Audiology Clinical Practice Guideline

Cleft Palate/Craniofacial And Syndromic Patients (Website version)

2012-March

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I. SUMMARY

Background:

There are three Cleft Palate (CP) programs in BC, including the BCCH program which also serves the craniofacial population (CP/CF). There is wide variability in Audiology follow up for this group of patients resulting in duplication and gaps in service. A need was identified for a model for care for these patients, as well as for craniofacial and other Syndromic patients not typically seen within the three programs.

A thorough literature review revealed there were no distinct protocols reported for these patients. The one relevant guideline (*The American Cleft Palate-Craniofacial Association – Parameters of Care*) was reviewed as an 'expert opinion' document. Their recommendation for individual ear hearing results by 3 months of age for this population was not currently being met in BC. Telephone and in person interviews were conducted with the major Audiology and Cleft/Craniofacial programs across Canada. Extensive literature reviews were conducted using relevant search terms and sources.

Given the lack of evidence reviewed guidelines, best practice audiology principles for young children were applied to inform this Guideline for this population.

The Guideline was developed by a working group including BC Children's Hospital (BCCH) Audiology Department, BCCH CP/CF Team and the BC Early Hearing Program (BCEHP). It was reviewed by Public Health Audiologists; BCCH Otolaryngology Department; BCCH Quality Improvement Project; BCEHP Regional Coordinators Council; Health Authority Audiology Professional Practice Leads or Senior Audiologists; BCEHP, Parent Coordinator; BCEHP Steering Committee Health Authority Representatives and the BCCH, Kelowna and Victoria CP Teams.

General Description of the Guideline

This Guideline recommends a care path specific to the hearing loss risk for each group of children. Specific recommendations include:

- All CP/CF and Syndromic (see specific list of syndromes on page 20) infants in BC will receive a full diagnostic ABR prior to 3 months of age, regardless of their screening outcome. This is due to the mild, atypical and temporary hearing loss configurations common to this population.
- Care paths for patient follow up have been developed for cleft palate, craniofacial and syndromic patients, including Down Syndrome, based on their particular type of degree of risk. This includes:

  Close periodic follow-up is required in groups at risk for recurrent middle ear disorders

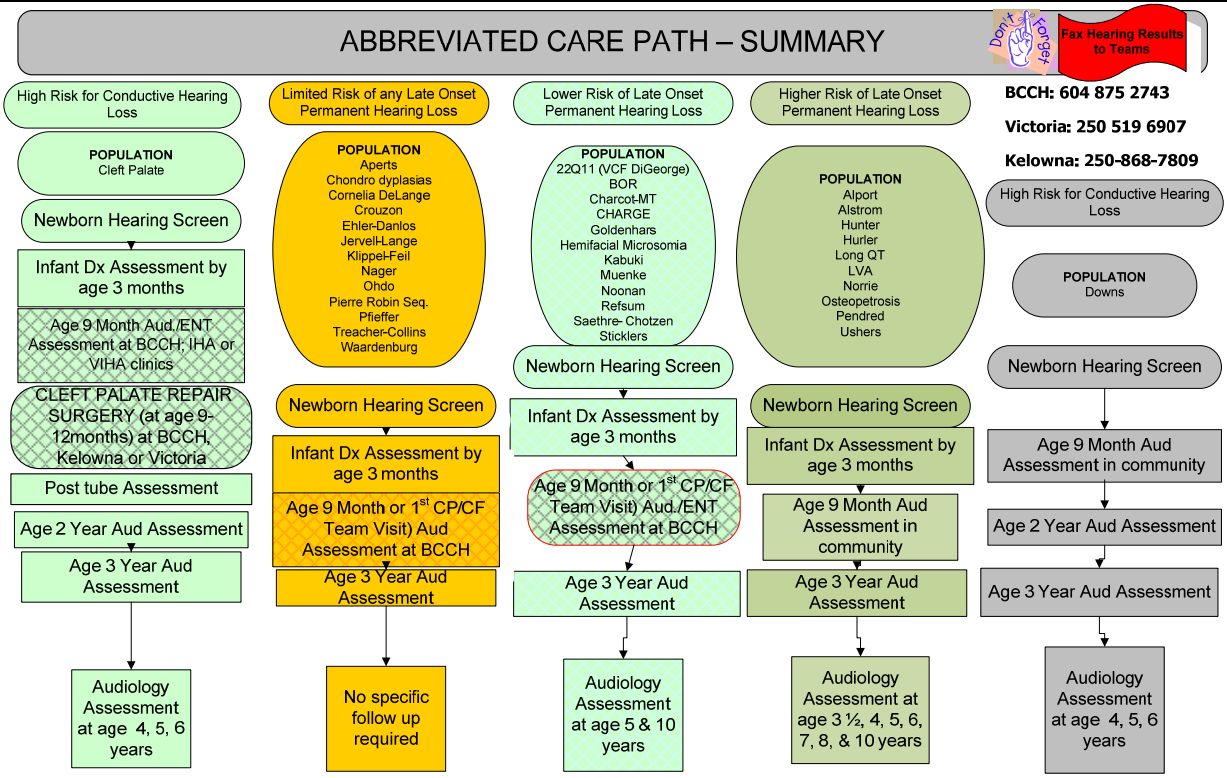
  Periodic follow–up for all infants with a permanent hearing loss

  Use of high frequency probe-tone tympanometry for infants under 6 months of age

  Assessment of acoustic reflexes ipsilaterally, using broad band noise stimuli

- A full review of recent literature as well as survey information from various stakeholders will be conducted to inform future revisions by BCCH Audiology every two years.
- Exceptions to locations and timing of services are encouraged to accommodate patient centred care.

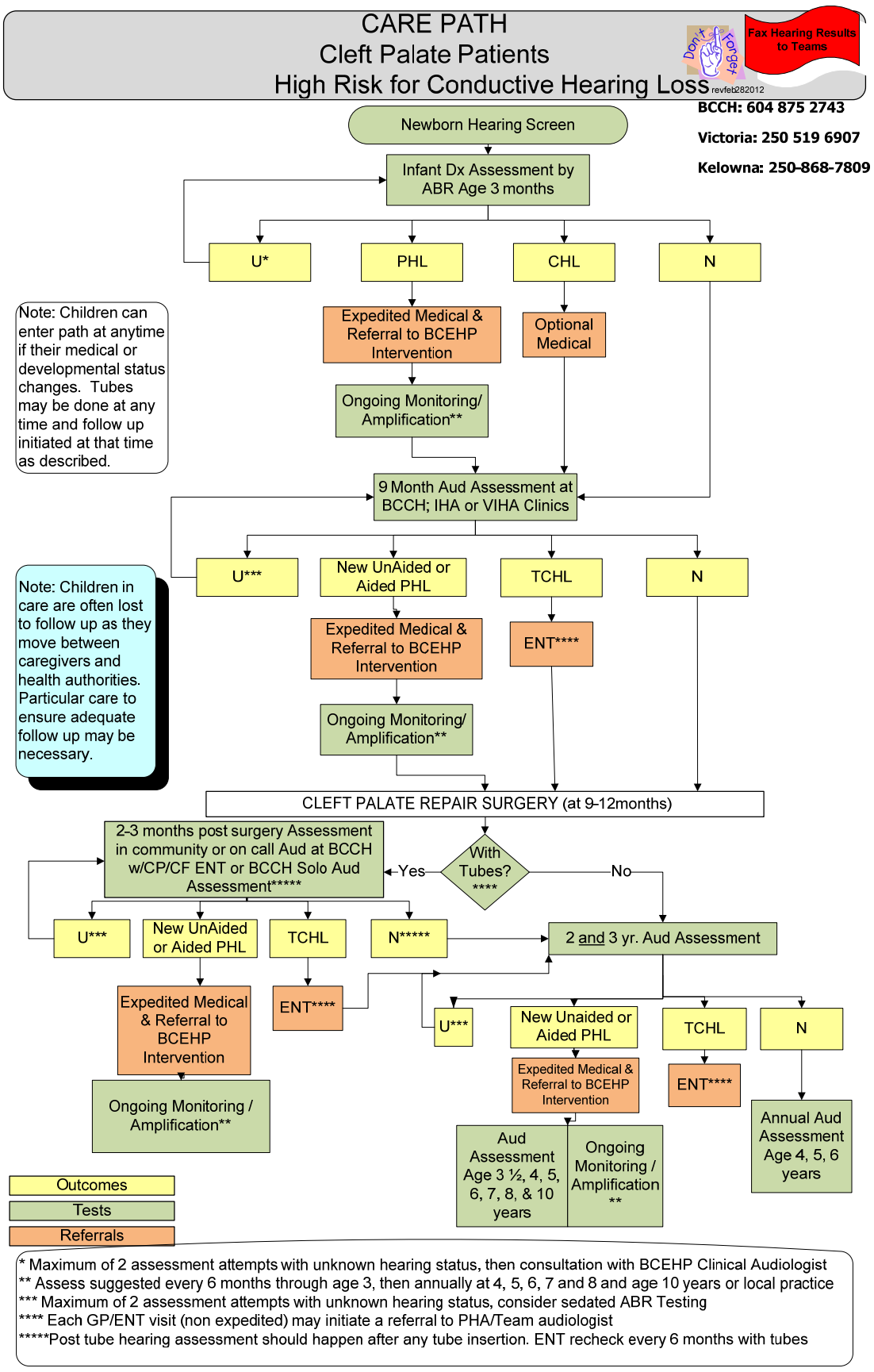
CARE PATHS



Notes: Crosshatched steps are done at BCCH (or Victoria). Children can enter path at anytime if their medical or developmental status changes. Anytime temporary conductive hearing loss found referral to GP or ENT recommended. Each GP/ENT visit (non-expedited) may initiate a referral to PHA/Team audiologist. Many of these children will be referred from ENT. Maximum of 2 Infant Dx. Assessment attempts with unknown hearing status, then consultation with BCEHP Clinical Audiologist. Maximum of 2 assessment attempts with unknown hearing status, consider sedated ABR Testing
For any aided PHL, assess suggested every 6 months through age 4, then annually at 5, 6, 7 and 8 and age 10 years or local practice
Deviation to ongoing monitoring determined on individual basis with community care team. Tubes may be done at any time and follow up initiated at that time as described. Follow up post tube assessment either at BCCH audiology or community (see specific Care Pathway).

Legend for all Care Pathways

PHL	Permanent (Sensorineural, Mixed or Conductive Hearing Loss)
CHL	Conductive Hearing Loss
TCHL	Temporary Conductive Hearing Loss
BCEHP	BC Early Hearing Program
U	Unknown Hearing Status
N	Hearing thresholds within normal limits bilaterally
PHA	Public Health Audiology
ENT	Otolaryngology
CP/CF	Cleft Palate / Craniofacial Team
Aud	Audiology


CARE PATH
Cleft Palate Patients
High Risk for Conductive Hearing Loss
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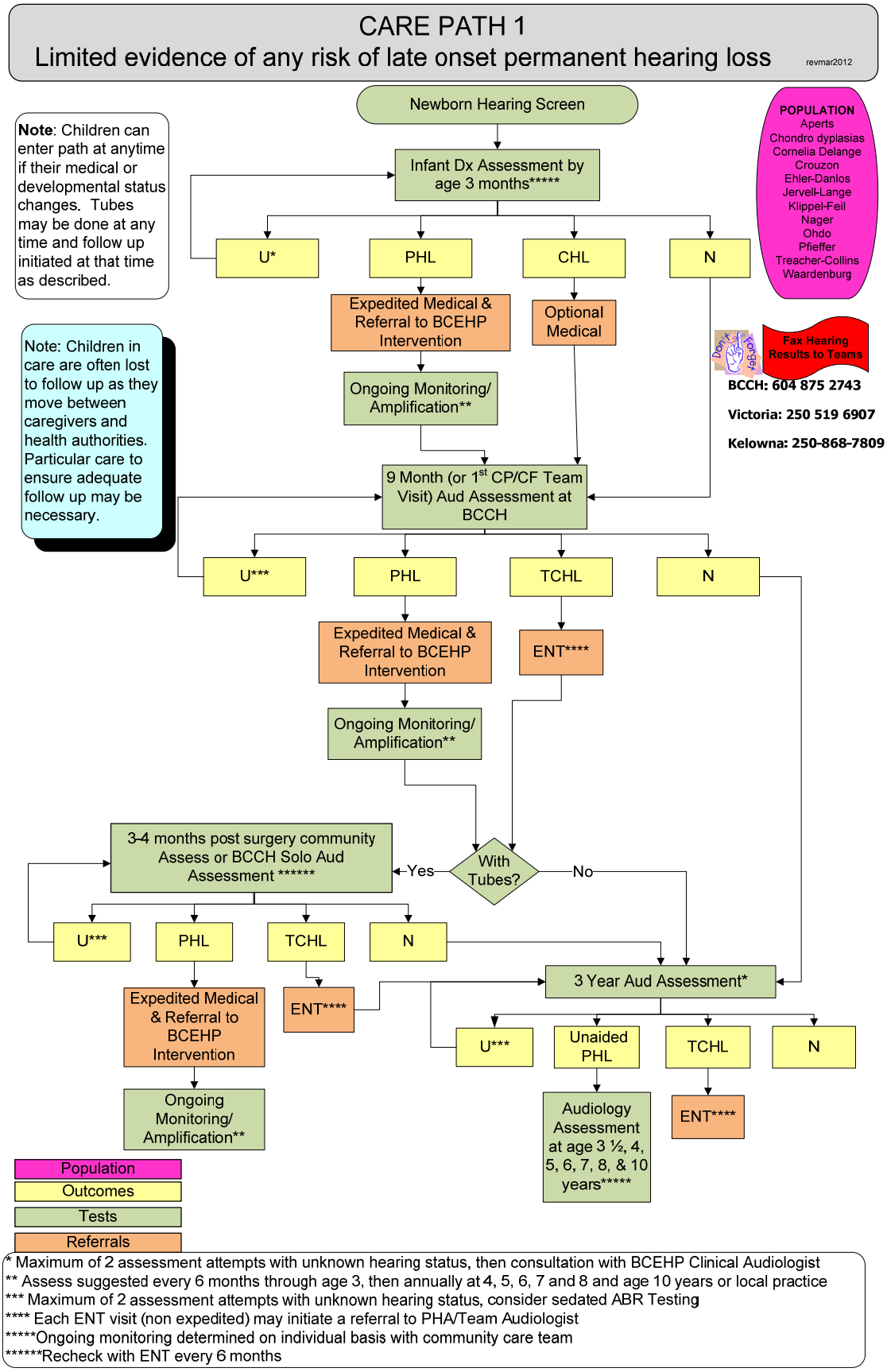
Don't Forget
Fax Hearing Results to Teams
BCCH: 604 875 2743
Victoria: 250 519 6907
Kelowna: 250-868-7809
Newborn Hearing Screen
Infant Dx Assessment by ABR Age 3 months
U*
PHL
CHL
N
Note: Children can enter path at anytime if their medical or developmental status changes. Tubes may be done at any time and follow up initiated at that time as described.
Expedited Medical & Referral to BCEHP Intervention
Optional Medical
Ongoing Monitoring/ Amplification**
9 Month Aud Assessment at BCCH; IHA or VIHA Clinics
U***
New UnAided or Aided PHL
TCHL
N
Note: Children in care are often lost to follow up as they move between caregivers and health authorities. Particular care to ensure adequate follow up may be necessary.
Expedited Medical & Referral to BCEHP Intervention
ENT****
Ongoing Monitoring/ Amplification**
CLEFT PALATE REPAIR SURGERY (at 9-12months)
2-3 months post surgery Assessment in community or on call Aud at BCCH w/CP/CF ENT or BCCH Solo Aud Assessment*****
Yes
With Tubes? ****
No
U***
New UnAided or Aided PHL
TCHL
N*****
2 and 3 yr. Aud Assessment
Expedited Medical & Referral to BCEHP Intervention
ENT****
Ongoing Monitoring / Amplification**
U***
New Unaided or Aided PHL
TCHL
N
Expedited Medical & Referral to BCEHP Intervention
ENT****
Aud Assessment Age 3 ½, 4, 5, 6, 7, 8, & 10 years
Ongoing Monitoring / Amplification **
Annual Aud Assessment Age 4, 5, 6 years
Outcomes
Tests
Referrals
* Maximum of 2 assessment attempts with unknown hearing status, then consultation with BCEHP Clinical Audiologist
** Assess suggested every 6 months through age 3, then annually at 4, 5, 6, 7 and 8 and age 10 years or local practice
*** Maximum of 2 assessment attempts with unknown hearing status, consider sedated ABR Testing
**** Each GP/ENT visit (non expedited) may initiate a referral to PHA/Team audiologist
*****Post tube hearing assessment should happen after any tube insertion. ENT recheck every 6 months with tubes


CARE PATH 1
Limited evidence of any risk of late onset permanent hearing loss
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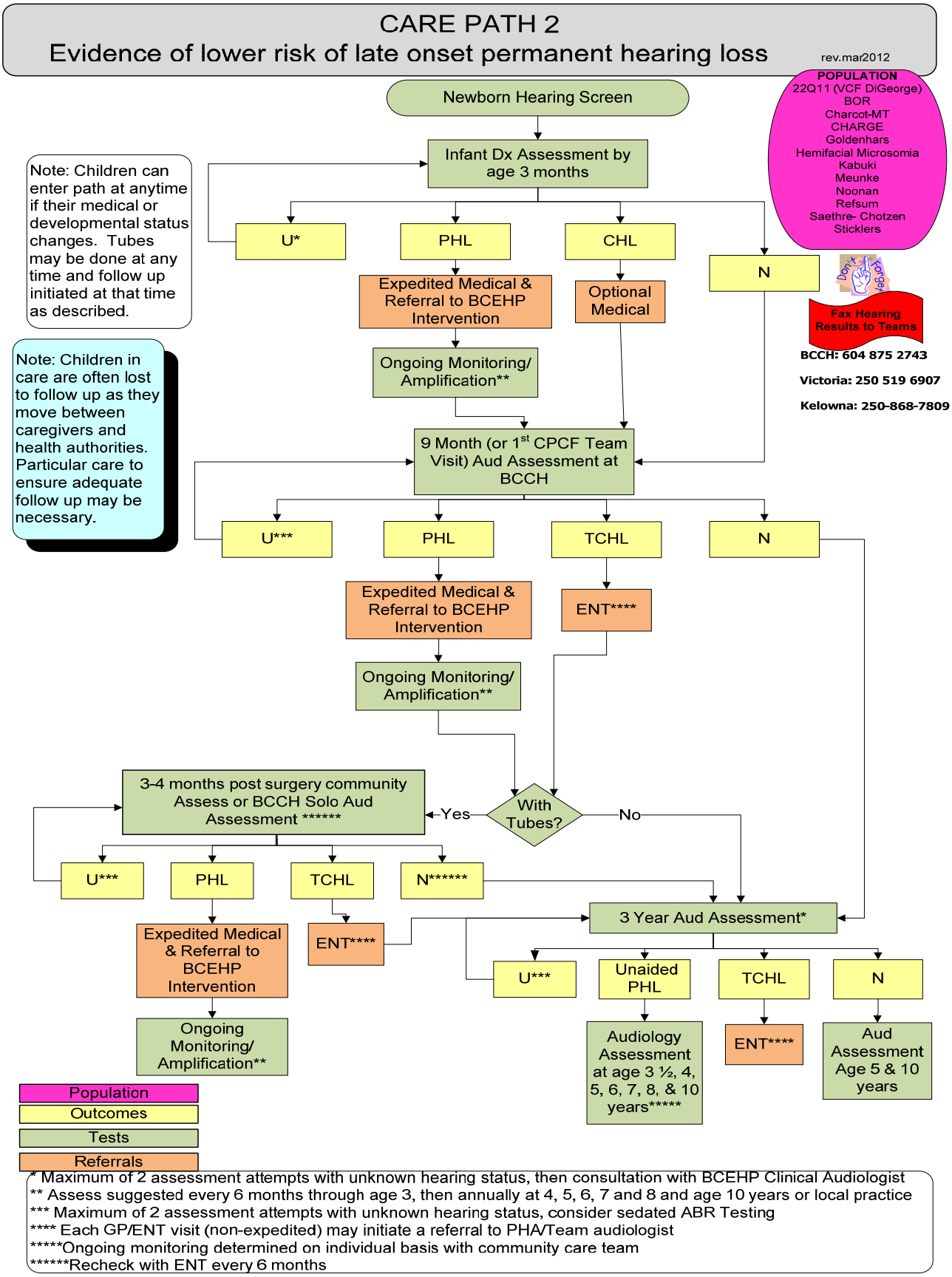
Newborn Hearing Screen
Infant Dx Assessment by age 3 months*****
U*
PHL
CHL
N
Expedited Medical & Referral to BCEHP Intervention
Optional Medical
Ongoing Monitoring/ Amplification**
9 Month (or 1st CP/CF Team Visit) Aud Assessment at BCCH
U***
PHL
TCHL
N
Expedited Medical & Referral to BCEHP Intervention
ENT****
Ongoing Monitoring/ Amplification**
With Tubes?
Yes
No
3-4 months post surgery community Assess or BCCH Solo Aud Assessment ******
U***
PHL
TCHL
N
Expedited Medical & Referral to BCEHP Intervention
ENT****
Ongoing Monitoring/ Amplification**
3 Year Aud Assessment*
U***
Unaided PHL
TCHL
N
Audiology Assessment at age 3 ½, 4, 5, 6, 7, 8, & 10 years*****
ENT****
Note: Children can enter path at anytime if their medical or developmental status changes. Tubes may be done at any time and follow up initiated at that time as described.
Note: Children in care are often lost to follow up as they move between caregivers and health authorities. Particular care to ensure adequate follow up may be necessary.
POPULATION
Aperts
Chondro dyplasias
Cornelia Delange
Crouzon
Ehler-Danlos
Jervell-Lange
Klippel-Feil
Nager
Ohdo
Pfieffer
Treacher-Collins
Waardenburg
Don't forget
Fax Hearing Results to Teams
BCCH: 604 875 2743
Victoria: 250 519 6907
Kelowna: 250-868-7809
Population
Outcomes
Tests
Referrals
* Maximum of 2 assessment attempts with unknown hearing status, then consultation with BCEHP Clinical Audiologist
** Assess suggested every 6 months through age 3, then annually at 4, 5, 6, 7 and 8 and age 10 years or local practice
*** Maximum of 2 assessment attempts with unknown hearing status, consider sedated ABR Testing
**** Each ENT visit (non expedited) may initiate a referral to PHA/Team Audiologist
*****Ongoing monitoring determined on individual basis with community care team
******Recheck with ENT every 6 months



* Maximum of 2 assessment attempts with unknown hearing status, then consultation with BCEHP Clinical Audiologist
** Assess suggested every 6 months through age 3, then annually at 4, 5, 6, 7 and 8 and age 10 years or local practice
*** Maximum of 2 assessment attempts with unknown hearing status, consider sedated ABR Testing
**** Each GP/ENT visit (non-expedited) may initiate a referral to PHA/Team audiologist
*****Ongoing monitoring determined on individual basis with community care team
******Recheck with ENT every 6 months

CARE PATH 3
Evidence of higher risk of late onset permanent hearing loss

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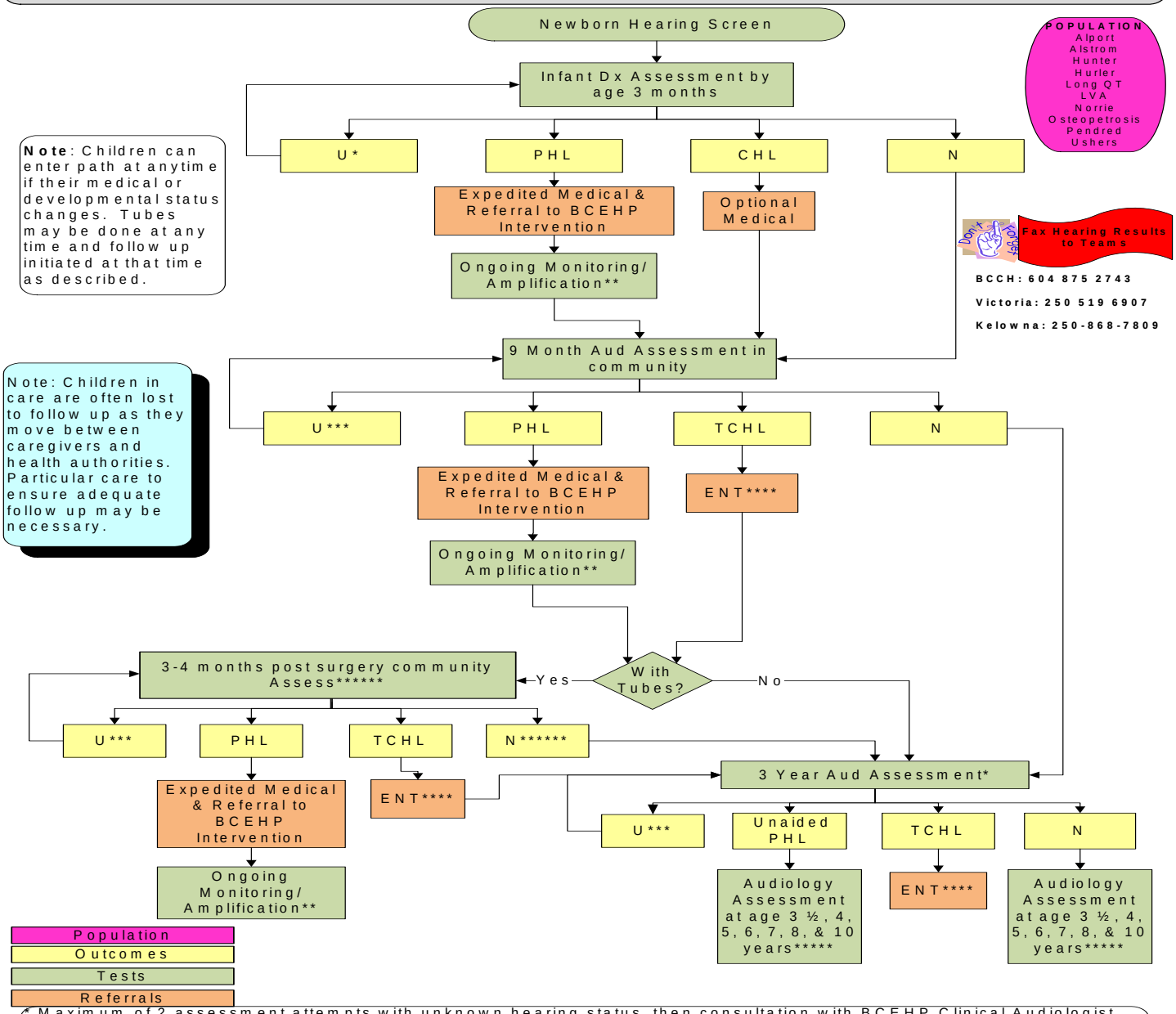

* Maximum of 2 assessment attempts with unknown hearing status, then consultation with BCEHP Clinical Audiologist
** Assess suggested every 6 months through age 3, then annually at 4, 5, 6, 7 and 8 and age 10 years or local practice
*** Maximum of 2 assessment attempts with unknown hearing status, consider sedated ABR Testing
**** Each GP/ENT visit (non-expedited) may initiate a referral to PHA/Team audiologist
***** Ongoing monitoring determined on individual basis with community care team
****** Recheck with ENT every 6 months

CARE PATH 4
Downs Syndrome Patients
High Risk of Conductive Hearing Loss

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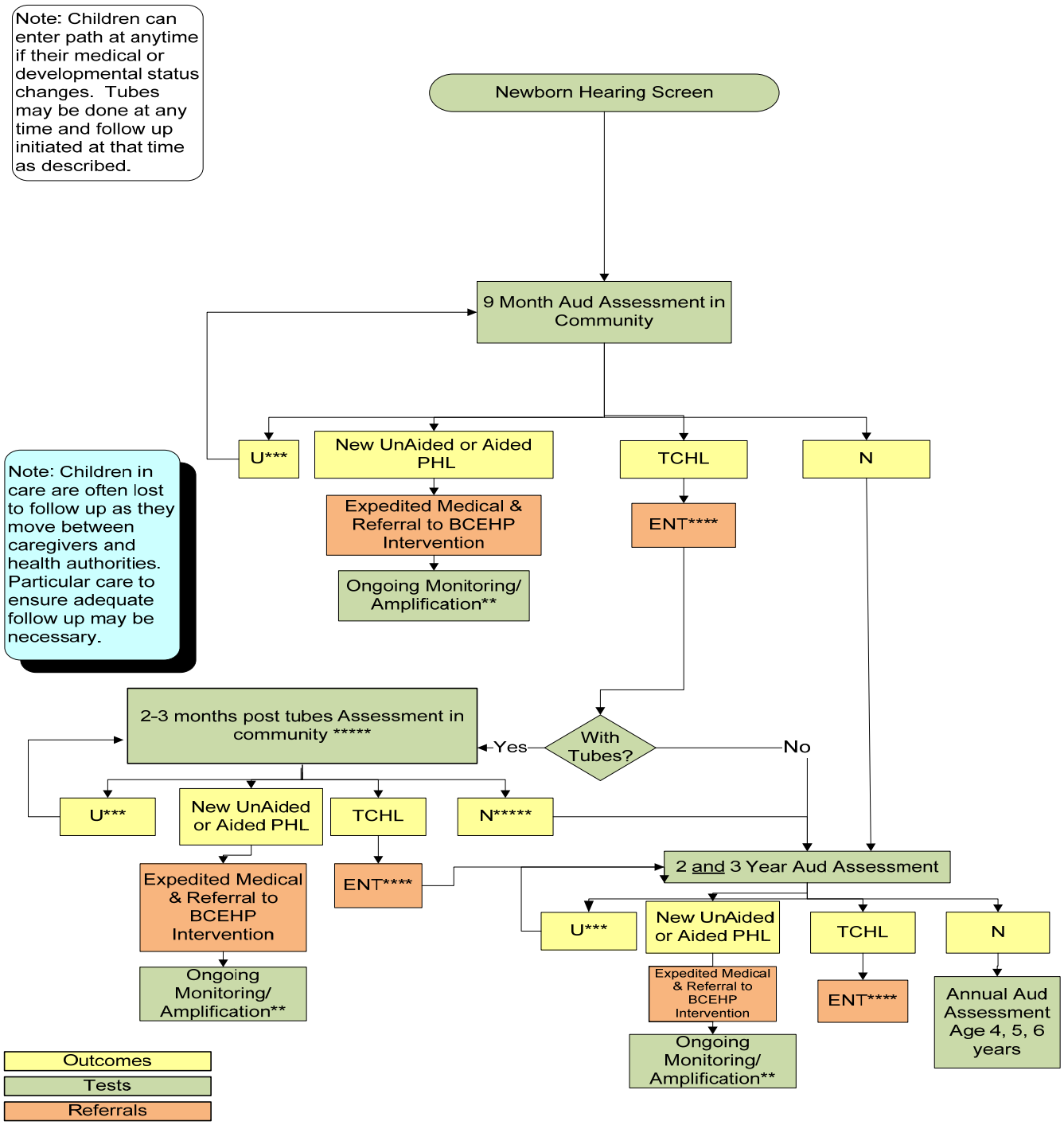

** Assess suggested every 6 months through age 3, then annually at 4, 5, 6, 7 and 8 and age 10 years or local practice
*** Maximum of 2 assessment attempts with unknown hearing status, consider sedated ABR Testing
**** Each GP/ENT visit (non expedited) may initiate a referral to PHA/Team audiologist
*****Tubes can happen at any time, a post assessment in the community is recommended. Recheck with ENT every 6 months

APPENDIX A: SCOPE, PURPOSE, PRINCIPLES & TARGET POPULATION

SCOPE:
This guideline was developed to support consistent Audiologic care of infants/young children in B.C. with Cleft Palate/Craniofacial Anomalies (hereafter referred to as CP/CF) or Syndromes (hereafter referred to as CP/CF/S) associated with hearing loss. It details the type of testing, when and how often testing ought to be done, as well as recommendations for where the testing ought to be conducted. It does not include follow up schedules for children who wear amplification. The guideline came from the review of audiology services provided by BCCH Audiology for children and their families seen by the BCCH Cleft Palate/Craniofacial Team (CP/CF).

PURPOSE:
There is limited evidence for distinct Audiologic protocols for infants with CP/CF/S. Modern protocols for Audiologic assessment in infants and preschool children are directed at specific types and aetiologies of hearing loss, irrespective of the derived patient population. The purpose of this guideline is to provide direction for the Audiologic care of patients with CP/CF/S in BC. The audience for this guideline is primarily Audiologists in Public Health in BC, but other care providers of these children (including Otolaryngologists or Speech Language Pathologists, etc.) might also find it helpful.

PRINCIPLES:
This Guideline meets the following service principles:

- Care coordinated by the Cleft Palate/Craniofacial (CP/CF) teams (BC Children's Hospital (BCCH) in Vancouver; the Queen Alexandra Centre for Children's Health (QACCH) in Victoria, and the Interior Health Cleft Lip/Palate Clinic in Kelowna), to patients of that team, but provided at the local level whenever possible
- Evidence-based practice, including American Cleft Palate Association (ACPA) Practice Guidelines
- Takes advantage of new services available through BC Early Hearing Program and its' clinical information system (BEST)
- "Right" service by "right" resource at "right" time that is seamless and integrated/coordinated from a patient perspective
- Accommodates unique and atypical clinical situations
- Family centered care
- Encourages consistency in service access and provision across BC
- Aligns with the CP/CF Teams, BCCH Audiology reviewed service priorities and those of the Public Health Audiology Programs

- Assists the local Public Health Audiologist with service timelines dependant on the infants' diagnosis.

TARGET POPULATON DESCRIPTION:

The patient population for this practice guideline includes:

- Children from birth to age 17 years AND
- Children with cleft of the palate and cleft lip and palate OR
- Children with any craniofacial anomaly or syndrome known to have increased hearing loss risk

Many of these children, but not all, are served by one of the three CP/CF programs. There are currently three surgical cleft palate programs. The BCCH program also serves craniofacial patients. Many syndromic patients without clefting or craniofacial anomalies are not typically seen by any of the programs however they are covered in these Practice Guidelines as their inclusion was felt appropriate.

This practice guideline excludes children with isolated cleft lip. The literature does not support that these children have an increased incidence of hearing loss, other than if the cleft lip is associated with a syndrome with increased risk of hearing loss.

APPENDIX B: DESCRIPTION, LOCATION & OUTCOMES

Patient Population	Service Description	Service Location	Clinical Outcome
All CP/CF/S infants	**Newborn Hearing Screen** – Enter in BEST	Various as per EHP protocol	As per BCEHP Screening Protocols
All CP/CF/S infants (except Downs')	**By 3 mo., ABR**-Based Initial Diagnostic Assessment to inform surgical decisions and identify mild unusual configurations common with this population	Fraser Health and Vancouver Coastal Health infants seen at BCCH; Other Health Authorities seen at their regional diagnostic Audiology centres	Ear and frequency specific A/C at .5, 2 and 4 kHz. And BC thresholds at .5 and 2 kHz As per BCEHP ABR-based Initial Diagnostic Assessment
All CP/CF/S infants	**9 mo Audiology Assessment** or 1st Team Visit – Enter in BEST	If child followed by one of the CP/CF teams, seen at their 'typical' audiology site (BCCH Audiology for BCCH team, etc.); otherwise seen at regional Public Health Audiology clinics	1. Establish middle ear status - Ipsilateral WN (or BB) Reflexes; 2. Behavioural thresholds: .5, 1, 2, 4k and 6 kHz bilaterally by A/C and B/C if elevated A/C thresholds[1] 3. DP- OAEs if middle ear status normal 4. Link with needed community resources, including schools 5. Anticipatory Guidance (see description on page 20) Optional: Sound field hearing demonstration if hearing loss found

[1] Hearing loss in the craniofacial anomaly patient population without clefting tend to be unusual configurations including cookie bite hearing losses, up-sloping hearing losses, slight/mild and therefore every effort to obtain mid frequencies is urged when possible.

Patient Population	Service Description	Service Location	Clinical Outcome
CP infants with myringotomy and/or ventilating tube insertion	**2-3 months post tube Audiology Assessment**	Seen at Public Health Audiology OR seen at BCCH through Audiology On Call Service at time of BCCH Otolaryngology recall visit  ***Implementation Idea: BCCH CP/CF Team will make referral in BEST to Public Health Audiology if its' needed, for this post tube assessment, at the time of their 9mo. assessment***	1. Establish middle ear status - Ipsilateral WN (or BB) Reflexes; 2. Behavioural thresholds: .5, 1, 2, 4k and 6 kHz bilaterally by A/C and B/C if elevated A/C thresholds 3. DP- OAEs if middle ear status normal 4. Link with needed community resources, including schools Optional: Sound field hearing demonstration if hearing loss found
CP and Down Syndrome infants	**Age 2 Audiology Assessment**	Public Health Audiology Clinics	1. Establish middle ear status - Ipsilateral WN (or BB) Reflexes; 2. Behavioural thresholds: .5, 1, 2, 4k and 6 kHz bilaterally by A/C and B/C if elevated A/C thresholds 3. DP- OAEs if middle ear status normal 4. Link with needed community resources, including schools Optional: Sound field hearing demonstration if hearing loss found

Patient Population	Service Description	Service Location	Clinical Outcome
Cleft Palate patients without hearing loss at 9 month assessment or post tube assessment	**Age 3 ,4, 5 & 6 year** Audiology **Assessment** and on referral - Ongoing medical management of middle ear function during this time, physician responsible to refer to Audiology for additional assessments if there are concerns	Public Health Audiology Clinics	1. Establish middle ear status - Ipsilateral WN (or BB) Reflexes; 2. Behavioural thresholds: .5, 1, 2, 4k and 6 kHz bilaterally by A/C and B/C if elevated A/C thresholds 3. DP- OAEs if middle ear status normal 4. Speech discrimination as indicated 4. Link with needed community resources, including schools Optional: Sound field hearing demonstration if hearing loss found

Patient Population	Service Description	Service Location	Clinical Outcome
CF/S Patients without any hearing loss at 9 months (or 1st team assessment) and limited evidence of risk of permanent late onset hearing loss These are: **Aperts** **Chondro dyplasias** **Cornelia DeLange** **Crouzon** **Down** **Ehler-Danlos** **Jervell-Lange** **Klippel-Feil** **Nager** **Ohdo** **Pfieffer** **Treacher-Collins** **Waardenburg**	Audiology assessment at 3 years. There is limited evidence of late onset permanent hearing loss with these patients, however community consensus suggests a 3 year assessment	Public Health Audiology Clinics	1. Establish middle ear status - Ipsilateral WN (or BB) Reflexes; 2. Behavioural thresholds: .5, 1, 2, 4k and 6 kHz bilaterally by A/C and B/C if elevated A/C thresholds 3. DP- OAEs if middle ear status normal 4. Speech discrimination as indicated 5. Link with needed community resources, including schools Optional: Sound field hearing demonstration if hearing loss found

Patient Population	Service Description	Service Location	Clinical Outcome
CF/S Patients without any hearing loss at 9 months (or 1st team assessment) and lower risk of permanent late onset hearing loss **These are:** **22Q11 (VCF/DiGeorge)** **Charcot-MT** **CHARGE** **Goldenhars** **Hemifacial Microsomia** **Kabuki** **Meunke** **Noonan** **Refsum** **Saethre- Chotzen** **Sticklers**	**Audiology assessment at age 3, 5 and 10 years.**	Public Health Audiology Clinics	1. Establish middle ear status - Ipsilateral WN (or BB) Reflexes; 2. Behavioural thresholds: .5, 1, 2, 4k and 6 kHz bilaterally by A/C and B/C if elevated A/C thresholds 3. DP- OAEs if middle ear status normal 4. Speech discrimination as indicated 5. Link with needed community resources, including schools Optional: Sound field hearing demonstration if hearing loss found

Patient Population	Service Description	Service Location	Clinical Outcome
CF Patients without any hearing loss at 9 months (or 1st team assessment) and with higher risk of permanent late onset hearing loss **This population includes:** **Alport** **Alstrom** **Hunter** **Hurler** **Long QT** **LVA** **Norrie** **Osteo Imperfecta** **Pendred** **Ushers**	**Audiology assessment** at 3, 3 ½, annually 4-8 years and 10 years.	Public Health Audiology Clinics	1. Establish middle ear status - Ipsilateral WN (or BB) Reflexes; 2. Behavioural thresholds: .5, 1, 2, 4k and 6 kHz bilaterally by A/C and B/C if elevated A/C thresholds 3. DP- OAEs if middle ear status normal 4. Speech discrimination as indicated 5. Link with needed community resources, including schools Optional: Sound field hearing demonstration if hearing loss found
Patients diagnosed with the following syndromes need an individualized care path as their age of diagnosis varies widely and is often in later childhood or adolescence: **Friedreich ataxia** **Klinefleter** **Neurofibromatosis** **Osteogenesis imperfecta** **Turners** **Van Buchem**	**Audiology assessment to be determined**	Public Health Audiology Clinics or BCCH	Dependant on clinical questions to be answered.

Patient Population	Service Description	Service Location	Clinical Outcome
Ongoing Monitoring of Patients with Documented Unaided Permanent hearing loss (Sensorineural, Conductive, Mixed, and ANSD)	**Audiologic assessment** every 6 months for at least a year or until stable by Audiologist judgment; ongoing monitoring as per Audiologist.	Public Health Audiology Clinics	1. Establish middle ear status - Ipsilateral WN (or BB) Reflexes; 2. Behavioural thresholds: .5, 1, 2, 4k and 6 kHz bilaterally by A/C and B/C if elevated A/C thresholds 3. DP- OAEs if middle ear status normal 4. Speech discrimination as indicated 4. Link with needed community resources, including schools Optional: Sound field hearing demonstration if hearing loss found

Fax all Audiology results to local CP/CF teams:

BCCH: 604 875 2743

Victoria: 250 519 6907

Kelowna: 250-868-7809

Anticipatory Guidance: At initial sessions with families and ongoing as needed, the following counselling information is reviewed. Guidance and education will be individualized for each family and will vary depending on the age of the first full team review and the diagnosis. The syndromic diagnosis may not be known at the first team review.

- Parental concerns
- Parental observations
- Discussion of previous test results
- Anatomy and physiology review
- Risk of hearing loss in the population, compared to normal population
- Auditory development
- Effects of mild and/or fluctuating hearing loss on behaviour and development
- Explanation of why and how we assess hearing
- Explanation of results
- Observations of the child's skills and abilities from an audiologist's point of view
- Description of timing and coordination of surgical intervention, timing and coordination of community monitoring, intervention and follow-up
- Importance of close medical and audiological monitoring
- Importance of ongoing communication between parents, team and community partners

Red Flag Indicators for CP/CF/S Patients to trigger Referral to BCCH:

- Second opinion, more than two (2) community visits where hearing status is not confirmed and ABR needed.
- Socio-economic, geographic and other barriers to community audiology service such that service uptake in community impacts patient care significantly such as children in and out of care for assessment and poor follow up is experienced.
- Child's lack of progress in development (cognitive, speech and language) is unexplained or not consistent with known conditions.

APPENDIX C. SYNDROMES

There are many known syndromes associated with hearing loss. Many of these have clefting and/or craniofacial anomalies, some of them don't. This list was generated by combining the BCCH Audiology Department list of syndromes and the BCEHP Late Onset Monitoring Risk Factor Syndromes. That list was then compared with those found in the *"Hereditary Hearing Loss and It's Syndromes"* and reviewed by all of the reviewers of this Guideline for completeness. This resulted in the syndromes listed below which are associated with hearing loss.

A literature review was conducted using Pub Med, PEDLYNX, and OMIM databases. Search terms were ('name of syndrome' as listed in Appendix B AND ('Audio*' OR Hear*') in title or abstract, from 1999 to 2010, all languages. Citations were screened by a two reviewers for relevance. Published, peer reviewed articles were selected based on level of evidence with recently published articles describing well-designed randomised controlled trials with comparatively large sample groups taking precedence. High quality systematic reviews and retrospective reviews of clinical data were also used. Case studies of noteworthy results were occasionally noted as a matter of interest or possible focus of higher level literature to be reviewed in the future (when published), but were not considered in determining association of a syndrome with late onset SNHL. If the results of a study were inconclusive or the literature could not clearly associate a syndrome with late onset SNHL (ie. small subject pool or insufficient baseline information) such information was noted but the syndrome was labelled as *not* associated with late onset SNHL.

Four distinct care paths were developed dependant on the level of risk assessed for late onset permanent hearing loss for syndromic children. One care path was developed specifically for Down Syndrome children. Each syndrome was assigned to one of the 4 care paths described. If age of diagnosis of the syndrome was known to be after childhood it is suggested that their care path be individualized (see Osteogenesis Imperfecta; Freidricks Ataxia; Turners, Klinefelter, Van Buchem and NF2). All of these infants will have had at least a newborn hearing screening and a 9 month Audiology assessment. The **BOLDED** syndromes are typically not seen through the CP/CF teams and therefore their 9 month assessment ought to be completed in their local Public Health Audiology Clinics.

SYNDROME /Description	Risk of late onset Permanent HL? Y= Yes evidence found N= No evidence found	Age of Diagnosis of Syndrome	Care Path **Cleft Palate** **1**-minimum f-up **2**-moderate f-up **3**-closest f-up **4**- Downs Syndrome f-up 5- Individualized
22Q11 (VCF/DiGeorge) Velocardiofacial Syndrome Cayler, Shprintzen: typical characteristics include cardiac abnormality (especially Fallot's Tetralogy), abnormal facies, thymic aplasia, can have cleft palate, hypocalcemia Zarchi et al, 2011. Digillio, 1999. Estimated prevalence: 1: 4,000. ~10% cleft palate	N: Primarily CHL related to auricular anomalies and cleft. ~11-20% congenital SNHL possibly related to vascular abnormalities. Case evidence of labrynthine anomalies.	Commonly at birth due to the congenital heart disease and abnormal facies, present in most all cases. If not heart problems can be later diagnosed.	2
DiGeorge sequence: cardiac defects, Thymus hypoplasia and/or T call-mediated immunodeficiency, and hypocalcemia and/or absence of parathyroids- (part of deletion 22q11 spectrum) Digillio, 1999. Erkki et al, 2007. Belmont et al, 2011. Estimated prevalence 1: 4,000.	Y: although primarily CHL related to auricular anomalies and cleft and ~11-20% congenital SNHL as well as some cases of LVA . Hearing loss can be unilateral and can be likely related to vascular abnormalities) as well as some case reports of labyrinthine anomalies. Not enough evidence to determine if significant risk of late onset snhl.	Commonly at birth due to the congenital heart disease and abnormal facies, present in most all cases. If no heart problems can be later diagnosed	2

SYNDROME /Description	Risk of late onset Permanent HL? Y= Yes evidence found N= No evidence found	Age of Diagnosis of Syndrome	Care Path **Cleft Palate** **1-**minimum f-up **2-**moderate f-up **3-**closest f-up **4-** Downs Syndrome f-up 5- Individualized
Alport syndrome: collagen synthesis disease characterized by renal disease. Alves et. al, '08 & Kashtan, '10	Y: BL/HF by late childhood/early adolescence for ~80-90% XL males & AR males & females. In some mutations (i.e. AD) SNHL may not occur until adulthood.	**Variable dependant on gene mutation and extent of kidney problems**	3
Alström syndrome: pigmentary retinopathy, diabetes mellitus, and obesity. Joy et al '07, Marshall et al '11 **Estimated prevalence <1:1,000,000 in general population**	Y: BL/HF progressive late childhood/early adolescence for ~80%. Some incidences of CHL & chronic OM. Symptom onset usually in infancy, but both onset and severity highly variable.	**Variable**	3
Apert Syndrome: FGFR2 craniosynostosis, syndactyly of hands and feet, mental retardation Rajenderhumar, 2005. Curch et al, 2007. Zhou et al, 2009. Robin et al, 2011. Prevalence: ~1: 100,000 to 200,000 live births (differing reports).	N: 3-6% Congenital CHL, >56% CHL ~10-20 yrs. Due to OME. Persistent to adulthood.	At birth or pre-natally	1

SYNDROME /Description	Risk of late onset Permanent HL? Y= Yes evidence found N= No evidence found	Age of Diagnosis of Syndrome	Care Path **Cleft Palate** **1-**minimum f-up **2-**moderate f-up **3-**closest f-up **4-** Downs Syndrome f-up 5- Individualized
Branchio-Oto-Renal syndrome: kidney, ears, and neck abnormalities Kemperman et al, 2004. Henricus et al, 2010. Kimberling et al, 2011. Huang et al, 2012. General prevalence: 1: 40,000 Onset variable, early childhood to early adulthood. Incidence in profoundly deaf children: ~2% ***Kemperman et al: 10/16 cases showed sig. SNHL progression in longitudinal anal. Including some fluctuation assoc. with enlarged endolymphatic duct/sac.***	Y: BL congenital. CHL (~50%), SNHL (~25%) & mixed HL.	Variable	2
Charcot-Marie-Tooth: inherited motor and sensory neuropathy, nephritis Postelmans, 2006. Kabzinska, 2010 Incidence: ~1: 2500 Prevalence varies between subclasses (0-15%). More common for auto-dom. Often slow progression.	Y: Late onset SNHL assoc. with demyelization of CN VIII.	Variable, especially if family history unknown, usually late childhood or early adulthood.	2

SYNDROME /Description	Risk of late onset Permanent HL? Y= Yes evidence found N= No evidence found	Age of Diagnosis of Syndrome	Care Path **Cleft Palate** **1-**minimum f-up **2-**moderate f-up **3-**closest f-up **4-** Downs Syndrome f-up 5- Individualized
CHARGE syndrome: acronym for the set of congenital features: Coloboma of the eye, Heart defects, Atresia of the nasal choanae, Retardation of growth and/or development, Genital and/or urinary abnormalities, and Ear abnormalities and deafness. P*rogressive/LO assoc. with LVA (19% of SNHL).* SNHL Correlated with Facial palsy (P<.025 N=20) Edwards et al, 2002. Morimoto et al, 2006. . Huang et al, 2012 Prevalence: 1: 15,000	N: HL=81% of those: CHL (24%), SNHL or mixed (76%). Chronic OME & infections in CHL.	While features may be present at birth & many are diagnosed pre-natally or in the 1st few weeks, others not until other diagnoses have been ruled out.	2
Chondrodysplasias, e.g. Achondroplasia Szymko-Bennett, 2003. Collins, 2007. Pannier et al 2009. Braverman et al, 2010. Tokgoz-Yilmas et al. 2011. Incidence: 1: 15,000- 1: 40,000 live births (varies by type- Achondro. most common). Estimated prevalence of Rhizomelic Chondro. Punctata Type 1 < 1: 100,000.	N: CHL ~50%--OM & OME. Sporadic report of SNHL, insufficient data/ conflicting evidence.	At birth or prenatally	1

SYNDROME /Description	Risk of late onset Permanent HL? Y= Yes evidence found N= No evidence found	Age of Diagnosis of Syndrome	Care Path **Cleft Palate** **1-**minimum f-up **2-**moderate f-up **3-**closest f-up **4-** Downs Syndrome f-up 5- Individualized
Cornelia Delange Syndrome(also Long QT variant, aka Brachmann De Lange): slow growth before and after birth, severe to profound intellectual disability, skeletal abnormalities distinctive facial features, excessive body hair, microcephaly, some cleft palate 1:10,000-30,000	N: high incidence of congenital severe-profound SNHL	Typically at birth	1
Crouzon Syndrome: FGFR2 craniosynostosis, maxillary hypoplasia, shallow orbits. Church et al, 2007. Karam, 2011. Robin et al, 2011. Prevalence: 1.6: 100,000	N: CHL ~55%--OM & Stenosis or Atresia	Usually 1st year	1
Downs Syndrome aka Trisomy 21 Blaser, 2006. Shott, 2006. Park et al, 2012 **Incidence: 1: 600-800 live births**	**N: 80% CHL. 4-20% mixed or SNHL possibly associated with unresolved/untreated chronic OM, anomalies of the cochlea, internal auditory canal and LVA. Variable data.**	**Typically at birth**	4
Ehlers-Danlos syndrome: synthesis of collagen defects, characterized by hypotonia, ocular abnormalities, joint hypermobility. Miyajima, 2007. Fransiska, 2010 Baumann et al, 2012 Estimated prevalence 1: 20,000.	N: CHL primarily related to otosclerosis or TM immobility Evidence of a variant associated with bilateral high frequency SNHL unknown onset age.	Birth or early infancy	1

SYNDROME /Description	Risk of late onset Permanent HL? Y= Yes evidence found N= No evidence found	Age of Diagnosis of Syndrome	Care Path **Cleft Palate** **1-**minimum f-up **2-**moderate f-up **3-**closest f-up **4-** Downs Syndrome f-up 5- Individualized
Friedreich ataxia: spinocerebellar, resulting in progressive gait ataxia **Delatycki, 2009 & Rance et al 2010** **Prevalence: 2-4: 100,000**	**Y: Progressive SNHL (~10-25%) related to axonal degeneration. Onset typically prior to age 25.**	**mean onset of gait symptoms between 10 and 15 years.**	5- Individualized
Goldenhar syndrome: incomplete development of the ear, nose, soft palate, lip, and mandible (part of the oculo-auriculo-vertebral spectrum) Bisdas et al, 2005. Martelli et al, 2009. Skarzynski, 2009. Prevalence estimated to range from 1: 3,500- 7,000 live births.	N: Cond. component (~70%) assoc. With Cleft palate. SNHL assoc. with cochlear malformation (Skarzynski: 5/14) Congenital. Possible evidence of progressive losses (maybe LVA related)	Within 1st year	2
Hemifacial microsomia: abnormal development of the lower half of the face, most commonly the ears, the mouth and the mandible (part of the oculo-auriculo-vertebral spectrum) Vrabec, 2010. Collett et al, 2011. Incidence: 1: 3,500- 4,500.	N: Primarily CHL. 6-16% prevalence SNHL related to cochlear & vestibular anomalies. Rate of progressive/late onset vs. congenital undetermined.	Usually within 1st year	2

SYNDROME /Description	Risk of late onset Permanent HL? Y= Yes evidence found N= No evidence found	Age of Diagnosis of Syndrome	Care Path **Cleft Palate** **1-**minimum f-up **2-**moderate f-up **3-**closest f-up **4-** Downs Syndrome f-up 5- Individualized
Hunter syndrome (mucopolysaccharidosis II): a lysosomal storage disease characterized by progressive intellectual impairment, death between 10 and 15 years. Rate of progression ~1 db/year. Often will present through ENT due to airway and neck problems. Wold, 2010. Keilmann, 2011. Prevalence ~1:100,000 live births (affects mainly males).	**Y: Progressive SNHL as early as age 2 and more commonly age 4. CHL also common.**	**Variable age of onset**	3

SYNDROME /Description	Risk of late onset Permanent HL? Y= Yes evidence found N= No evidence found	Age of Diagnosis of Syndrome	Care Path **Cleft Palate** **1-**minimum f-up **2-**moderate f-up **3-**closest f-up **4-** Downs Syndrome f-up 5- Individualized
Hurler syndrome (mucopolysaccharidosis I): lysosomal storage disease characterized by coarse facial features, skeletal malformations, recurrent OM, hepatosplenomegaly, and macroglossia, developmental delay. Often will present through ENT due to airway and neck problems. Two basic types (severe and attenuated). Shortened lifespan common (severe - <age 10 and attenuated varies from 20 to normal) Gunilla et al, 2008. Wold et al 2010. Clark et al, 2011. **Prevalence: 1: 100,000 for severe form and 1: 500,000 for attenuated form.**	**Y: L-O SNHL progressing to profound coinciding with developmental delay ~1-4 years of age. Involvement of CNVIII is common. Also CHL, OM & infections.**	**no clinical presentation at birth.** ***Severe MPS I*: feature onset ~1 year,** ***Attenuated MPS*: clinical onset from age 3-10 years,**	3

SYNDROME /Description	Risk of late onset Permanent HL? Y= Yes evidence found N= No evidence found	Age of Diagnosis of Syndrome	Care Path **Cleft Palate** **1**-minimum f-up **2**-moderate f-up **3**-closest f-up **4**- Downs Syndrome f-up 5- Individualized
Jervell and Lange-Nielsen syndrome: variant of long QT syndrome (see below) Mohiddin et al, 2004. Baig 2011, Tranebjaerg, 2010 Estimated prevalence 1.6: 1,000,000 worldwide (higher in areas where consanguineous marriage is common or identified "founder mutation" is present-ie. Norway, 1: 200,000)	N: Long QT Characterized by bilateral congenital profound SNHL	Variable Half of children identified by age 3 due to cardiac issues.	1
Klinefelter syndrome (XXY): hypogonadism, infertility Evans et al, 2000. Visootsak, 2006. Prevalence 1: 500- 1,000 males	N: CHL due to chronic OM, some reports of congenital snhl.	Later childhood	5- Individualized
Klippel-Feil Sequence: fused cervical vertebrae, webbed neck, can have cleft palate Incidence: 1: 40,000 to 50,000 live births.	N: 30% SNHL or CNHL congenital	Early infancy	1
Kabuki: postnatal growth deficiency, onset <1st yr.craniofacial abnormalities, some have cleft palate, some cardiac deficiencies. Barozzi, 2008. Matsumoto et al, 2003. Wessels, 2002. Estimated Prevalence: 1: 32,000 live births	N: 32% CNHL and ongoing OME.	Typically age 2	2

SYNDROME /Description	Risk of late onset Permanent HL? Y= Yes evidence found N= No evidence found	Age of Diagnosis of Syndrome	Care Path **Cleft Palate** **1-**minimum f-up **2-**moderate f-up **3-**closest f-up **4-** Downs Syndrome f-up 5- Individualized
Large Vestibular Aqueduct Syndrome: enlargement of vestibular aqueduct in the inner ear Arjmand, 2004. Dewan et al, 2009. Santos et al, 2010. Gopen et al, 2011. Estimated Prevalence in clinical population 5-15%.	**Y: BL (~67%) or Uni (~33%). Prevalence of L-O SNHL ~96%. Onset of hearing loss is highly variable, ranging from birth to adolescence.**	Early infancy	3
Long QT syndrome: prolongation of QT on ECG, syncope, and sudden death Sopontammarak, 2003. Mohiddin et al, 2004. Gritli et al, 2010. Belmont et al, 2011. Incidence 1: 2,500 Accounts for ~.21% of SNHL.	**Y: Age of onset & severity vary with type & severity of cardiac condition. Penetrance as high as 50%.**	Variable dependant on when cardiac issues arise	3
Meunke Craniosynostosis – FGFR3 mutation, coronal craniosynostosis, fifth finger clinodactyly, Ptosis, developmental delay. Agochukwu et al, 2006. Honnebier et al, 2008. Robin et al, 2011. Estimated Prevalence: 1: 30,000 live births.	N: Typically mild bilateral, symmetric, low-mid frequency, SNHL congenitally.	Usually within 1st year	2

SYNDROME /Description	Risk of late onset Permanent HL? Y= Yes evidence found N= No evidence found	Age of Diagnosis of Syndrome	Care Path **Cleft Palate** **1**-minimum f-up **2**-moderate f-up **3**-closest f-up **4**- Downs Syndrome f-up 5- Individualized
Nager: similar to Treacher-Collins, micrognathia, low set,posteriorly rotated ears, atresia, can have cleft palate Danziger et al, 1990. Opitz, 2003. Hermann et al, 2005. Prevalence unknown, 70 published cases.	N- CHL due to middle and conductive ear pathology.	Variable dependant on severity	1
Neurofibromatosis II (NF2): tumours of the central and peripheral nervous system, including non-malignant vestibuloschwannomas Evans, 2009 Incidence reports range from 1: 40,000 to 1:25,000; and the prevalence from 1:200,000 to 1:80,000.	**Y: Incidence of CN VIII tumours ~90%.**	**Average age of onset 18- 24 years dependant on penotype**	5- Individualized
Noonan syndrome: short stature, characteristic facial features, hypotonia, cardiac abnormalities Tartaglia, 2009. Pierpont, 2010 Estimated prevalence of 1: 1,000 -2,500 live births.	Y: SNHL ~26- 40%. Associated with temporal bone structural anomalies. Can also result in structural CHL & OM.	Most apparent in preschool years	2
Norrie syndrome: retinal detachment, often born blind, possible mental retardation Rehm et al, 2002.Halpin 2005 & 2008 Incidence/ prevalence unknown (case reports only)	**Y: X-linked. Complete penetrance of L-O progressive (mild-profound, assym. HF) SNHL in late childhood/early adolescence. Stable ~35 years.**	Early childhood	3

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Ohdo syndrome: mental retardation, congenital heart disease, blepharophimosis/ptosis, hypoplastic teeth Aizeddin et al, 1998 White et al, 2003 Verloes et al, 2006. Beckett et al, 2008 Prevalence/incidence data unavailable,literature consists of case reports.	N: Sporadic case study reports, controversy over classification. Literature indicates SNHL and CHL. Very little specific information on hearing assessment available (including degree, age of onset/diagnosis & in some cases, type)	Early childhood	1
Osteogenesis imperfecta: disorder of type I collagen metabolism characterized by bone fragility Sainz et al, 2009. Marini, 2010. Forlino et al, 2011. **Prevalence: 1: 15,000- 20,000**	**Y: SNHL or mixed hearing loss including structural CHL & otic capsule demineralization & dehiscence. Typically type III & IV. 6-7% experience loss of mild or greater by 9 years of age. SNHL in 25-60% of cases.**	**Variable**	5 -Individualized
Osteopetrosis: increased osseous density due to defects in osteoclastic resorption Dozier et al, 2005. Fattore et al , 2008. **Incidence of autosomal recessive form: 1: 250,000** **Incidence of autosomal dominant form: 5: 100,000**	**Y: Closure of bone foramina causing CN VIII compression. Can also present with otosclerosis and external auditory canal stenosis.**	**Variable onset and severity of clinical features (infancy or early childhood for autosomal recessive form and early adulthood for autosomal dominant form).**	3

SYNDROME /Description	Risk of late onset Permanent HL? Y= Yes evidence found N= No evidence found	Age of Diagnosis of Syndrome	Care Path **Cleft Palate** **1-**minimum f-up **2-**moderate f-up **3-**closest f-up **4-** Downs Syndrome f-up 5- Individualized
Pendred syndrome: goitre and hypothyroidism Luxon et al, 2003. Huang et al, 2012. Ito et al, 2011. **Estimated prevalence 7.5- 10: 100,000.**	**Y: congenital** or **L-O (devel. by age 3). Progressive & some fluctuation due to LVA and membranous labyrinth abnormalities.**	Variable	3
Pfeiffer syndrome: FGFR1/2 craniosynostosis Church et al, 2007. Desai et al, 2010. Robin et al, 2011. Incidence for all forms combined reported as 1: 100,000.	N: CHL related to ossicular fixation & stenosis ~50-70%. Congenital Mixed/SNHL ~20%	Usually in 1st year	1
Pierre Robin sequence: craniofacial abnormalities incl. cleft palate. Gruen 2005. Medard, 1999. Estimated Prevalence 1: 8500-10,000.	N*: CHL associated with middle ear pathology. Congenital SNHL with PR in isolation. *Associated with many other syndromes that may be associated with L-O SNHL. Ie. *Stickler in ~25% of PR. Case reports of LVA*	Early infancy	Cleft Palate Care Path

SYNDROME /Description	Risk of late onset Permanent HL? Y= Yes evidence found N= No evidence found	Age of Diagnosis of Syndrome	Care Path **Cleft Palate** **1-**minimum f-up **2-**moderate f-up **3-**closest f-up **4-** Downs Syndrome f-up 5- Individualized
Refsum syndrome: phytanic acid storage disease characterized by microcephaly, severe developmental delay, hypotonia, hepatomegaly, retinitis pigmentosa and dysmorphic facial features Bamiou et al 2003. Raine et al 2008. Wanders et al, 2010. Prevalence and incidence data is unavailable but estimated to be very low.	Y: SNHL (predominantly high frequency) related to progressive toxic effects of elevated phytanic acid on peripheral nerves. Progressive, often asymmetrical hearing loss 50-70%. Evidence of CN VIII involvement. Symptom onset, with retinitis pigmentosa usually the first symptom, ranges from 7 months of age to adulthood.	Variable	2
Saethre-Chotzen Syndrome: craniofacial anomalies including variable craniosynostosis Lee et al 2000. Robin et al 2011. 1:25,000-50,000	N: Typically CHL. Sporadic case reports of mixed or SNHL. Single cases at BCCH of presumed late onset.	Due to its variability, age is also variable	2
Stickler syndrome Type 1: flat midface, cleft palate, myopia with retinal detachment and cataracts, musculo-skeletal findings Robin et al, 2010. Francomano, 2010. Szymko-Bennett, 2001. Incidence 1:10,000 ~20% cleft palate	Progressive SNHL in 60% Y: SNHL ~40-50% more severe & *progressive* in type 2 & 3. Can be congenital or late onset. Also CHL often related to CP.	Often after 1st year as myopia not identified	2

SYNDROME /Description	Risk of late onset Permanent HL? Y= Yes evidence found N= No evidence found	Age of Diagnosis of Syndrome	Care Path **Cleft Palate** **1-**minimum f-up **2-**moderate f-up **3-**closest f-up **4-** Downs Syndrome f-up 5- Individualized
Stickler syndrome Type 2 &3: flat midface, cleft palate, myopia with retinal detachment and cataracts, musculo-skeletal findings Robin et al, 2010. Francomano, 2010. Szymko-Bennett, 2001. All 3 types:~20% cleft palate; 1-3:10.000	SNHL in 90% Y: SNHL ~40-50% more severe & *progressive* in type 2 & 3. Can be congenital or late onset. Also CHL often related to CP.	Often after 1st year as myopia not identified	2
Treacher Collins syndrome (mandibulofacial dysostosis): craniofacial abnormalities Pagon et al 2006 1 :50,000 births ~35% have cleft palate	N: Permanent CHL related to outer & middle ear malformation	Typically at birth (sometimes prenatally)	1
Turner syndrome: XO genotype characterized by short stature, infertility, renal abnormalities, chronic otitis media Verver et al, 2010 **1:2000 live females**	Y: Often mid-frequency or high frequency onset in adolescence. Differs with karyotype. **Prevalence of SNHL** varies 10- 66%.CHL ~35%.	Adolescence	5 - Individualized
Usher syndrome types I and II: retinits pigmentosa and vitiligo Friedman et al 2011. Jaijo 2004 **3-5:100,000** **Type 1 constitutes 90% of syndrome.**	N: in type I congenital severe to profound SNHL, in type II usually stable congenital loss in low freq sloping to severe or profound in high frequencies, may also be progressive.	*Typically diagnosed as a result of the congenital hearing loss diagnosis, therefore likely <1 year*	3

SYNDROME /Description	Risk of late onset Permanent HL? Y= Yes evidence found N= No evidence found	Age of Diagnosis of Syndrome	Care Path **Cleft Palate** **1**-minimum f-up **2**-moderate f-up **3**-closest f-up **4**- Downs Syndrome f-up 5- Individualized
Usher Type III **Aller, 2004. Friedman et al 2011.** Prevalence 6% of all Usher cases.	Y: Type III often born with normal hearing. Onset of SNHL may be as early as 3-5 or in adolescence or late childhood. Continues to progress to severe and profound in 4-5th decade of life.	Typically diagnosed as a result of SNHL after age 3	3
Van Buchem Syndrome: skull otosclerosis facial changes over time Two types: Type I (Van Buchem's disease) progressive form for lifetime; Type II (Worth disease) the pathologic bone deposition stops at 20 years of age. The disease is incurable; surgical treatment aims to reduce the intracranial pressure and to correct bones deformity.	Onset of mixed and SNHL around age 15	Variable, often in late childhood	5 - Individualized
Waardenburg Syndrome: three types (I, II and III)white forelock, heterochromia of irises. Rehm, 2008. Toriello, 2011 Prevalence 1:42,000	N: Type I: SNHL can be BL or UL. Typically congenital hearing loss can range from normal to severe SNHL. Type II: ***Can be progressive (70%). 5% also present with CLP & associated OM. Type III: least likely to have hearing loss	Typically at or near birth	1

APPENDIX D. REFERENCES

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