Childhood Hearing Clinic – causes of congenital hearing loss
Audit of results of investigations

Dr Karen Liddle - 20th May 2017
9th Australasian Newborn Screening Conference
Childhood Hearing Clinic

• Multidisciplinary clinic for babies with permanent hearing loss detected on the newborn screen
• Operating since October 2011
• Brisbane and Townsville
• 2 Brisbane sites merged to Lady Cilento Children’s Hospital in November 2014
• Coordinator (nurse), Speech Pathology, Australian Hearing, Paediatrician, Audiologist case conferences
• Parent mentor and QHLFSS family support facilitator as required
• Liaise with ENT
Aims of the CHC:

1. To provide a full medical assessment and other services within the first 6 months.

2. Provides initial medical investigations, developmental assessment, hearing aid clearance and early amplification, and opportunities for early intervention from allied health and other external agencies as well as referral to other specialists as required.
Aims of this Audit

• To describe and understand our caseload
• To see how our caseload compares with other cohorts worldwide
• To inform better data collection
Scope of this audit

- Brisbane clinic: referrals from 2015 and 2016 births
- Permanent hearing loss – conductive and sensorineural
- 2 clinics per week
- N=298
- 6 excluded as hearing loss was transient
- Audit on n=292
Geographic location
Geographic location
Age at 1st CHC appointment

- Under 3 months: 159
- 3 to 6 Months: 97
- 6 months to 1 year: 26
- 1 to 2 years: 4
- Over 2 years: 2
- No Apt with CHC: 10
Time from diagnosis to CHC appointment

- Under 2 weeks: 64
- 2 weeks to 1 month: 93
- 1 to 2 months: 49
- 2 to 3 months: 11
- 3 to 6 months: 13
- 6 months to 1 year: 5
- More than 1 year: 1
- No Apt with CHC: 10
- No HL Outcome date: 17
- Seen before HL Outcome: 35
Medical assessment

• History, physical and developmental assessment
• Understand the cause of the hearing loss if possible
• Identify and manage comorbidities
• Referrals to ENT, ophthalmology, genetics, cardiology etc and family audiograms as required
• Order investigations: bloods, urine, imaging if families want this done
Aetiology of hearing loss

Figure 3: Environmental and genetic contributions to total congenital SNHL

Smith et al, Lancet, 365; 879-90; 2005
Aetiology, categorisation and coding

• Bilateral moderate or greater vs unilateral and mild bilateral
• Most common aetiology is unknown – around 40% in most studies
  • Unknown: 37.7%
  • Genetic  32%– non syndromic 29% and syndromic
  • Prenatal 11% - TORCH, alcohol, drugs, other – CMV 0.92 % in studies 1990-2002 – with DBS screening we currently see up to 10%
  • Perinatal 9.6% - kernicterus, asphyxia, prematurity, drugs
  • Postnatal 8.2% - meningitis, trauma, chemo, ECMO, measles, other
Imaging and genetic testing

- Reports of high yield from radiographic imaging of the inner ear (Mafong et al, Use of laboratory evaluation and radiologic imaging in the diagnostic evaluation of children with sensorineural hearing loss. Laryngoscope 2002; 112: 1-7)

98 children imaged – 0-18y Californian sample
40% showed abnormalities

| TABLE II. Radiologic Findings in Children With Sensorineural Hearing Loss. |
|-----------------------------|-----|-----|
| Finding                     | No. | Percent |
| Normal                      | 59  | 60.8  |
| Isolated CT abnormalities   |     |       |
| Large vestibular aqueduct   | 7   | 7.2   |
| Lateral semicircular canal dysplasia | 5   | 5.1   |
| Cochlear dysplasia          | 3   | 3.1   |
| Otic capsular lucency       | 1   | 1.0   |
| Small internal auditory canals | 1   | 1.0   |
| Hypoplastic cochlea         | 1   | 1.0   |
| Multiple CT abnormalities   |     |       |
| Cochlear dysplasia          | 7   | 7.2   |
| Large vestibular aqueduct   | 6   | 6.2   |
| Lateral semicircular canal dysplasia | 6   | 6.2   |
| MRI findings distinct from CT |     |       |
| Lipoma within quadrigeminal cistern | 1   | 1.0   |
| Fistulous connection        | 1   | 1.0   |
| Flattened posterior fossa floor | 1   | 1.0   |
| Butterfly vertebrae         | 1   | 1.0   |

*CT = computed tomography; MRI = magnetic resonance imaging.*
Imaging

- Review 0-18y Cincinatti 1993-2002
- 616 patients underwent imaging
- Abnormalities detected in 27.4%
Genetic testing

- From early 2000s connexin 26 mutation testing (GJB2) became available widely
- Initially 2 common mutations tested (30/35 del G); now the entire gene is sequenced
GJB2 (connexin 26) mutation testing

Fig 2. GJB2 mutation screen yield by SNHL severity category. Patients with biallelic nonsense mutations had a significantly higher likelihood of having severe to profound SNHL, whereas patients with at least one missense mutation had a significantly higher likelihood of having mild to moderate SNHL (*P = 0.006).
Unilateral Hearing Loss

• Much more likely to have structural abnormalities
• Much less likely to be genetic (non-syndromic) but still can be syndromic – Waardenburg, LVAS
Results of other cross-sectional studies

• LOCHI study n= 364
• Australian cohort
• 5 year cohort born 2002-2007 recruited over 2 years (2005-2007)

- N= 569 1997-2011 (14 years)
- Detected on newborn hearing screen
- NB 1/3 OME
Cohort from Antwerp

- N= 170 1998-2006 (8 years) of which 116 had permanent HL and 29 lost to follow up (87 tested)


• N=198 2005-2007
• Children with PHL referred to ENT clinic
• Older children – more acquired non-infectious
SSD and asymmetrical hearing loss cohort from Japan  Usami et al Etiology of single-sided deafness and asymmetrical hearing loss, Acta Oto-Laryngologica, 2017, epub ahead of print

• N = 210 congenital/early onset cases: 2006-2016
• ENT clinic

Figure 1. (A) The causes of SSD in congenital/early-onset cases. (B) The causes of AHL in congenital/early-onset cases.
Our data – All Hearing Losses

- N = 292  2015-2016
- Conductive and sensorineural
- Mild through to profound
Bilateral

• N=168  2015-2016
• Mild through to profound
• Conductive included
Unilateral

- N=124 2015-2016
- Includes conductive
- Mild to profound
Bilateral causes:

- Absent nerve: 1
- Structural - other: 3
- LVAS: 8
- Cleft palate: 7
- Microtia/atresia: 4

Total: 23, 14% of total

Unilateral causes:

- Absent/hypoplastic nerve: 17
- Structural - other: 3
- LVAS: 3
- Cleft palate: 0
- Microtia/atresia: 23

Total: 46, 37% of total

Bilateral structural causes:

- Absent nerve: 4%
- Structural - other: 13%
- LVAS: 35%
- Cleft palate: 31%
- Microtia/atresia: 17%

Unilateral structural causes:

- Absent/hypoplastic nerve: 37%
- Structural - other: 6%
- LVAS: 50%
- Cleft palate: 0%
- Microtia/atresia: 17%
Genetic causes – bilateral vs unilateral

**Bilateral n= 67  40% of total**

- Genetic - connexin: 18
- Genetic - familial: 15
- Genetic - syndromic: 23
- Genetic - other: 9
- Consanguinity: 2

**Unilateral n= 14  11% of total**

- Connexin: 2
- Familial: 3
- Syndromic: 9

**Bilateral Genetic Causes**
- Genetic - connexin: 27%
- Genetic - familial: 22%
- Genetic - syndromic: 34%
- Genetic - other: 14%
- Consanguinity: 3%

**Unilateral Genetic Causes**
- Connexin: 14%
- Familial: 22%
- Syndromic: 64%
## Syndromic causes unilateral vs bilateral

<table>
<thead>
<tr>
<th>Bilateral</th>
<th>Unilateral</th>
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<tbody>
<tr>
<td>Trisomy 21</td>
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<tr>
<td>Treacher Collins</td>
<td>Waardenburg</td>
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<tr>
<td>Pallister Killian</td>
<td>Goldenhar</td>
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<tr>
<td>Waardenburg Type 2</td>
<td>Dandy Walker</td>
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<tr>
<td>17 microdeletion syndrome</td>
<td>VACTERL</td>
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<tr>
<td>Cornelia de Lange</td>
<td>Isolated hemihyperplasia</td>
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<td>Zellweger</td>
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<tr>
<td>CHARGE syndrome</td>
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<td>Di George Syndrome</td>
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<tr>
<td>Branchio-otic syndrome</td>
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<td>Branchio oto renal syndrome</td>
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<tr>
<td>Goldenhar</td>
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<td>Cockayne</td>
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<td>Pierre Robin Syndrome</td>
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Summary

• Understanding this caseload
• Large numbers – larger series than what is in the literature; huge potential
• Different causes unilateral and bilateral
• Would be good to separate out conductive causes also and analyse by degree of hearing loss
• Consistent coding will enable better and more refined analysis – aiming to build this in to our clinical care
• Potential to collaborate with other states – more powerful data
• Huge benefit in linking CHC data into pathway data (QChild)
• Many research questions to answer!
Thank you