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

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Background

CONTROVERSY

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

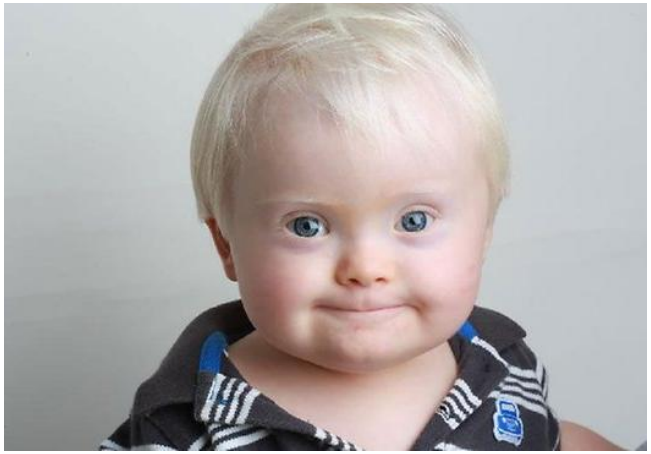
2012 REVIEW

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

Hearing Loss - Syndrome and condition list for congenital and progressive hearing loss

	Title	Description	Hearing loss (HL)
A	Achondroplasia	Dwarfism, skeletal ossification disorder	Conductive & Sensorineural HL
	Albers-Schonberg Disease of Osteopetrosis	Brittle, thickened, chalky bones	Conductive & Sensorineural HL
	Albinism with blue irides	Pigmentation disorder eyes,skin, hair	Sensorineural HL
	Alport's Syndrome	Nephritis and cataracts	Progressive Sensorineural HL
	Apert Syndrome	Craniosynostosis, midface anomalies, middle ear involvement	Conductive HL
	Aplasias (errors during embroyonic development)		
	Michel aplasia	Complete absence of inner ear & auditory nerve	Sensorineural HL
	Mondini aplasia	Abnormal development of the structure (turns) of the cochlear	Sensorineural HL
	Scheibe aplasia	Abnormal formation of the cochlear membrane	Sensorineural HL
	Asphyxia at birth / neonatal period	Rescusitation required/poor APGARs, seizures, neurological involvement	Sensorineural HL, auditory neuropathy
B	Bacterial meningitis	Auditory involvement, can have sudden permanent H'loss	Sensorineural HL, central effects
	Bjornstad Syndrome	Dry, brittle, flat, twisted hair	Sensorineural HL
	Branchio-Oto-Renal syndrome (BOR)	Renal anomalies, auricular pits, pinnae malformations	Conductive, sensorineural & mixed HLs
	Carraro Syndrome	Absence of the Tibia bone	Sensorineural HL
C	Camurati-Engelmann Disease	Skeletal - enlarged diaphysis of the long bones	Conductive & Sensorineural HL
	Chemotherapy medications (mother & baby)	Cisplatin, Carboplatin - inner ear hair cells affected	Sensorineural HL
	Cerebral palsy	Hypoxic episode during development or birth asphyxia	Sensorineural HL
	Craniofacial abnormalities		
	Atresia of the ear canal	Atresia, stenosis of the ear channel	Conductive, Sensorineural & Mixed HLs
	Absence or malformed pinna	Atresia, stenosis, malformation of the pinnae	Conductive, Sensorineural & Mixed HLs
	Cleft palate	Malformation of the hard palate (Exclude cleft lip if only feature present)	Conductive HL
	CHARGE syndrome	Coloboma - eyes, Heart, Atresia of the nares, Genital, Ear - deafness	Conductive, sensorineural & mixed HLs. Can have aud neuropathy
	Cleidocranial Dysostosis	Retarded ossification, narrowed auditory canal	Conductive & Sensorineural HL
	Cockayne's syndrome	Growth failure and neurologic delay, retinal atrophy,	Sensorineural Hearing Loss
	Cornelia de Lange Syndrome	SGA, limb malformations, cardiac defects, cleft palate	Conductive, Sensorineural or mixed HLs
	Crouzon's syndrome	Craniosynostosis, midface anomalies, outer & middle ear defects	Conductive, SN or mixed (Majority are conductive)
D	Dwarfism	Skeletal anomalies, shortness, short fingers	Sensorineural HL
	Down syndrome	Middle ear anomalies - ossicles, otitis media infections	Conductive, Sensorineural or mixed HLs
E	Encephalitis	Infection, Auditory involvement	Sudden permanent Sensorineural HL
	Engelmann's Syndrome	Bone dysplasia, increased skeletal density affecting auditory function	
F	Fanconi's anaemia syndrome	Impaired renal transport, growth delay	Sensorineural HL
	Family history of hearing loss	Permanent HL evident in early infancy < 6 years (see QH - S&R list)	Conductive or sensorineural HL
	Fetal Alcohol Syndrome	LBW, skeletal anomalies, cleft palate, pinnae anomalies	Conductive & Sensorineural HL
	Fraser Syndrome	Adherent eyelids, external ear malformations, syndactyly	Conductive & Sensorineural HL
	Friedreich Ataxia	Progressive ataxia, cataracts	Sensorineural HL
G	Goldenhar's syndrome	Eye, ear and mouth anomalies	Conductive or Sensorineural HL
H	Hemifacial microsomia	Abnormal development on one side of the face, atresia/ stenosis canal	Conductive or Sensorineural HL
	Hermann's Syndrome	Late onset of disease. Epilepsy, speech, ataxia, renal disease	Sensorineural HL
	Hyperbilirubinemia	Dampening of the Auditory nerve function due to excessive bilirubin	Sensorineural HL, may have aud neuropathy
	Hypoxic Ischaemic Encephalopathy **HIE**	Severe aphyxia with neurological sequalae, hypotonic limbs, significant morbidity	Sensorineural HL, may have aud neuropathy
	Hydrocephalus	IVH Grade 3 & 4, internal cranial anomalies, 8th Cranial Nerve involvement	Sensorineural HL
	Hunter's and Hurler's Syndrome	Progressive manifestation of coarse facial features	Mixed HL
I	**Infections**		
	Cytomegalovirus	Herpes virus 5, microcephaly, hepatosplenomegaly, jaundice, IUGR	Sensorineural HL
	Herpes	Congenital neonatal herpes infection HSV-1 & 2 - High mortality	Sensorineural HL
	Rubella	LBW, purpura, jaundice, Organ of Corti degeneration	Sensorineural HL
	Toxoplasmosis	Parasitic infection, chorioretinitis, cerebral calcification, convulsions	Sensorineural HL
	Syphillis	Nasal discharge, rash, anaemia, jaundice, osteochondritis	Sensorineural HL
	Intraventricular haemorrhage **IVH**	Bleeding within the brain structures causing adverse neurological complications	Sensorineural HL & Central effects
J	Jervell and Lange-Nielsen syndrome	Cardiovascular disorder, fainting, sudden death a feature, auditory involvement	Sensorineural HL
K	Keratopachyderma & digital Constrictions Nephrosis	Pigment disorder, may include renal disease	Sensorineural HL
L	Laurence-Moon-Biedl-Bardet Syndromes	Retinitis pigmentosa, polydactyly	Sensorineural HL
	LEOPARD Syndrome (Multiple lentigines syndrome)	Pigment disorder, café au lait spots, cardiac, ocular, genital, growth delay	Sensorineural HL
	Long QT Syndrome	Cardiac condition	
	Low birth weight <1500 gms	Multifactorial predictor for increased perinatal morbidity risks	Sensorineural HL & Central effects
M	Marshall Syndrome	Short stature, skeletal defects, cataracts	Sensorineural HL
	Meningitis	Inner hair cells in cochlear damaged by virus	Sensorineural HL

Hearing Loss - Syndrome and condition list for congenital and progressive hearing loss

	Title	Description	Hearing loss (HL)
	Mitochondrial disorders	DNA - Maternal inheritance pattern	
	Moeibus(Mobius)Syndrome	Connective tissue disorder, facial paralysis Cranial nerves 6 & 7, middle ear anomalies	Conductive & Sensorineural HL
	Muckle-Wells Syndrome	Onset in teens, urticaria, renal failure	Sensorineural HL
N	Neurofibromatosis type II	Intracranial tumours, 8th Cranial nerve, acoustic neuroma	Sensorineural HL
	Noonan's Syndrome	See Leopard syndrome, café au lait spots	Sensorineural HL
	Norries Syndrome	Eye disorder, auditory impairment	Sensorineural HL
O	Oculo-Auriculo-Vertebralia Spectrum (OAV)	Facial asymmetry, anomalies of external, middle ear, cranial nerve	Sensorineural HL & Central effects
	Optic atrophy and polyneuropathy	Progressive visual loss, polyneuropathy in childhoods	Sensorineural HL (progressive)
	Ototoxic Medication - affecting inner ear hair cells	Neomycin, Amikacin, Gentamycin, Kanamycin, Sisomicin, Tobramycin, Dibekacin, Steptomycin	Sensorineural HL
		Frusemide (loop diuretic used in conjuction with Antibiotics) Quinine - malarial treatment	Sensorineural HL
	Osteogenesis imperfecta	"brittle bones", stapes malformation	Conductive & Sensorineural HL
P	Paget's disease	Juvenile skeletal disorder, bone pain, swelling	Progressive mixed h'loss
	Persistent Pulmonary Hypertension of the Newborn **PPHN**	Ventilation, progressive hypoxia, persistent fetal circulation	Sensorineural HL & Central effects
	Pierre Robin Syndrome	Craniofacial anomaly, micrognathia, glossoptuosis, may have cleft palate	Conductive & Sensorineural HL
	Periauricular abnormalities	Periauricular pits, tags, fistulas, ear canal atresia, facial paralysis	Conductive or sensorineural
	Periventricular leucomalacia **PVL**	Ischaemic cystic changes in the brain matter predisposing to Cerebral palsy	
	Piebaldness	Lack of pigment in hair, ataxia, blue irides	Sensorineural HL
	Pendred's Syndrome	Thyroid goiter - iodine imbalance in inner hair cells	Sensorineural HL
	Pyle's Syndrome	Enlargement and sclerosis of the facial bones, ribs, clavicles	Sensorineural HL
Q			
R	Refsum's Syndrome	Organ of Corti degeneration, inner ear anomalies, eye disorder	Progressive Sensorineural HL
	Richards-Rundle Syndrome	CNS disorder, ataxia muscle wastings	Progressive Sensorineural HL
S	Stickler syndrome	Flattened facial profile, cleft palate, ocular changes	Conductive & Sensorineural HL
T	Treacher Collins syndrome	Head and neck anomalies, atresia of canal, abnormal middle ear	Conductive HL
	Trisomy 21 (Down syndrome)	Recurrent Middle ear infections	Conductive & Sensorineural HL
	Trisomy 13 -15 & 18	High mortality rate	Conductive or Sensorineural HL
	Turner's Syndrome	Gonadal dysgenesis, webbed neck & digits, micrognathia	Conductive & Sensorineural HL
U	Usher Syndrome	Retinitis pigmentosa, tunnel vision, vertigo organ of Corti degeneration	Sensorineural HL
V	Ventilation	Mechanical ventilation for longer than 5 days - increased neonatal risks	Sensorineural HL
	Van der Hoeve's syndrome	"brittle bone", stapes malformation	Conductive & Sensorineural HL
	Vohwinkel-Nockemann Syndrome	See Keratopachyderma reference above	Sensorineural HL (may be progressive)
	Von Reckinghausen's Syndrome	Hyperkeratosis of palms, soles, knees, elbows, acoustic neuroma, renal	Sensorineural HL
W	Waardenburg's Syndrome (Type 1&2)	White forelock, iris colour different in one eye, prominent mandible, cleft	Sensorineural HL
	Wildervanck's Syndrome	Dysmorphic facial features, atresia of ear canals, eyeball retraction,	Sensorineural HL or mixed
	Winter Syndrome	Renal anomalies, genital malformation, malformed ear and canals	Conductive HL
XYZ			

John Muir Medical Centre USA - Hearing loss indication list 2000

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

- Eyes –cataracts, albinism, eyelid
- malformation, coloboma
- Mouth – hemifacial microsomia
- glossoptuosis
- Chin - micrognathia

__Excluded from screening__

Ears – major pinna malformations, microtia, stenosis, atresia

►Direct referral to Audiology

What the literature says – population studies

Wood et al	Beswick et al
2013	2012
NHS England	Queensland
N= 2 307 880	N= 261,328
Whole population	Whole population
Highest prevalence risk factors: 1. Syndrome other than Downs 3. Craniofacial 4. Down syndrome	Syndrome, craniofacial anomalies, severe asphyxia had the highest yield of postnatal hearing loss

What the literature says – condition specific studies

Park et al	Raut et al	Viswanathan et al	Chen et al
2012	2011	2008	2008
Utah	Singapore	Chelmsford	Boston
N=344	N=45	N=90	N=114
Down syndrome	Down syndrome	Cleft palate	Cleft palate
43.5% developed conductive hearing loss 0.4% developed sensorineural loss	82.3% (14) of assessed children (17) had a hearing loss. At 1 year:- • 4 WNL • 3 mild to moderate loss • 5 LTFU	ABR < 3 months of age • 74 (82%) had a hearing loss the majority • 7 infants had a mixed loss	15 children were identified with permanent hearing loss • 13 referred at birth • 2 identified at 1 year of age

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

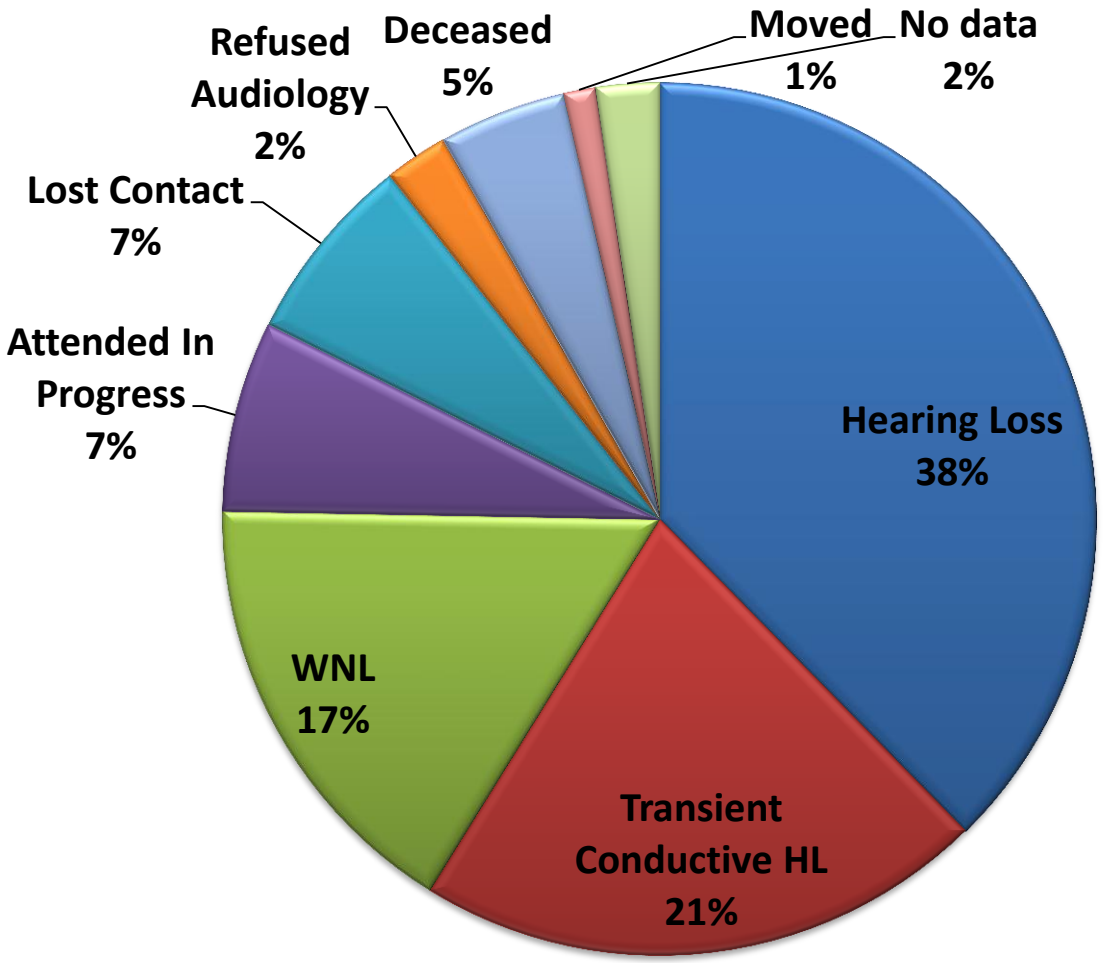
35 CFA
14 CFA + other RFs
23 Syndrome
3 Syndrome + other RFs
10 CFA + Syndrome

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

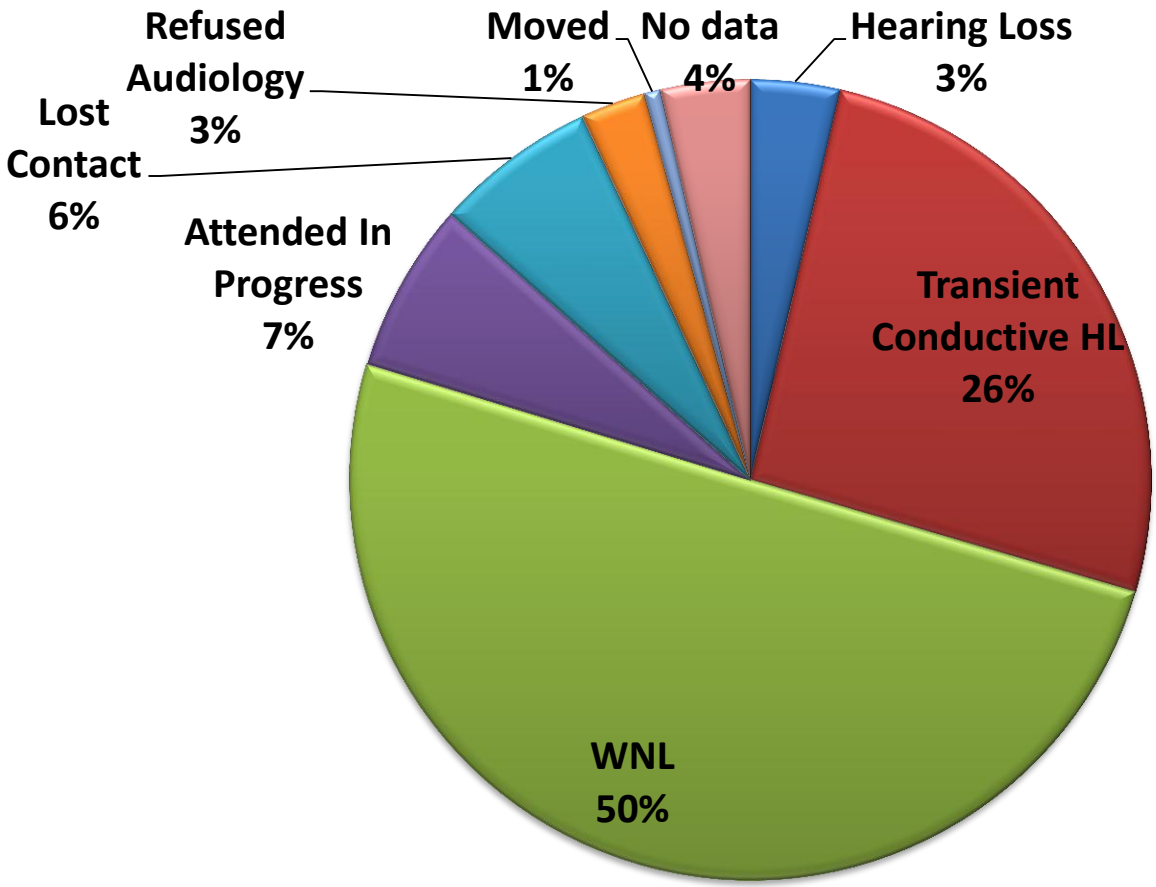
Did not pass the newborn hearing screen or bypassed the screen



N= 85

Early Targeted Surveillance

Passed newborn hearing screen and seen at Audiology at 6 weeks



N= 306

Early Targeted Surveillance

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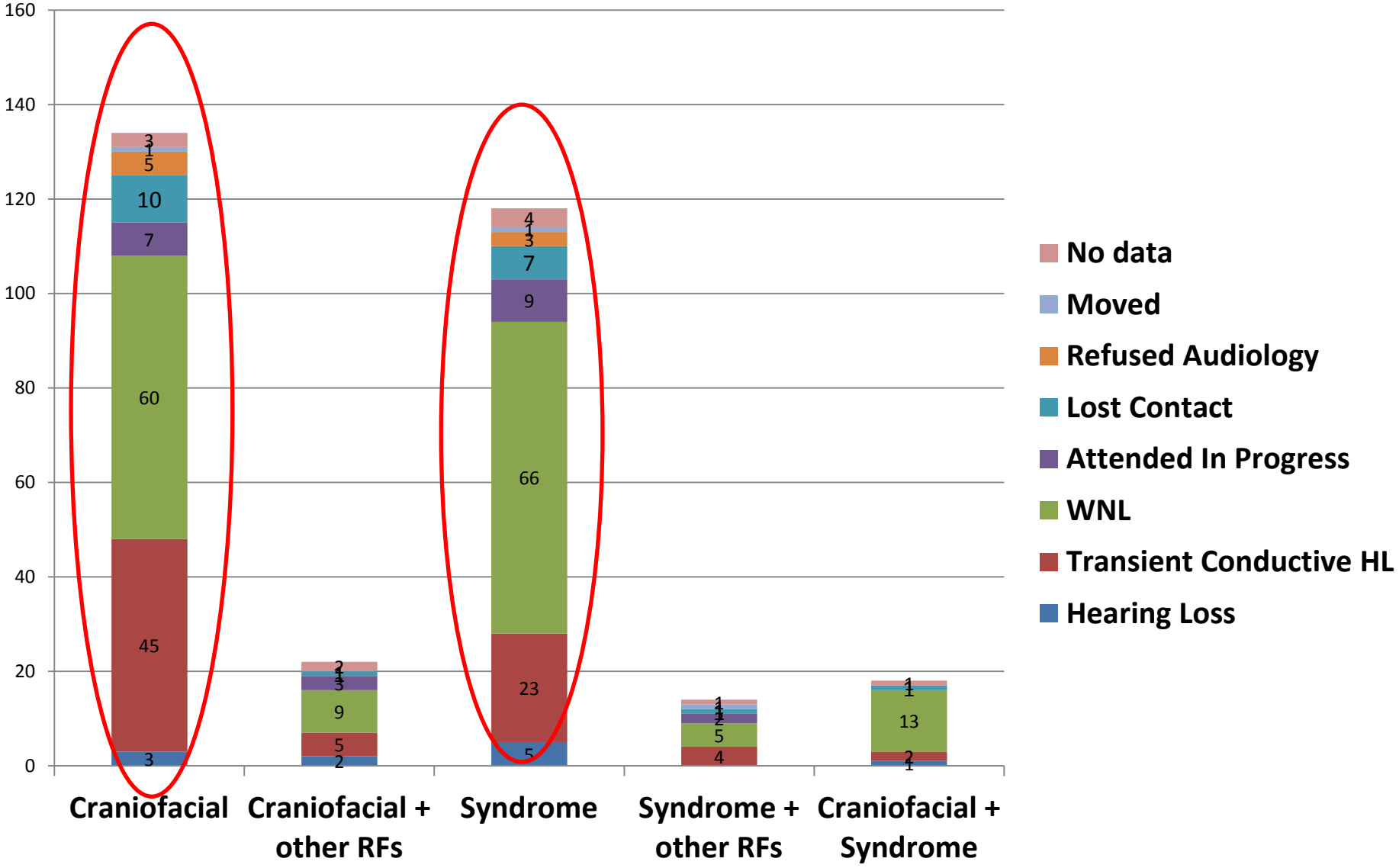

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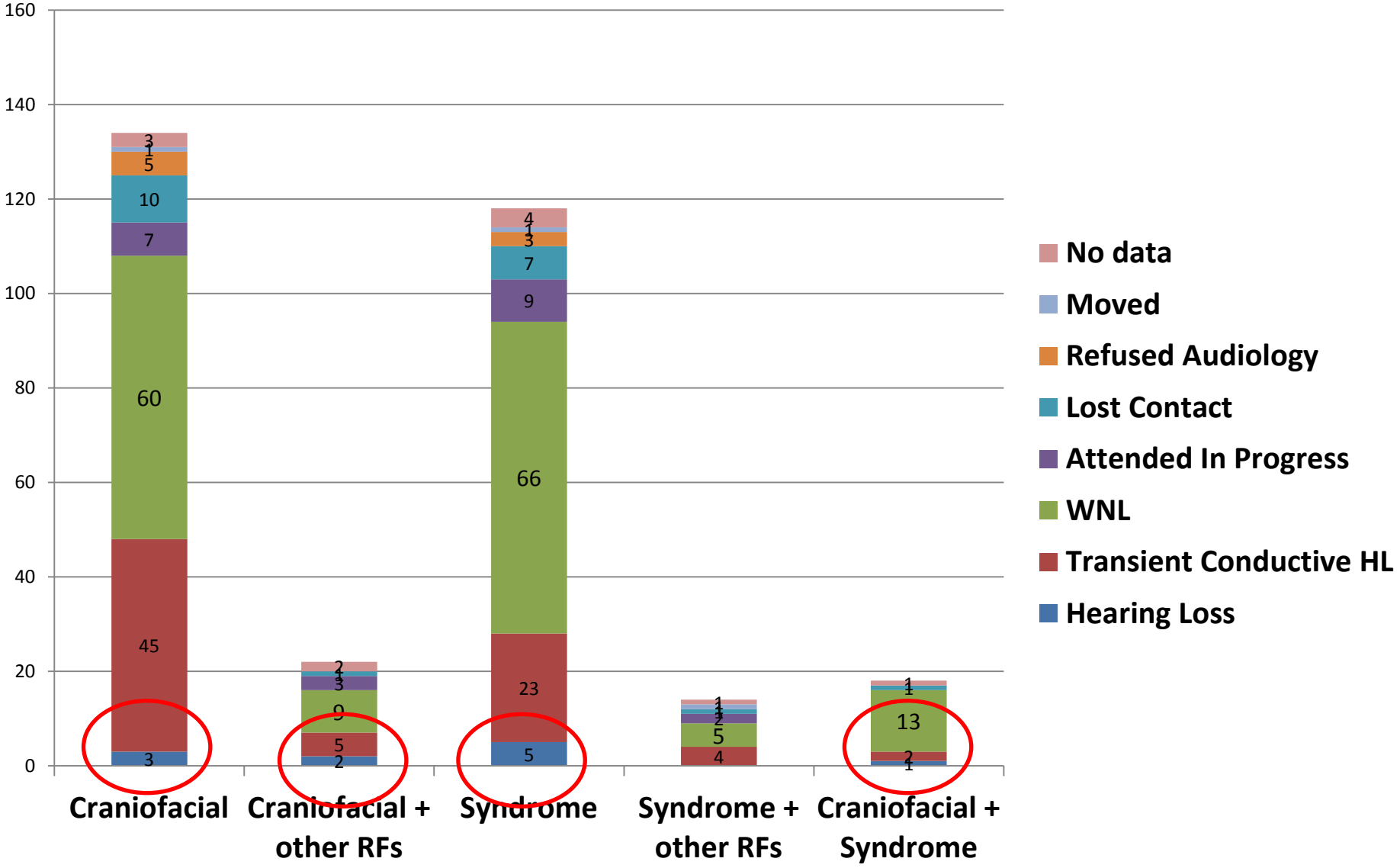

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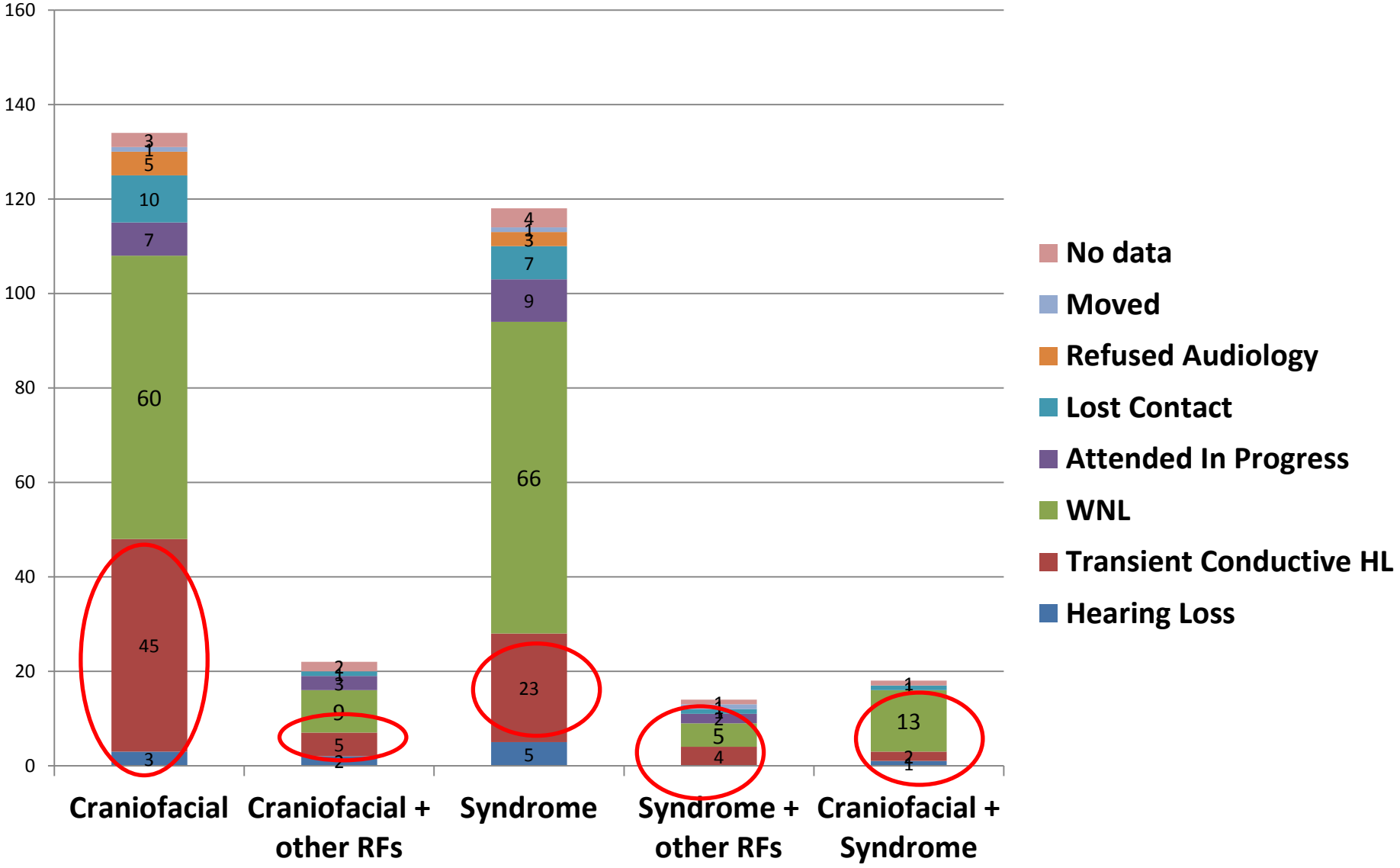

What does this tell us?

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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