Guidelines for Assessment of Children with Hearing Loss 2022 (QLD)

Queensland Government

Clinical Assessment

History

History to assess aetiology and guide management of hearing loss	
Antenatal history	• Spontaneous/recurrent miscarriages • Infections, febrile/flu-like illnesses • Drug/alcohol use • Medication use (e.g. ototoxic medications) • Known in-utero infections: cytomegalovirus (CMV), toxoplasmosis, herpes simplex virus (HSV), varicella, rubella, syphilis, human immunodeficiency virus (HIV) • Maternal immunisation status and serological results: rubella immunity (rubella IgG>10IU/ml), syphilis, HIV and varicella testing • Other investigations: ultrasound, amniocentesis, chorionic villus sampling, first trimester combined screen, non-invasive prenatal testing
Birth history	• Gestation • Delivery type • Condition at birth (e.g. Apgar scores) • Birth weight, length and head circumference (chart percentiles) • Complications/interventions at delivery (e.g. resuscitation, respiratory/cardiovascular support, admissions to special care nursery/intensive care, duration of admission, sepsis, hypoglycaemia, seizures, use of ototoxic medications e.g. aminoglycosides/loop diuretics) • Jaundice requiring phototherapy/exchange transfusion, peak serum bilirubin • Abnormalities noted/suspected/diagnosed at birth
Post-natal history	• Bacterial meningitis • Head injuries • Proven/suspected congenital infections (e.g. CMV/toxoplasmosis/HSV/rubella/syphilis) • Exposure to ototoxic medications (e.g. aminoglycosides, loop diuretics, chemotherapy especially cisplatin) • Cranial irradiation • Motor delay/balance issues (consider vestibular dysfunction) • Haematuria (Alport syndrome)
Family history: Three-generation family tree	• Audiograms from first degree relatives • Consanguinity • Family history of hearing loss, goitre (Pendred syndrome), pigment abnormalities (Waardenburg syndrome), congenital renal anomalies, renal failure (Alport syndrome, branchio-oto-renal syndrome), short stature (Stickler syndrome, 22q deletion), cardiac malformations (22q deletion, CHARGE syndrome), arrhythmias, sudden death (Jervell Lange Nielson syndrome), vision issues (Usher syndrome), ear and neck malformations, developmental delays, neurological conditions
Social history	• Other specialists/services involved, access to financial supports/service pathways (including Carer Allowance, National Disability Insurance Scheme) • Migration history (including risk factors for congenital infection) • Language, family structure, employment • Adjustment, impact on family, resilience and coping factors, including supports

Examination

Key characteristics on physical examination		
Examination systems	**Physical features**	**Examples of diagnoses to consider**
Growth (weight, length, head circumference percentiles)	• Microcephaly • Low birth weight	• Congenital infections
Development	• Developmental delay/regression • Gross motor delay	• Congenital infections/metabolic disorder • Neurological causes (e.g. prematurity, hypoxic ischaemia encephalopathy • Gross motor delay may be an early manifestation of vestibular dysfunction (consider Pendred, Usher)
General	• Dysmorphology • Pigmentation: skin / hair • Coarse features • Blood pressure	• Syndromic causes • Waardenburg syndrome • Metabolic / storage disorder • Renal causes
Head and neck	• Facial asymmetry • Abnormal external ears • Abnormal ear canals • Preauricular sinuses / pits / tags • Cleft palate / submucous cleft / bifid uvula • Tympanic membrane status • Goitre	• Syndromic / developmental causes (especially with unilateral hearing loss e.g. branchio-oto-renal syndrome) • Middle ear fluid • Pendred (goitre onset usually in mid-late childhood)
Eyes	• Cataracts • Retinal scarring (fundoscopy) • Microphthalmia • Hypertelorism / heterochromia iridium • Retinitis pigmentosa (fundoscopy)	• Congenital infection • Congenital infection • Syndromic cause • Waardenburg syndrome • Usher syndrome (retinitis pigmentosa onset usually in mid-late childhood; electroretinography best screening tool)
Neurologic	• Hypotonia • Ataxia • Focal neurological signs	• Neurological causes (e.g. prematurity, hypoxic ischaemia encephalopathy, space occupying lesions)
Cardiac	• Murmur	• Cardiac causes
Skeletal	• Spine • Digits / nails / hyperextendable joints	• Bony dysplasias • Connective tissue disorders
Abdominal	• Organomegaly	• Storage disorders
Urine dipstick / microscopy	• Haematuria	• Alport (onset usually late childhood)

Investigations

Investigations for childhood sensorineural hearing loss (SNHL) and auditory neuropathy (AN)

Tier 1: First line recommendations		
Investigation	**Bilateral SNHL/AN**	**Unilateral SNHL/AN**
CMV PCR	< 21 days - saliva (>1hr after breast feed) > 21 days - dried blood spot (Guthrie card)	< 21 days - saliva (>1hr after breast feed) > 21 days - dried blood spot (Guthrie card)
MRI scan: **brain and internal auditory canal**	All infants early (attempt feed and wrap +/- sedation), esp. if: • Severe to profound HL • Asymmetric HL • Auditory neuropathy • Other indications Children (under GA): • Cochlear implant candidate • Progression of HL • Auditory neuropathy • Other indications	All infants early (attempt feed and wrap +/- sedation): • Moderate or worse HL Children (under GA): • Cochlear implant candidate • Auditory neuropathy • Progression of HL • Other indications
Connexin 26/30	All patients Not recommended if: • Conductive HL • Structural external ear anomalies • Developmental delay/regression • Congenital anomalies • Dysmorphic features • Complex phenotype	Not recommended
Ophthalmology exam	Any age: • Profound or progressive HL • Concerns about vision (esp. night vision) • Congenital infections • Syndromic features • Developmental motor delay, hypotonia, poor coordination Age 18 months: Not yet walking School entry: All patients (visual acuity)	Any age: • Profound or progressive HL • Concerns about vision (esp. night vision) • Congenital infections • Syndromic features • Developmental motor delay, hypotonia, poor coordination Age 18 months: Not yet walking School entry: All patients (visual acuity)
Family audiograms (1st degree relatives)	All patients	All patients

Tier 2: For specific clinical indications		
Investigation	**Bilateral SNHL/AN**	**Unilateral SNHL/AN**
Chromosome microarray	• Developmental delay/regression • Congenital anomalies • Dysmorphic features • Complex phenotype • Non-syndromic SNHL with negative connexin testing	• Developmental delay/regression • Congenital anomalies • Dysmorphic features • Complex phenotype
Mitochondrial DNA	• SNHL following aminoglycoside therapy • Family history of SNHL with matrilineal inheritance • Clinical and/or family history suggestive of mitochondrial disorders (e.g. diabetes, cardiomyopathy, epilepsy, myopathy)	• SNHL following aminoglycoside therapy • Family history of SNHL with matrilineal inheritance • Clinical and/or family history suggestive of mitochondrial disorders (e.g. diabetes, cardiomyopathy, epilepsy, myopathy)
Thyroid function	• Developmental delay • Abnormal growth • Goitre • Early newborn bloodspot screen (<48hrs of age)	• Developmental delay • Abnormal growth • Goitre • Early newborn bloodspot screen (<48hrs of age)
Perinatal infection testing	• 'At-risk' with known maternal infections • Suggestive ophthalmology findings	• 'At-risk' with known maternal infections • Suggestive ophthalmology findings
Metabolic testing	As clinically indicated, including: • Developmental delay/regression • Intellectual disability • Autism • Encephalopathy • Hepatosplenomegaly • Features of storage disorders • Progressive SNHL • Family history of Brown-Vialetto-Van Laere syndrome	As clinically indicated, including • Developmental delay/regression • Intellectual disability • Autism • Encephalopathy • Hepatosplenomegaly • Features of storage disorders • Progressive SNHL • Family history of Brown-Vialetto-Van Laere syndrome
CT scan **petrous bone / inner ear**	As determined by ENT clinicians for indications such as: • Permanent conductive HL • Further information beyond MRI required	As determined by ENT clinicians for indications such as: • Permanent conductive HL • Further information beyond MRI required
Renal ultrasound	• Multisystem anomalies • Family history of renal malformations associated with HL • Preauricular pits, cup ears or ear anomalies AND cochlear/vestibular malformations, branchial anomalies, family history of HL, or maternal gestational diabetes	• Multisystem anomalies • Family history of renal malformations associated with HL • Preauricular pits, cup ears or ear anomalies AND cochlear/vestibular malformations, branchial anomalies, family history of HL, or maternal gestational diabetes
Urinalysis	• Delayed onset HL • Progressive HL • Family history of renal disease (e.g. Alport syndrome, MYH9-related disorder, Fabry disease, Alstrom syndrome, and distal renal tubular acidosis)	• Delayed onset HL • Progressive HL • Family history of renal disease (e.g. Alport syndrome, MYH9-related disorder, Fabry disease, Alstrom syndrome, and distal renal tubular acidosis)

Vestibular testing	• Progressive SNHL • Developmental motor delay, poor balance • Temporal bone anomalies • Suspicion of Usher or Pendred syndrome	• Progressive SNHL • Developmental motor delay, poor balance • Temporal bone anomalies • Suspicion of Usher or Pendred syndrome
ECG	• Severe to profound SNHL • Syncopal episodes • Family history of unexplained sudden death, arrhythmias, or syncope	Not recommended

Tier 3: Aetiology still unknown and/or older children		
Investigation	**Bilateral SNHL/AN**	**Unilateral SNHL/AN**
Genomic testing **Targeted gene panel / whole exome sequencing (WES) / whole genome sequencing (WGS)** **Recommend discussion with clinical genetics service*	Microarray testing negative AND: • Developmental delay/regression • Congenital anomalies • Dysmorphic features • Complex phenotype Connexin/microarray testing negative: • Non-syndromic SNHL	Microarray testing negative AND: • Developmental delay/regression • Congenital anomalies • Dysmorphic features • Complex phenotype
MRI scan: brain and internal auditory canal	School entry (non GA)	School entry (non GA)
Ophthalmology exam	Age 7-9 years (+/- electroretinography for Usher syndrome)	Age 7-9 years (+/- electroretinography for Usher syndrome)
Urinalysis	Age 10 years	Age 10 years

Referrals

Referrals for children with hearing loss	
Service / Health professional	**Indications and timing**
Audiology	All patients • At diagnosis • 6-9 months • 6 monthly-yearly until age 5 years • Yearly > 5yrs (unless deterioration)
ENT surgery	• Consideration of cochlear implant • Progressive HL • Permanent conductive HL (incl. microtia) • Middle ear disease/consideration of grommets
Paediatrician	All patients at diagnosis of HL for assessment Further review by paediatrician or GP AND child health/school based nurse: • 9 months • 15-18 months • 2 years • 3 years • 4 years • 5 years (school entry) • 10-11 years (primary school exit) • 12 years (secondary school entry)
Early intervention / allied health	• Moderate to profound bilateral HL • Mild HL if developmental concerns • Global developmental delay
Clinical genetics	As clinically indicated and with agreement with patient/parents • Diagnostic assessment for syndromic HL • Diagnostic genomic testing • Reproductive counselling for parents/patient Not recommended if: • Non-syndromic unilateral SNHL • Non-syndromic unilateral/bilateral conductive HL
Optometry	All patients • From 12 months • 5 years (school entry)
Ophthalmology	Any age: • Profound or progressive HL • Concerns about vision (esp. night vision) • Congenital infections • Syndromic features • Developmental motor delay, hypotonia, poor coordination Age 18 months: Not yet walking School entry: All patients (visual acuity) Age 7-9 years: Aetiology unknown (+/- electroretinography)
Nephrology	• Renal malformations • Abnormal urinalysis
Cardiology	• Cardiac malformations • Abnormal ECG • Family history of cardiomyopathy
Neurology	• Neurological impairment • Abnormal MRI brain • Family history of neuropathy
Family audiograms	All patients at diagnosis
**Consult local audiology department for local services and pathways*	

References:

Sung et. al. Childhood Hearing Australasian Medical Professionals network: Consensus guidelines on investigation and clinical management of childhood hearing loss. J Paediatrics Child Health (2019) 55:1013-1022