Should infants with a syndrome or craniofacial anomaly be screened for a hearing loss at birth?

Rachael Beswick
Background

**CONTROVERSY**
Significant debate regarding “off-label” use of screening equipment for children with a syndrome or CFA

**2012 REVIEW**
Risk factor review revealed that children with a syndrome or CFA had an increased risk of postnatal hearing loss

Development of Early Targeted Surveillance Category
What is ETS?

Children who have passed newborn hearing screening but have the risk factors of craniofacial anomaly and/or syndrome are classified as ETS and are seen at Audiology by 6 weeks of age.
### Syndrome associated with a hearing loss

<table>
<thead>
<tr>
<th>Title</th>
<th>Description</th>
<th>Hearing loss (HL)</th>
</tr>
</thead>
<tbody>
<tr>
<td>A</td>
<td><strong>Achondroplasia</strong></td>
<td>Dwarfism, skeletal ossification disorder</td>
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<tr>
<td></td>
<td><strong>Alpers-Schonberg Disease of Otosclerosis</strong></td>
<td>Brittle, thickened, chalky bones</td>
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<td></td>
<td><strong>Alström Syndrome</strong></td>
<td>Neuronal and cytopathic</td>
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<td></td>
<td><strong>Apert Syndrome</strong></td>
<td>Craniohyperostosis, malleus anomalies, middle ear involvement</td>
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<tr>
<td>B</td>
<td><strong>Apnea (errors during embryonic development)</strong></td>
<td>Complete absence of inner ear &amp; auditory nerve</td>
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<td></td>
<td><strong>Arnold-Chiari malformation</strong></td>
<td>Absence of development of the brain (in the) cochlear</td>
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<td></td>
<td><strong>Basal Ganglia (BGR)</strong></td>
<td>Absence of the Nucleus of the thalamus</td>
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<td></td>
<td><strong>Behçet's Syndrome</strong></td>
<td>Dry, brittle, flat, telangiectatic</td>
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<td></td>
<td><strong>Craniosynostosis</strong></td>
<td>Absence of the anterior fontanelle</td>
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<td></td>
<td><strong>Chemotherapy medications (mother &amp; baby)</strong></td>
<td>Clavicular, Cartilaginous - inner ear hair cells affected</td>
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<td></td>
<td><strong>Cerebral Palsy</strong></td>
<td>Hypoplastic aplasia during development or birth asphyxia</td>
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<td></td>
<td><strong>Cranial abnormalities</strong></td>
<td>Atelesia of the ear canal</td>
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<td></td>
<td><strong>Claustrum</strong></td>
<td>Atelesia, akinesia of the pinnus</td>
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<td></td>
<td><strong>Cleft palate</strong></td>
<td>Malformation of the hard palate (Exclude cleft lip if only feature present)</td>
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<tr>
<td></td>
<td><strong>CHARGE syndrome</strong></td>
<td>Coloboma - eyes, Heart, Atelesia of the nasae, Genital, Ear - deafness</td>
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<td></td>
<td><strong>Coarctation of the aorta</strong></td>
<td>Narrowing of the circulator</td>
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<td></td>
<td><strong>Hemifacial Microsomia</strong></td>
<td>Abnormal development on one side of the face, atresia of the outer ear</td>
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<tr>
<td></td>
<td><strong>Hypertrophic osteoarthropathy</strong></td>
<td>Hyperplasia of the cartilaginous surface, hypoplastic limb, significant deformity</td>
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<td></td>
<td><strong>Hypophosphatemia</strong></td>
<td>Vitamin D &amp; phosphate deficiency</td>
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<td></td>
<td><strong>Hypermocomenia</strong></td>
<td>Increase in the production of antibodies</td>
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<td></td>
<td><strong>Hypermotility</strong></td>
<td>Increased muscular activity</td>
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<tr>
<td></td>
<td><strong>Idiopathic</strong></td>
<td>Idiopathic</td>
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<tr>
<td></td>
<td><strong>Infectious</strong></td>
<td>Infection, Auricular involvement</td>
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<tr>
<td></td>
<td><strong>Joseph's Syndrome</strong></td>
<td>Bone dysplasia, increased skeletal density affecting auditory function</td>
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<td></td>
<td><strong>Kawasaki's Disease</strong></td>
<td>Perianal pain, growth retardation</td>
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<td></td>
<td><strong>Legionnaires' Disease</strong></td>
<td>Periventricular lesions, intracranial hemorrhage</td>
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<tr>
<td></td>
<td><strong>Liebgold's Syndrome</strong></td>
<td>Periventricular lesions, intracranial hemorrhage</td>
</tr>
<tr>
<td></td>
<td><strong>Lisch's Ataxia</strong></td>
<td>Progressive ataxia, oculopathy</td>
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<tr>
<td></td>
<td><strong>Lissencephaly</strong></td>
<td>Primary brain defect</td>
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<tr>
<td></td>
<td><strong>Marfan's Syndrome</strong></td>
<td>Abnormal development of the bone, atresia of the outer ear</td>
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<tr>
<td></td>
<td><strong>Meckel's syndrome</strong></td>
<td>Abnormal development of the bone, atresia of the outer ear</td>
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<tr>
<td></td>
<td><strong>Meningitis</strong></td>
<td>Meningitis</td>
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<tr>
<td></td>
<td><strong>Neurofibromatosis I (NF1)</strong></td>
<td>Absence of development of the brain (in the) cochlear</td>
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<tr>
<td></td>
<td><strong>Neurofibromatosis II (NF2)</strong></td>
<td>Absence of development of the brain (in the) cochlear</td>
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<tr>
<td></td>
<td><strong>Neurofibromatosis type III</strong></td>
<td>Absence of development of the brain (in the) cochlear</td>
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<tr>
<td></td>
<td><strong>Norrie's Syndrome</strong></td>
<td>Absence of development of the brain (in the) cochlear</td>
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<td></td>
<td><strong>Otomatolabyrinthic Dysplasia (OAD)</strong></td>
<td>Facial asymmetry, anomalies of external, middle ear, cranial nerve</td>
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<td></td>
<td><strong>Otosclerosis</strong></td>
<td>Progressive progressive loss, pulpanetopathy in children</td>
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<td></td>
<td><strong>Pendred's Syndrome</strong></td>
<td>Thyroid goiter - iodine imbalance in inner ear cells</td>
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<td><strong>Pierre Robin Syndrome</strong></td>
<td>Craniosynostosis, micrognathia, glaucoma, may have cleft palate</td>
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<td></td>
<td><strong>Polychromatophilia</strong></td>
<td>Polychromatophilia</td>
</tr>
<tr>
<td></td>
<td><strong>Pseudohypoparathyroidism type 1B</strong></td>
<td>Hypoplastic anomalies, middle ear involvement</td>
</tr>
<tr>
<td></td>
<td><strong>Progeria</strong></td>
<td>Accelerated aging</td>
</tr>
<tr>
<td></td>
<td><strong>Quadruplet syndrome</strong></td>
<td>Absence of development of the bone, atresia of the outer ear</td>
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<tr>
<td></td>
<td><strong>Rett's Syndrome</strong></td>
<td>Absence of development of the bone, atresia of the outer ear</td>
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<tr>
<td></td>
<td><strong>Russell-Silver syndrome</strong></td>
<td>Hypoplastic anomalies, middle ear involvement</td>
</tr>
<tr>
<td></td>
<td><strong>Schober's syndrome</strong></td>
<td>Absence of development of the bone, atresia of the outer ear</td>
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<tr>
<td></td>
<td><strong>Shaw's syndrome</strong></td>
<td>Absence of development of the bone, atresia of the outer ear</td>
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<tr>
<td></td>
<td><strong>Sjögren's syndrome</strong></td>
<td>Absence of development of the bone, atresia of the outer ear</td>
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<tr>
<td></td>
<td><strong>Tuberous Sclerosis</strong></td>
<td>Absence of development of the bone, atresia of the outer ear</td>
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<tr>
<td></td>
<td><strong>Turner's syndrome</strong></td>
<td>Absence of development of the bone, atresia of the outer ear</td>
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<tr>
<td></td>
<td><strong>Usher's syndrome</strong></td>
<td>Absence of development of the bone, atresia of the outer ear</td>
</tr>
<tr>
<td></td>
<td><strong>Von Willebrand's disease</strong></td>
<td>Absence of development of the bone, atresia of the outer ear</td>
</tr>
</tbody>
</table>

### Hearing loss (HL)

- **Conductive HL**
- **Sensorineural HL**
- **Central factors**
- **Mixed HL**
- **Progressive mixed HL**

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**Children’s Health Queensland Hospital and Health Service**

Craniofacial anomalies
Some examples of cranio-facial “high risk” factors
(not exhaustive list)

- Cleft palate (*not cleft lip)
- Facial anomalies, asymmetry
- Dysmorphic, flattened features
- Paralysis
- Nasal anomalies
- Craniosynostosis
- Microcephaly, hydrocephaly
- Neck webbing

*Excluded from screening*
Ears – major pinna malformations, microtia, stenosis, atresia
► Direct referral to Audiology
### What the literature says – population studies

<table>
<thead>
<tr>
<th>Wood et al</th>
<th>Beswick et al</th>
</tr>
</thead>
<tbody>
<tr>
<td>2013</td>
<td>2012</td>
</tr>
<tr>
<td>NHS England</td>
<td>Queensland</td>
</tr>
<tr>
<td>N= 2 307 880</td>
<td>N= 261,328</td>
</tr>
<tr>
<td>Whole population</td>
<td>Whole population</td>
</tr>
<tr>
<td>Highest prevalence risk factors:</td>
<td>Syndrome, craniofacial anomalies, severe asphyxia had the highest yield of postnatal hearing loss</td>
</tr>
<tr>
<td>1. Syndrome other than Downs</td>
<td></td>
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<tr>
<td>3. Craniofacial</td>
<td></td>
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<tr>
<td>4. Down syndrome</td>
<td></td>
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</tbody>
</table>
What the literature says – condition specific studies

<table>
<thead>
<tr>
<th></th>
<th>Park et al</th>
<th>Raut et al</th>
<th>Viswanathan et al</th>
<th>Chen et al</th>
</tr>
</thead>
<tbody>
<tr>
<td>Year</td>
<td>2012</td>
<td>2011</td>
<td>2008</td>
<td>2008</td>
</tr>
<tr>
<td>Location</td>
<td>Utah</td>
<td>Singapore</td>
<td>Chelmsford</td>
<td>Boston</td>
</tr>
<tr>
<td>Sample size</td>
<td>N=344</td>
<td>N=45</td>
<td>N=90</td>
<td>N=114</td>
</tr>
<tr>
<td>Condition</td>
<td>Down syndrome</td>
<td>Down syndrome</td>
<td>Cleft palate</td>
<td>Cleft palate</td>
</tr>
<tr>
<td><strong>% developed conductive hearing loss</strong></td>
<td>43.5%</td>
<td>82.3% (14) of assessed children (17) had a hearing loss. At 1 year:-</td>
<td>ABR &lt; 3 months of age • 74 (82%) had a hearing loss the majority • 7 infants had a mixed loss</td>
<td>15 children were identified with permanent hearing loss • 13 referred at birth • 2 identified at 1 year of age</td>
</tr>
<tr>
<td><strong>% developed sensorineural loss</strong></td>
<td>0.4%</td>
<td>0.4%</td>
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</tbody>
</table>
Method

- All neonates who were born in Queensland, Australia between July 2012 – December 2014 who had completed Healthy Hearing’s newborn hearing screen (2-stage AABR) and had the risk factors of syndrome and/or CFA.

- Data was extracted from Healthy Hearing’s database, QChild, and de-identified prior to data analysis.
Results - July 2012 - December 2014

153,897 eligible for screening

391 identified with a syndrome and/or CFA

85 Infants were direct refer/medical exclusion
- 35 CFA
- 14 CFA + other RFs
- 23 Syndrome
- 3 Syndrome + other RFs
- 10 CFA + Syndrome

306 Early Targeted Surveillance (seen at 6 weeks)
- 134 CFA
- 22 CFA + other RFs
- 118 Syndrome
- 14 Syndrome + other RFs
- 18 CFA + Syndrome
Direct Refer/medical exclusion
Did not pass the newborn hearing screen or bypassed the screen

- Hearing Loss: 38%
- Transient Conductive HL: 21%
- WNL: 17%
- Refused Audiology: 5%
- Lost Contact: 7%
- Attended In Progress: 7%
- Deceased: 5%
- Moved: 1%
- No data: 2%

N= 85
Early Targeted Surveillance
Passed newborn hearing screen and seen at Audiology at 6 weeks

- Hearing Loss: 3%
- Transient Conductive HL: 26%
- WNL: 50%
- Refused Audiology: 3%
- Attended In Progress: 7%
- Lost Contact: 6%
- Moved: 1%
- No data: 4%

N= 306
Early Targeted Surveillance
Passed newborn hearing screen and seen at Audiology at 6 weeks

- Craniofacial: 60 cases
- Craniofacial + other RFs: 66 cases
- Syndrome: 23 cases
- Syndrome + other RFs: 5 cases
- Craniofacial + Syndrome: 13 cases

- No data
- Moved
- Refused Audiology
- Lost Contact
- Attended In Progress
- WNL
- Transient Conductive HL
- Hearing Loss

Children’s Health Queensland Hospital and Health Service
Early Targeted Surveillance

Passed newborn hearing screen and seen at Audiology at 6 weeks

Craniofacial

Craniofacial + Syndrome

Syndrome + other RFs

Craniofacial + Syndrome

No data
Moved
Refused Audiology
Lost Contact
Attended In Progress
WNL
Transient Conductive HL
Hearing Loss

Children’s Health Queensland Hospital and Health Service
Early Targeted Surveillance
Passed newborn hearing screen and seen at Audiology at 6 weeks
What does this tell us?

Targeted Surveillance at 9-12 months is not enough for every child with a risk factor.

>25% of ETS children had a HL at 6 weeks.
Future directions

Continue to refer children for Early Targeted Surveillance

Develop screening pathway guidelines for children with the risk factors of craniofacial anomalies and syndrome
Should infants with a syndrome or craniofacial anomaly be screened for a hearing loss at birth?

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