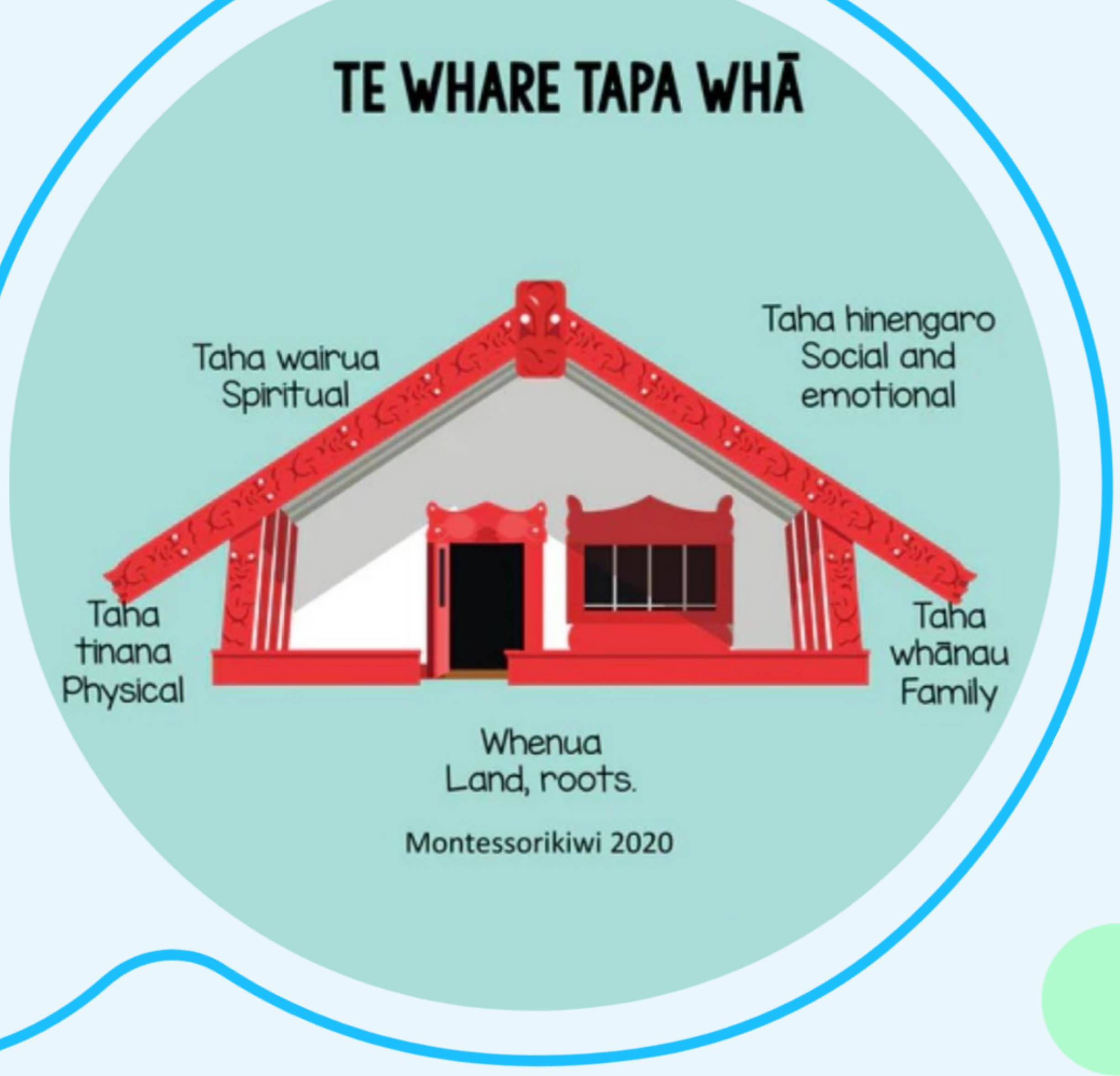
### Hearing aids fitted bilaterally

Pēpi's hearing development and needs continue to be monitiored

Currently, there is **no cure** for ECHS1 deficiency. Few individuals survive into adulthood with multiple disabilities.



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**Health New Zealand**
**Te Whatu Ora**

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