Healthy Hearing Program

Medical Guidelines for Children born with a Permanent Hearing Loss
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Disclaimer

These guidelines have been prepared to promote and facilitate standardisation and consistency of practice, using a multidisciplinary approach.

Information in this guideline is current at time of publication.

Queensland Health does not accept liability to any person for loss or damage incurred as a result of reliance upon the material contained in this guideline.

Clinical material offered in this guideline does not replace or remove clinical judgement or the professional care and duty necessary for each specific patient case.

Clinical care carried out in accordance with this guideline should be provided within the context of locally available resources and expertise.

This Guideline does not address all elements of standard practice and assumes that individual clinicians are responsible to:

- Discuss care with consumers in an environment that is culturally appropriate and which enables respectful confidential discussion. This includes the use of interpreter services where necessary.
- Advise consumers of their choice and ensure informed consent is obtained.
- Provide care within scope of practice, meet all legislative requirements and maintain standards of professional conduct.
- Apply standard precautions and additional precautions as necessary, when delivering care.
- Document all care in accordance with mandatory and local requirements.
## Abbreviations

<table>
<thead>
<tr>
<th>Abbreviation</th>
<th>Description</th>
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<tr>
<td>CI</td>
<td>Cochlear implant</td>
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<tr>
<td>CMV</td>
<td>Cytomegalovirus</td>
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<td>CT</td>
<td>Computerised tomography</td>
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<td>DBS</td>
<td>Dried blood spot</td>
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<td>EVA</td>
<td>enlarged vestibular aqueduct</td>
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<td>FH</td>
<td>Family history</td>
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<tr>
<td>FSF</td>
<td>Family Support Facilitator</td>
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<td>GA</td>
<td>General anaesthetic</td>
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<td>HH</td>
<td>Healthy Hearing</td>
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<tr>
<td>HIE</td>
<td>Hypoxic ischaemic encephalopathy</td>
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<td>IAM</td>
<td>Internal auditory meati/meatus</td>
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<td>IgG</td>
<td>Immunoglobulin G</td>
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<tr>
<td>IgM</td>
<td>Immunoglobulin M</td>
</tr>
<tr>
<td>IPPV</td>
<td>Intermittent positive pressure ventilation</td>
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<td>JLNS</td>
<td>Jervell and Lange-Nielsen syndrome</td>
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<td>MRI</td>
<td>Magnetic resonance imaging</td>
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<td>PAP</td>
<td>positive airway pressure</td>
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<td>PCHL</td>
<td>Permanent childhood hearing loss</td>
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<tr>
<td>PCR</td>
<td>Polymerase chain reaction</td>
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<td>PHL</td>
<td>Permanent hearing loss</td>
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<td>PHR</td>
<td>Personal health record</td>
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<td>QHLFSS</td>
<td>Queensland Hearing Loss Family Support Service</td>
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New born screen - Guthrie card  
DBS  

interchangeable
1. Introduction

1.1 Background

Children born with a permanent hearing loss run the risk of life long speech deficits and delays in the acquisition of language. Deficits in speech and language may result in poor academic performance, psycho-social difficulties, behaviour and emotional problems.

Technological developments in recent years have widened the scope of potential outcomes for the development of communication skills for children born with a permanent hearing loss. This together with early screening processes and research providing evidence of the value of prompt early intervention services (Yoshinaga-Itano, 2004) provide a window of time for making a real difference in long term outcomes for children with PHL and their families. To make the best use of these opportunities, it is necessary to develop and set in place efficient systems to guide and co-ordinate parents and practitioners through the complex medical assessments and investigations involved with this diagnosis.

Up to 40% (Moeller, 2007) of children with a hearing loss have additional health needs or developmental problems and 20% have more than 2 (Fortnum and Davis 1997, 1998). The complexity of referral pathways and the number of specialities and practitioners that may be involved, make medical guidelines essential in order to deliver well co-ordinated, quality service and to avoid any replication or unnecessary procedures (both from a patient safety point of view and financially). Accurate information and record keeping shared between parents and their medical contacts can lead to greater engagement, clearer understanding and enable informed decision making in the care pathway process.

Since the Queensland Healthy Hearing Statewide Screening programme commenced in 2004, children with hearing loss are now being identified in their first few months. Having identified these children, we need to have effective, efficient pathways for the management of their future health, development and family needs.

1.2 Aims of the medical guidelines for children born with permanent hearing loss

a. To ensure effective, efficient and accurate assessment, diagnosis and management of children born with permanent hearing loss.
b. To ensure consistency and equity of access to quality medical services for all children with PHL statewide.
c. To ensure that medical evaluation and diagnostic procedures are evidence based reflecting best practice.
d. To provide a basis for collection of accurate data for evaluation and reporting purposes.
1.3 Why investigate the causes of hearing loss?

- To answer the parents question ‘Why is my child deaf?’
- Identification of associated conditions, medical or developmental problems.
- The results of investigations can assist the health practitioners to make informed decisions about the most appropriate management plan in terms of frequency of follow up, cross referrals to other specialities, amplification needs, further investigations, educational programmes and family counselling etc as indicated.
- To give families advice about likely outcomes, progression of hearing loss, complications which may arise and prevention of complications eg for those children with cardiac conduction defects.
- To provide genetic information and offer counselling.
- Epidemiological information for research and planning, surveillance and prevention of hearing impairment.

Identifying the cause and extent of the hearing loss can be a long ongoing process and in 30-40% of cases, the aetiology remains unknown. (Parving, 1984, Maki-Torkko 2003)

The timing of investigations will depend on the family’s readiness to proceed with the investigations and how well the child can cooperate with the tests and other factors such as timing of anaesthetics. Investigation for causes of hearing loss may be delayed if the child has additional medical problems.

While we acknowledge the evidence supporting a sequential pattern of investigation,(Preciado, 2005) clinical reasoning needs to be applied to strike a balance between cost effectiveness and the least intrusive process of care for the child and their family.

Reasons for carrying out aetiological investigations early:

- CMV: to monitor development and progression of hearing loss and referral for the mother to an infectious diseases specialist.
- EVA: to educate parents with a view to minimising fluctuations/progression of hearing loss and to monitor thyroid function.
- Long QT: to minimise risk of cardiac complications
- Early genetic opinion on recurrence risks and syndromal diagnosis
- Some investigations could be carried out under natural sleep
- Early intervention for both the hearing loss and all the developmental needs of the child can be planned and managed.

“Clinical Pathways never replace clinical judgement. Care outlined in this pathway must be altered if it is not clinically appropriate for the individual patient”.
1.4 Definition of Terms

**Permanent hearing loss (PHL)** – permanent hearing loss, which includes both sensorineural hearing loss and permanent conductive hearing loss, and mixed hearing loss (sensorineural and conductive), but excludes temporary conductive hearing loss such as associated with otitis media with effusion (OME) or “glue ear”. For the purpose of this document PHL includes both bilateral and unilateral HL, and any degree of severity (mild, moderate, severe and profound). The document will most commonly be utilised for children with congenital PHL, but also is relevant to children with late-onset PHL such as that due to congenital CMV infection or genetic progressive hearing loss.

**PCHL** – permanent childhood hearing loss

1.5 Family Centre Care

Medical investigations including aetiology needs to be available to all children diagnosed with HL with full parent involvement.

“Parents must be given comprehensive and unbiased information about the medical investigation which is carried out to identify the cause of the hearing impairment and the diagnosis and treatment of any co-existing conditions, including both the benefits and the disadvantages, and be given every opportunity to further discuss their views and concerns with the doctor so that they can make an informed decision about whether they want their child to have these investigations and if so, the nature and timing........ Parents must be kept informed at every stage of the investigations and should have reasonable access to the doctor for further information and explanation as required.”

*Medical management of infants with significant congenital hearing loss identified through the national newborn hearing screening programme- Best Practice Guidelines (NHSP in conjunction with BAAP and BACADA)2004*

- **Dignity and respect** needs to be shown for the family and their part in the decision making process and their right to decline an investigation if they feel it is not right for them. Opportunities should be provided to revisit these decisions at a later stage, being mindful that the adjustment stage may influence readiness/ openness to investigations. There needs to be respect and understanding for the adjustment stages the families of children with this diagnosis will be going through.

- **Information sharing** - The parent booklet that forms part of the Personal Health Record contains information on the roles of the various professionals that may be involved after a diagnosis of hearing loss. It also contains details of the types of investigations and procedures that may be required as well as information on developmental milestones etc.

- **Participation** - Providing detailed information to parents enables them to make informed decisions around management planning of the hearing loss and future investigations. It gives them the rationale for the importance of early intervention for optimum outcomes in the development of their child’s speech and language, promoting the need for timely medical investigation.

- **Collaboration** - The personal health record provides opportunities for a collaborative approach between families and professionals with the family holding the child’s information and sharing it across sites. It relies on the clinician to be an active participant in maintaining this record.
2. Companion Documents

- Hearing Loss Management Summary
- Algorithm
- Summary sheet
- Poster
- Parent booklet
- Investigation results

Part of the Personal Health Record

<table>
<thead>
<tr>
<th>Resource</th>
<th>Application</th>
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| Medical Guidelines Forms  | **Hearing loss Management Summary**  
                          • Filed in the child’s medical chart when children are first diagnosed with hearing loss.  
                          • Investigation results and referrals to be entered by the initiator.  |
|                           | **Algorithm**  
                          • Used as a clinical guide and aide-memoire                                  |
|                           | **Summary Sheet**  
                          • Used as a clinical guide with greater detail and text form               |
| Medical Guidelines        | **Poster**  
                          • A flow chart of the medical assessment pathway                             |
| Parent information        | **Parent Booklet**  
                          • Information for parents on medical investigations, referrals and the roles of the different professionals that may be involved. Timelines, developmental stages etc.  |
| Investigation pages       | **Investigation pages**  
                          • Used to record information and test results. To be taken to all medical appointments and updated.  
                          • These pages and the booklet are additions to be added to the child’s Personal Held Record. They form part of the Possibilities and Pathways kit that is given to families of children with hearing loss (by QHLFSS or Audiology).  |

Medical Guidelines for the Assessment of Children with Permanent Hearing Loss (including unilateral loss)

Clinical Assessment

History
- pre-natal, birth history, post-natal, family history - don't forget 3 generation family tree, audiology results

Examination
- general, dysmorphology, head and neck, cardiac, neurological, developmental

Referrals

Audiology
- Family audiology testing (mother, father, siblings)

ENT
- All children ASAP

OHLFSF
- (family support facilitator)

Australian Hearing
- All children ASAP

Paediatrician
- All children ASAP then 4-6 months, 12, 18, 24 months

Genetics
- All children 6-12 months

Ophthalmologist
- All children approximately 6 months
- If not walking at 18 months and aetiology unknown, review for Usher's Syndrome
- If no known aetiology by 6 yrs, ERG to assess for Usher's Syndrome

Speech Pathology

Early Intervention
- All children by 6 months

Investigations

Blood
- FBC
- U&Es
- Thyroid function
- CMV, rubella, toxoplasmosis IgG3 and IgM, syphilis serology
- CMV PCR from Newborn Screening Card (DSS)
- Chromosomes if developmental delay or dysmorphic features
- Connexin 26 common mutation screen unless clear diagnosis of syndrome associated with HL

Urine
- All children: protein - microscopy
- CMV PCR (if DSS positive)
- Urine metabolic screen if developmental delay or failure to thrive

Radiology
- MRI inner ear and internal auditory meatus, brain screen
- children with severe bilateral SNHL or greater
- children with moderate unilateral or greater
- progressive SNHL
- auditory neuropathy
- structural renal abnormalities
- congenital CMV infection (or as indicated)
- CT petrous bone, brain scan
- children with severe bilateral SNHL or greater
- progressive unilateral or bilateral SNHL
- structural renal abnormalities (or as indicated)
- Renal Ultrasound
- children with suspected branchio-oto-renal syndrome: auricular pits, branchial anomalies or cysts
- multiple or multi system abnormalities
- family history of structural renal problems
- Mondini defect/in imaging

ECG (+/- holter tape)
- Children with severe bilateral SNHL or greater
- may need repeating when child is older
- interpretation by Paediatric Cardiologist
- if QT interval > expected for age, refer to Paediatric Cardiologist
- if QT interval > expected for age, refer other family members for ECG
4. **Guidelines**

4.1 **Audiology**

<table>
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<tr>
<td>• Confirm permanent sensorineural or conductive hearing loss</td>
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<td>• Progress referral to ENT</td>
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<tr>
<td>• Send hearing assessments results to relevant professionals</td>
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<tr>
<td>• Progress referral of parents and siblings for hearing assessment</td>
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On the completion of the diagnostic process, the diagnostic audiologist is responsible for:

- Progressing a referral to an Ear Nose and Throat Specialist (ENT)
- Sending the assessment results to relevant professionals, including:
  - Ear Nose and Throat Specialist
  - Queensland Hearing Loss Family Support Facilitator
  - General Practitioner/Family Doctor
  - Paediatrician present at child’s birth
  - Paediatrician child will visit for post-diagnostic assessment if different from original Paediatrician present at the child’s birth
  - Neonatologist (if applicable)
  - Australian Hearing
  - Habilitation centre/s once parents have made these decisions
  - Childhood Hearing Clinic (Mater, RCH, Townsville)
  - Other relevant professionals

- Facilitating parent and sibling hearing assessment
  
  Except in circumstances it can be confidently ruled out that the child’s hearing loss was acquired, it is recommended that hearing assessment of both the infant’s parents and all of the infant’s siblings should be undertaken even where newborn hearing screen was negative.

If there is any doubt about whether or not the child’s hearing loss was acquired, family hearing assessment should be undertaken.

In order to reduce the travel demands on families living in regional, rural or remote areas, parent and sibling hearing assessment may be able to be undertaken at a location other than the original diagnosing centre (e.g. other Queensland Health facility, private audiologist, Australia Hearing). The diagnosing audiologist will facilitate the referral process to alternate services where this is required.
4.2 Queensland Hearing Loss Family Support Service- QHLFSS

QHLFSS offer support to families following newborn screening ‘double refer’ results. Typically Family Support Facilitators (FSF) contact families from the time of audiology confirmation of diagnosis of hearing loss. If a parent chooses to include a FSF in their ongoing support, the FSF will be available to assist them to co-ordinate their access to the post-diagnostic aetiological, developmental and health assessment processes.

Examples of the roles FSF may fulfil, in collaboration with other professionals and services, include assisting families:

- Through the delivery of information and guidance to support parents in planning and making informed decisions
- Through the delivery of information that is timely and supports understanding.
- Through promoting adjustment and positive emotional and health outcomes for families and children.
- Through support and advocacy, that includes negotiating and assisting co-ordinated approaches to the provision of services.
- To explore early intervention and communication options that are appropriate to needs.
- To understand the outcomes and implications of the integrated picture of the different assessments undertaken.

4.3 Medical Evaluation

This may be carried out by the ENT and/or the Paediatrician

All children
- Child and family history, physical examination
- Order primary blood tests
- Order CMV investigation from Guthrie Cards (DBS)

Child and Family History

- **Antenatal history**
  - immunization for rubella- IgG>10 IU/ml
  - screening for syphilis
  - drug use
  - alcohol use
  - spontaneous abortions
  - exposure to ototoxic medications ✰ risk factors for PHL (aminoglycosides, platinum derivatives, loop diuretics, quinine derivatives, salicylates)

- in-utero infection ✰ risk factor for PHL- toxoplasmosis, CMV, herpes, rubella, syphilis
- medications taken

(Updated: 24/05/2012, Review November 2013)
- antenatal ultrasound
- other interventions
- other complications

• Birth
  - weeks gestation (X/40)
  - spontaneous/induced/reason for induction
  - mode
  - duration
  - delivery complications
  - interventions
  - instrumentation used
  - post partum complications (mother)
  - post partum interventions (mother)

• Post-natal history
  - Apgar 1 minute, Apgar 5 minutes
  - Birth Weight
  - Length, head circumference
  - Complications on delivery
  - Interventions on delivery
  - Post-partum complications (mother)
  - Post-partum interventions (mother)
  - NICU admission and duration
  - SCN admission and duration

  - Respiratory status at birth/ resuscitation
    ☢ risk factor for PHL
    Severe asphyxia
    (convulsions/HIE)

  - Ventilation- number of days
    ☢ risk factor for PHL
    ≥ 5 days (IPPV/PAP)

  - Hypoxic/anoxic episodes in post-natal period
    ☢ risk factor for PHL

  - Maximum serum bilirubin level
    ☢ risk factor for PHL
    Term: >450µmol/l,
    Preterm: >340µmol/l

  - Phototherapy
  - Abnormalities noted at birth
    ☢ risk factor for PHL
    Craniofacial anomalies
    (excluding cleft lip and skin tags)

  - Diagnosed/ suspected syndrome
    ☢ risk factor for PHL:
    syndrome

  - Other investigations
  - Immunizations
  - Early feeding: breast, bottle, tube fed, issues?

• History of exposure to risk factors ☢
  - Bacterial meningitis (proven/suspected)
  - Head injuries
  - Proven infections
toxoplasmosis, CMV, herpes, rubella, syphilis
  - Suspected infections
  toxoplasmosis, CMV, herpes, rubella, syphilis
- Antibiotics
- Ototoxic medications
- Other medications

• **Three generation family tree**
  Family history of hearing loss or risk factors associated with hearing loss in first and second degree relatives.
  - Grandparents
  - Parents
  - Siblings
  
  Risk factors associated with hearing loss in first and second degree relatives
  - structural renal problems (Alport Syndrome), goitre in Pendred’s syndrome

• **Family audiograms**
  A hearing assessment is recommended for first degree relatives. Previously unsuspected audiometric abnormalities may be identified. In families were there is a history of hearing loss, the degree of hearing loss may be variable within the same condition (eg, Waardenburg syndrome). Siblings need to be retested even if their newborn screen was negative.

**Physical Examination**

Including:

- head
- face
- neck
- eyes
- ears
- hands
- feet

**Investigations**

• **Order Primary Blood Tests**
  - Full blood count
  - Electrolytes
  - Serum creatinine
  - Glucose

• **CMV PCR from newborn screening card**

• **Order CMV, rubella, toxoplasma and syphilis serology**

  CMV
  - Detection of CMV by PCR in saliva, blood or urine or positive IgM prior to 3 weeks of age suggests congenital CMV infection.
  - After 3 weeks of age, viral detection or IgM could be due to either congenital or postnatally acquired infection. Postnatally acquired infection would not usually be associated with adverse outcome.
  - Detection of CMV by PCR from newborn screening card suggests congenital CMV infection. If the PCR is positive, mother should be...
referred to an Infectious Diseases Specialist for testing and if positive, monitoring and counselling for the risks involved in future pregnancy until tests come back negative.

Rubella
- IgM positivity prior to 6 months suggests congenital rubella infection
- Falling or absent IgG after 9 months of age suggests absence of infection
- Between 6 months and 3 years, absence of rubella specific T cell response (heparinised blood) excludes rubella.

Toxoplasmosis
- IgM or blood/CSF PCR positivity prior to 6 months of age suggests congenital infection.
- IgG titre significantly higher than mother’s IgG titre suggests congenital infection.
- IgG positivity after 6 months of age suggests congenital infection.

Syphilis
- Positive serology in baby warrants further investigation eg IgM testing and discussion with ID specialist

- Genetics
  - Chromosomes - if there is developmental delay or dysmorphic features
  - Connexin 26- the 30delG (35delG) mutation screen - unless clear diagnosis of a syndrome associated with hearing loss. Expanded genetic testing by Genetic Health Queensland as needed.

- Urine
  - All children –protein, microscopy
  - CMV PCR if DBS positive - only indicates congenital CMV if <3 weeks but may add to the total picture.
  - Urine metabolic screen if there is developmental delay or failure to thrive

4.4

**ENT**

- **All children**
  - Physical examination of head, neck
  - Refer to Australian Hearing, Paediatrician, Ophthalmologist, Clinical Geneticist*
  - Order primary blood tests
  - Discuss MRI and CT with all parents
  - Order CMV investigation from Guthrie Cards

- **Where indicated**
  - Refer for MRI
  - Refer for CT
  - Refer for renal ultrasound
  - Refer for ECG

*except if syndromic or non-syndromic genetic aetiology can be confidently ruled out

(Updated: 24/05/2012, Review November 2013)
MRI Imaging and CT
Discuss the purpose of MRI (Magnetic Resonance imaging) and CT (Computerised Tomography) with ALL families. CT and MR imaging are known to provide complementary information rather than one or the other procedure providing complete information alone.

MRI showing the soft tissue- brain, VIIth and VIIIth nerves and membranous labyrinth including the endolymphatic sac.

Recommend MRI inner ear and internal auditory meatus (IAMs), brain scan imaging for all children with:

- severe bilateral sensorineural hearing loss or greater
- moderate unilateral SNHL or greater
- progressive unilateral or bilateral hearing loss
- auditory neuropathy
- structural renal abnormalities (or as indicated)
- congenital CMV infection (or as indicated)

To ensure high quality images, a GA is required for all children up to approximately 4 years of age, although an early referral for feed and wrap MRI scan for young infants (approximately 4 months of age) should be considered if indicated, as the need for GA may be avoided.

Unless clinically indicated (e.g. assessment for early cochlear implantation), it is recommended that imaging does not occur on children under 6 months in order to minimise GA risks and radiation exposure.
CT scan involving radiation and showing the bony structures including middle ear ossicles.

Recommend CT petrous temporal bone, brain scan imaging for all children with:

- severe bilateral sensorineural hearing loss or greater
- progressive unilateral or bilateral hearing loss
- structural renal abnormalities (or as indicated)

CT scan is no longer a first line of investigation and should only be considered after consultation with the ENT in the cochlear implant team.

CT and MR imaging are only able to be completed with infants under GA at:

Royal Children’s Hospital
Mater Children’s Hospital
Mater Private Hospital
The Townsville hospital
Gold Coast hospital
Renal Ultrasound
- Children with suspected branchio-oto-renal syndrome—where pre-auricular pits, branchial sinuses or cysts are present
- Multiple or multi system abnormalities
- Family history of structural renal problems
- Mondini dysplasia on imaging

Electrocardiography ECG (with holter tape)
- Children with severe bilateral sensorineural hearing loss or greater
- May need repeating when the child is older
- ECG needs to be interpreted by a Paediatric Cardiologist
- If QT interval> expected for age, refer to a Paediatric Cardiologist
- If QT interval> expected for age, refer other family members for an ECG

Jervell and Lange-Nielsen syndrome (JLNS) is a rare autosomal recessive syndrome with long QT interval and sensorineural hearing loss due to the homozygosity for mutations in the KVLQT1 or KCNE1 genes.

Referrals
- Hearing Aid Clearance and referral to Australian Hearing (where appropriate)

- Referral to Paediatrician
  Childhood Hearing Clinics (CHC) now operate at the Royal Children’s Hospital, Mater Children’s Hospital and the Townsville Hospital. These are multidisciplinary clinics where families can see a paediatrician, QHLFSS, speech pathology, Australian Hearing and a parent mentor. The child receives a full diagnostic and developmental assessment in a series of three appointments after which they are referred to the paediatric hearing clinics or regional paediatricians if further paediatric follow up is needed.

- Referral to a Clinical Geneticist – (Genetic Health Queensland)
  -request for assessment at 6 months.

- Referral to Ophthalmologist
  -request for assessment at 6 -12 months. Assessment of visual acuity and fundoscopy.

40% of children with sensorineural hearing loss have additional needs and/or ophthalmic conditions. Problems may be non-specific to any underlying condition such as a squint or refractive errors or other findings on examination may help to clarify diagnosis such as Usher Syndrome, CHARGE, congenital CMV or rubella.
4.5 The assessment by the Paediatrician will include:

- Child and family history
- Examination
- Developmental history
- Examination
  - Growth parameters
  - Development
  - Neurological assessment
  - Cardiovascular system
  - Head
  - Face
  - Neck
  - Skin
  - Chest
  - Abdomen
  - Limbs
  - Nails
- Developmental Assessment
- Order Chromosome test when:
  - Developmental delay evident
  - Dysmorphic features evident
- Order metabolic screen when:
  - Child suspected of failure to thrive
  - Developmental delay noted
- Order MRI if appropriate

Paediatrician

All children
- Child and family history
- Physical, developmental, health examination
- Blood, urine and CMV if not already requested

Order chromosome test when:
- Developmental delay evident
- Dysmorphic features evident

Order metabolic screen when:
- Child suspected of failure to thrive
- Developmental delay noted

Where indicated:
- Order chromosome test (parent consent required)
- Order metabolic screen

request form for CMV with parent consent - download from

Where indicated:
- Order chromosome test (parent consent required)
- Order metabolic screen

If not already done by ENT

Where indicated:
- Order chromosome test (parent consent required)
- Order metabolic screen

If not already done by ENT
4.6

Clinical Geneticist- Genetic Health Queensland (GHQ)

- Assessment at 6 months

Not required if syndromic or non-syndromic genetic aetiology can be confidently ruled out

To ensure families have the most timely access to information relevant to their future family planning, it is recommended that assessment by a Clinical Geneticist occurs when the child is approximately 6 months of age, and preferably no later than 9 months.

GHQ provides services in Brisbane, Gold Coast, Nambour, Toowoomba, Ipswich, Bundaberg, Hervey Bay, Mackay, Mt Isa, Rockhampton, Townsville and Cairns.

The assessment by the Clinical Geneticist may include:

- Family History
- Physical Examination
- Examination and Measurement of craniofacial region, +/- W index
- Assessment as indicated for:

  - **Connexin 26** – common mutations, followed by full sequencing if necessary
    - not indicated in context of clear syndromic aetiology
    - not indicated where 3 generation family history indicates obvious dominant inheritance
    - if one mutation found on Connexin 26, then Connexin 30 should be completed

  - **Connexin 30** – common mutations, followed by full sequencing if necessary
    - not indicated in context of clear syndromic aetiology
    - if one mutation found on Connexin 26, then Connexin 30 should be completed

  - **Pendred Syndrome** – genetic testing to identify the Pendrin gene PDS SLC26A4
    - all children with large vestibular aqueduct syndrome (LVAS)
    - all children with cochlear dysplasia
    - all children with hypothyroidism
    - not indicated in context of clear syndromic aetiology
    - not indicated where 3 generation family history indicates obvious dominant inheritance

  - **A1555G (mitochondrial DNA mutation)**
    - All children with exposure to aminoglycosides
    - All children with a pedigree indicative of mitochondrial inheritance/maternal inheritance

This is our current understanding of genetics testing but as it is an area of rapid change, it will be under regular review.

(Updated: 24/05/2012, Review November 2013)
o Biochemical genetic assessment
  - as determined by Geneticist

o Chromosome assessment
  - all children with developmental delay
  - all children with dysmorphic features

For more detailed information:
http://hereditaryhearingloss.org an up-to-date overview of the genetics of hereditary hearing impairment for researchers and clinicians working in the field. Maintained by the universities of Antwerp and Iowa.

4.7 Ophthalmologist

- Assessment at 6 months
- Assessment at 18 months
- ERG at 6 years if cause of hearing loss not identified through other means

Approximately 40% of children with severe to profound hearing loss will also have ocular abnormalities. The majority of these will be refractive errors but other ocular pathologies may contribute to aetiology and confirm a syndrome (for example, Usher) or suggest a congenital infection (for example, congenital rubella).

Initial Assessment – 6 months

- It is recommended that all children are referred for assessment by an ophthalmologist for indicators of Usher Syndrome Type 1. Children not walking by 18/12 with unknown aetiology of PHL should be reviewed for Usher Syndrome.
- Children who show signs of night blindness should be assessed by an Ophthalmologist for indicators of Usher Syndrome
- Children who have had no known aetiology for their hearing loss identified by 6 years of age should be assessed using electroretinography to identify indicators of Usher Syndrome.
4.8 CT and MRI

- MRIImaging and CT
  - Recommend MRI imaging for all children with:
    - Severe bilateral SNHL or greater
    - Moderate unilateral SNHL or greater
    - Progressive SNHL
    - Auditory neuropathy
    - Structural renal abnormalities
    - Congenital CMV infection or as indicated
  - To ensure high quality images, a GA is required for all children up to approximately 4 years of age, although an early referral for feed and wrap MRI scan for young infants (approximately 4 months of age) should be considered if indicated as the need for GA may be avoided.
  - Unless clinically indicated (e.g. assessment for early cochlear implantation), it is recommended that imaging does not occur on children under 6 months in order to minimise GA risks and radiation exposure.
  - MRI imaging and CT are known to provide complementary information rather than one or the other procedure providing complete information alone. However, for a child who is known to have been totally deaf from birth, it is recommended that MRI is completed first to determine the presence or absence of the cochlear nerve and identification of other congenital abnormalities of the inner ear.
  - Recommend CT petrous bone, brain scan for all children with:
    - Severe bilateral sensorineural hearing loss or greater
    - Progressive unilateral or bilateral hearing loss
    - Structural renal abnormalities (or as indicated)
  - MRI imaging and CT are only able to be completed with infants under GA at:
    - Royal Children’s Hospital
    - Mater Children’s Hospital
    - Mater Private Hospital
    - The Townsville Hospital
    - Gold Coast Hospital

When children require both MRI and CT, consideration should be given to performing both scans under one GA.
• Infants with severe bilateral sensorineural hearing loss or greater are to be referred for an ECG
• The ECG is to be read by a paediatric cardiologist
• Where ECG results indicate that an infant’s QT interval is greater than expected for their age, referral to a Cardiologist for further management is recommended.

Renal Ultrasound and Nephrology Referral

• Renal ultrasound is recommended for all children with:
  o Suspected branchio-oto-renal syndrome (i.e. pre-auricular pits, branchial sinuses or cysts)
  o Multiple or multisystem abnormalities
  o Family history of structural renal problems
  o Mondini dysplasia on imaging

4.9 Neurology

• The need for referral to a Neurologist will be determined by a relevant medical practitioner (e.g. Paediatrician, ENT or Geneticist)

Other issues under consideration:-

• Urine testing for CMV in all infants with a double refer on newborn screening
• Use of the genetic chip
• Testing for mucopolysaccharidosis disorders when metabolic testing
• Vestibular testing
• Radiology – MRI and CT
Bibliography:

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http://hereditaryhearingloss.org

(Updated: 24/05/2012, Review November 2013)
Members of the Advisory Group
Medical guidelines for children born with a permanent Hearing loss

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Dr Susan Moloney  Paediatrician, Gold Coast Hospital
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Dr David Bell-Allen  ENT
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Lia Traves  Audiology,CI, RCH
Tim Wood  Team Leader, QHLFSS
Sreedevi Aithal  Director of Audiology, Townsville
Nuala Beahan  Senior Audiologist, Healthy Hearing
Dr Alan Sive  Paediatrician, Townsville
Heather Price  FSF, Townsville
John Gavranich  Director of Paediatrics
Katrina Roberts  Healthy Hearing, Townsville
Julie McGaughran  Genetic Health Queensland
Dr Anne Kynaston  Paediatrician
Dr Brian Morris  Director of Paediatrics
Richard Heazlewood  Pediatric Outreach
Luke Jardine  Neonatologist
Rachel Susman  Clinical Geneticist

(Updated: 24/05/2012, Review November 2013)
APPENDIX 1

Healthy Hearing Program Summary

Healthy Hearing Mission Statement

The Healthy Hearing program aims to improve health outcomes for Queensland children through the earliest possible detection and management of permanent childhood hearing loss. Newborn hearing screening is the first stage of a comprehensive approach to communication development which includes further assessment and early intervention. The program aims to systematically monitor its performance and be alert and responsive to emerging evidence in this field.

Background

The Healthy Hearing Program aims to detect permanent hearing loss (PHL) through newborn hearing screening for all Queensland babies before they reach three months of age, as well as ensuring appropriate diagnosis and early intervention for babies found to have a hearing loss. International evidence suggests that early detection of a hearing loss and commencement of early intervention through hearing aid provision and communication habilitation by the age of 6 months may be critical for speech and language development and can reduce the need for ongoing special education.

Children can have their hearing augmented by hearing aids from birth and/or with cochlear implants at around 12 months (although some children are implanted earlier). Access to sound as soon as possible is crucial to a child’s communication development.

Over 60,000 babies are born in Queensland each year. Approximately 1 to 2 per 1000 babies born will have a bilateral moderate or greater hearing loss. The program also detects hearing loss in infants with milder degrees of loss or unilateral losses and offers referral for these children.

Some children pass the hearing screen at birth, but have risk factors for progressive or delayed onset hearing loss. These children are identified at the time of screening and offered follow-up audiology assessment before 12 months of age. It is important for parents and medical staff to monitor a child’s hearing, as a pass at screening at birth is not a pass for life. Hearing can change over time.

Objectives

The objectives of the Healthy Hearing Program include:

- Optimise early detection of PHL in neonates by providing newborn hearing screening before 3 months of age.
- Provide diagnostic audiology assessment to relevant neonates by 6 months of age.
• Ensure equitable access to the Healthy Hearing Program for all neonates irrespective of social, economic, cultural or geographic circumstances.
• Ensure the Healthy Hearing Program is standardised and provided using multidisciplinary, evidence based screening, diagnostic, treatment and habilitation protocols.

Healthy Hearing Benchmarks

The Healthy Hearing Program has established the following targets:

<table>
<thead>
<tr>
<th>Screening Rates</th>
<th>All babies born in Queensland birthing facilities (public/private) offered a hearing screen. 95% of eligible babies will have their screen completed by 3 months of corrected age.</th>
</tr>
</thead>
</table>
| Referral Rates  | <4% of babies screened referred for diagnostic audiological testing;  
• Babies with a bilateral Refer result offered assessment by Audiology within 2 weeks.  
• Babies with unilateral Refer result offered assessment by Audiology within 6 weeks.  
(Diagnostic assessment for above babies will be completed by 6 months corrected age.)  
95% of babies referred for diagnostic audiological testing attend Audiology.  
<4% of babies screened, identified with risk factors for progressive hearing loss;  
• Babies identified with risk factors will be reviewed by their 1st birthday. |
| Intervention     | Average age for intervention/fitting of hearing aids will be 6 months corrected age.  
Average age for intervention/fitting of cochlear implants will be 12 months correct age. |

A comprehensive set of national quality standards and performance indicators for newborn hearing screening is being developed and Queensland will comply with those when they are finalised.

Hearing Screening Protocol

Hearing screening ideally takes place prior to the baby’s discharge from hospital, to optimise ‘capture rates’. A brochure titled “Your baby’s free hearing screen” is provided to parents and written consent is obtained from the parent/s prior to the screen. A nurse trained in hearing screening carries out the screen when the baby is quiet or asleep.

The screening process uses Automated Auditory Brainstem Response (AABR) equipment. The equipment used by Queensland Health is the Natus ALGO 3 (trolley mounted) and the ALGO 3i (hand held). The screen is non-invasive.
and easy to perform. Several sensor pads are placed on the baby’s head and soft clicking sounds are played into the baby’s ear through earphones. The sensor pads record the baby’s responses to the sounds.

The baby must be more than 34 weeks gestation to be screened. If a baby receives a ‘refer result’ on either or both ears a second screen of both ears is conducted at a later time to confirm the result. If the baby receives a ‘refer result’ on either or both ears on the second screen the baby is referred for diagnostic audiology assessment.

Statewide Diagnostic Audiology Protocols have been developed and are used throughout the state when a child referred from the Healthy Hearing Program is assessed.

A diagram of the hearing screening protocol is attached at Appendix 1.


**Recording the Results**
The screening equipment automatically records the results of each screen and the data is imported into a statewide database for tracking and further analysis at a district or state level. Any risk factors for progressive or delayed onset hearing loss, are identified. These risk factors, along with screening results are also recorded in the baby’s medical chart and Personal Health Record.

**Newborn Screening Hospitals**

<table>
<thead>
<tr>
<th>Public Birthing Hospitals</th>
<th>Private Hospitals</th>
</tr>
</thead>
<tbody>
<tr>
<td>Royal Brisbane &amp; Women’s Hospital</td>
<td>St Andrews Private, Ipswich</td>
</tr>
<tr>
<td>Townsville Hospital</td>
<td>Cairns Private</td>
</tr>
<tr>
<td>Mater Mother’s Hospital</td>
<td>Mater Private, Redland</td>
</tr>
<tr>
<td>Logan Hospital</td>
<td>Mater Private, Mackay</td>
</tr>
<tr>
<td>Cairns Hospital</td>
<td>Mater Private, Gladstone</td>
</tr>
<tr>
<td>Nambour Hospital</td>
<td>Sunnybank Private</td>
</tr>
<tr>
<td>Ipswich Hospital</td>
<td>Pindara Private</td>
</tr>
<tr>
<td>Gold Coast Hospital</td>
<td>John Flynn Private</td>
</tr>
<tr>
<td>Redcliffe Hospital</td>
<td>Mater Private, Rockhampton</td>
</tr>
<tr>
<td>Thursday Island Hospital</td>
<td>St Vincent’s, Toowoomba</td>
</tr>
<tr>
<td>Ingham Hospital</td>
<td></td>
</tr>
</tbody>
</table>

**Babies Born Outside Birthing Hospitals**
The Healthy Hearing program receives Client Directory data for all babies born in every Queensland public hospital each day. This data allows babies to be identified who are not born at a birthing hospital, and these babies are referred on the closest screening hospital to arrange the screen with the parent/s. The Healthy Hearing program also works closely with homebirth providers to ensure they
encourage parents to contact their local screening site to arrange a hearing screen.

**Hearing Screen Declined by Parents**
A parent’s decision to decline the hearing screen is respected. However, it is appropriate to ascertain reasons for declining, and to correct any misunderstandings regarding the hearing screening process and/or risks to the baby. It is also important that parents be fully informed of the potential implications should their baby have an undetected hearing loss.

If a screen is declined, a letter is sent to the baby’s medical practitioner advising of this, and advising of any risk factors for a progressive/delayed onset hearing loss (Appendix 2).

Medical practitioners and other health staff involved with the infant can advise parents that they may return for the screen before the baby is 3 months of age on an outpatient basis should they change their mind. Babies may be able to be screened up to 6 months. After that time an audiology appointment would be required. Hospital contact phone number and details should be provided on the ‘Your baby’s free hearing screen’ brochure.

**Support for Parents**
The Queensland Hearing Loss Family Support Facilitators Service (QHLFSF) is a state-wide service established to provide family-centred support and counselling to families of children diagnosed with a PHL. This service assists parents access a wide range of different professionals and services available to children with a PHL. The service is available to all children diagnosed with a PHL from birth through to completion of Grade 1. This includes children who access either public or private medical and habilitation services. QHLFSF Contact Information: Phone: (07) 3250 8555
Appendix 1

Healthy Hearing Program Screening Pathway

- Medically stable
- Older than 34 wks
- Near to discharge
- Parental consent
- Inpatient / Outpatient

Eligible to screen

AABR1 1st Screen

PASS both ears
No risk factors identified

Discharge
Parental & GP monitoring

PASS both ears
Risk factors identified

REFER to Audiology for surveillance assessment before baby's 1st birthday or earlier at discretion of Audiologist
Parental & GP monitoring

REFER one or both ears

AABR2 2nd Screen

PASS both ears
No risk factors identified

Discharge
Parental & GP monitoring
Discuss Otitis Media risk

PASS both ears
Risk factors identified

REFER to Audiology for surveillance assessment before baby's 1st birthday, or earlier at the Audiologist's discretion
Parental & GP monitoring

REFER one or both ears

Flip Flop Result
Unexpected reversal of previous pass result

AABR3 3rd Screen

PASS both ears
No risk factors identified

Discharge
Parental & GP monitoring
Discuss Otitis Media risk

PASS both ears
Risk factors identified

REFER to Audiology for surveillance assessment before baby's 1st birthday, or earlier at the Audiologist's discretion
Parental & GP monitoring

REFER result in same ear on two occasions

Refer to Audiology for immediate diagnostic assessment
- Appointment within 2 weeks - Bilateral refer
- Appointment within 6 weeks - Unilateral refer

Refer to Family Support Facilitator Service

Ineligible to screen

Exclusion to screening
Refer to Audiology for diagnostic assessment
Hearing loss risk factors include:

- Syndromes associated with hearing loss (eg. Downs, FAS) (See protocols for a complete list)
- Prolonged ventilation ≥ 5 days (IPPV / CPAP)
- Bacterial meningitis (confirmed / suspected)
- Severe asphyxia at birth (convulsions / HIE / PPHN)
- Craniofacial anomalies eg. cleft palates (excluding cleft lips and skin tags)
- Hyperbilirubinemia levels ≥450μmol/l (Term) or ≥340μmol/l (Preterm) Max SBR level
- Proven / suspected congenital infection of the baby (Toxoplasmosis, Rubella, CMV, Herpes, Syphilis)
- Professional concern
| Hearing Status | Referrals | Radiology | Urine Exam | Blood Tests | Family History (including Unilateral) | Other Genetic Tests | Chromosomes | Other Tests | Full Blood Count | Thyroid Function Test | Vitamin D, Calcium & Phosphorus screen | Family History (parents, siblings) | Family History (grandparents) | Family History (other relatives) |
|----------------|-----------|-----------|------------|-------------|--------------------------------------|--------------------|-------------|--------------|----------------|---------------------|----------------------------------|--------------------------------|--------------------------------|--------------------------------|--------------------------------|
| Cochlear Implant | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** |
| Speech Pathology | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** |
| Audiology | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** |
| Otolaryngology | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** |
| Genetics | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** |
| Ophthalmology | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** |
| Ent | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** |
| ECG | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** |
| CT of petrous temporal bones | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** |
| MRI inner ear and Auditory Brainstem | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** |
| Metabolic Screen | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** |
| Cytokine 26 | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** |
| **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** | **PD** |

**PD** = performed during the initial assessment.

**For all children**
APPENDIX 3

MEDICAL GUIDELINES FOR THE ASSESSMENT OF CHILDREN WITH PERMANENT HEARING LOSS –Summary

History – prenatal, birth, post-natal, family history - don’t forget 3 generation family tree!

Audiology results

Examination

**Audiology, Family audiology testing** (mother, father, siblings)

**QHLSF**

**Australian Hearing** – all children ASAP

**ENT** – all children ASAP

**Paediatrician** - all children ASAP then approx 4 – 6 mo, 12mo, 18mo, 24mo of age

**Genetics** – all children 6-12 months

**Ophthalmologist**

- all children @ approx 6mo
- if not walking @ 18mo and aetiology unknown, review for Usher’s Syndrome
- if no known aetiology by 6 years ERG to assess for Usher’s Syndrome

**Blood tests** All children

- FBC
- U&Es
- thyroid function
- CMV, rubella, and toxoplasmosis IgG and IgM, syphilis serology
- CMV from Newborn Screening Card

Chromosomes if developmental delay or dysmorphic features

Connexin 26 common mutation screen unless clear diagnosis of syndrome ass. with a HL

Expanded genetic testing by Genetic Health Queensland as needed

**Urine**

All children – protein, microscopy

Consider CMV PCR

Urine metabolic screen if developmental delay or failure to thrive

**Radiology**

**MRI inner ear and internal auditory meatus, brain screen**

- children with severe bilateral SNHL or greater
- children with moderate unilateral SNHL or greater
- children with progressive unilateral or bilateral SNHL
- auditory neuropathy
- structural renal abnormalities (or as indicated)
- congenital CMV infection (or as indicated)

**CT petrous temporal bone, brain screen**

- children with severe bilateral SNHL or greater
- progressive unilateral or bilateral SNHL
- structural renal abnormalities (or as indicated)

**Renal Ultrasound**

- children with suspected branchio-oto-renal syndrome: pre-auricular pits, branchial sinuses or cysts
- multiple or multi system abnormalities
- family history of structural renal problems
- Mondini defect on imaging

**ECG with holter tape**

- children with severe bilateral SNHL or greater

- may need repeating when child is older
- interpretation by Paediatric Cardiologist
- if QT interval > expected for age, refer to Paediatric Cardiologist
- if QT interval > expected for age, refer other family members for ECG’s

(Updated: 24/05/2012, Review November 2013)
APPENDIX 5

Parent handbook for Personal Health Record (Red Book)

The parent hand book is given to families as part of the Possibilities and Pathways package by the audiologist or support facilitator on receiving the diagnosis of hearing loss. It outlines the medical team that will be involved, their roles and details investigations that they may order. The booklet also has information on hearing development and early communication.

The book is intended to fit into the front pocket of the Personal Health Record (the Red Book) and comes with a set of loose sheets that fit into the clip side where health practitioners can enter investigations and test results pertaining to the hearing loss. Families should be encouraged to bring this book to all appointments.