Expanded Hearing Screening in Ontario

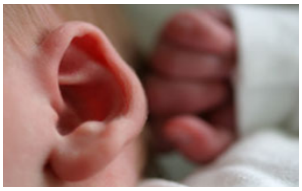
AN INTRODUCTION FOR COMMUNITY PEDIATRICIANS

Newborn Screening Ontario (NSO)

- Newborn Screening Ontario is the provincial program that coordinates dried blood spot testing for a variety of rare but treatable diseases.
- NSO also provides support regarding the standardization, quality assurance, and accessibility of point of care screening for Critical Congenital Heart Disease for infants in Ontario.

NEWBORN SCREENING **ONTARIO**
DÉPISTAGE NÉONATAL **ONTARIO**

The Infant Hearing Program (IHP)



- The Infant Hearing Program is a provincial program that identifies infants with permanent hearing loss and provides services to these children and their families to support language and early literacy development so they are ready to start school.
- IHP services also include monitoring for those infants born at risk of early childhood hearing loss.

Ontario
MINISTRY OF CHILDREN, COMMUNITY AND SOCIAL SERVICES

NSO and the IHP have partnered to offer **expanded hearing screening**, which will improve the detection of infants at risk for childhood hearing loss and introduce an etiologic focus to hearing loss identification.

Expanded Hearing Screening

Newborn Blood Spot Screening

Testing for rare, treatable diseases:

- Metabolic Diseases
- Endocrine Diseases
- Sickle Cell Disease
- Cystic Fibrosis
- Severe Combined Immune Deficiency

Hearing Screening

Audiometric screening

PASS
No further follow-up unless dried blood spot tests positive

REFER
Specialized audiology testing and/or surveillance

NEW

Parental consent obtained by IHP to test dried blood spot

NSO tests for hearing loss risk factors (July 2019)

1. Cytomegalovirus (CMV)

SCREEN POSITIVE

- CMV detected
- High likelihood of congenital CMV infection
- Evaluation including confirmation of infection by a pediatrician required
- If symptomatic (see below), urgent referral to Infectious Diseases Clinic
- Hearing status to be established by the IHP and surveillance as needed

SCREEN NEGATIVE

- CMV not detected
- Baby unlikely to have congenital CMV but does not rule out the possibility (very specific, ~80% sensitive)
- If clinical suspicion remains, diagnostic testing recommended

2. Genetic factors

- Testing of genes commonly associated with non-syndromic hearing loss (GJB2, GJB6 and SLC26A4)
- Only common mutations associated with a high risk for congenital hearing loss will be tested
- Most infants will have hearing loss at birth and identified by audiometric screening
- This etiologic information will allow for earlier connections with ENT and genetic counselling

Congenital Cytomegalovirus (cCMV)

cCMV signs and symptoms:

- petechiae
- jaundice
- hepatosplenomegaly
- microcephaly
- IUGR
- chorioretinitis/optic atrophy
- seizures
- intracranial calcifications
- permanent sequelae can include hearing loss, intellectual and motor disabilities, seizures, and/or vision loss

Note: the majority of infants with cCMV (~85%) will be asymptomatic.

Proposed initial evaluation of CMV screen positive infants by pediatrician:

Urine PCR
to confirm CMV infeciton

Physical exam
to look for petechiae, jaundice, hepatosplenomegaly, microcephaly, IUGR, abnormal neurologic exam

Diagnostic audiology
arranged by the Infant Hearing Program

Laboratory investigations
CBC, LFTs, bilirubin to look for thrombocytopenia, hepatitis, conjugated hyperbilirubinemia

Head ultrasound
to look for intracranial calcifications, white matter changes, microgyria, cysts

Opthalmologic assessment (if possible)
to look chorioretinitis, optic atrophy, or other ocular abnormalities

If symptomatic, referral to be made by pediatrician to Infectious Diseases Clinic for further assessment and treatment decision-making (within 1 month). Shared care between the pediatrician and ID specialist may be considered for ongoing management and monitoring.

cCMV and hearing loss:

cCMV accounts for up to **25%** of hearing loss in children

An infant with cCMV can pass their hearing screen at birth and still be at risk to develop CMV-related hearing loss in childhood.

Risk for hearing loss:

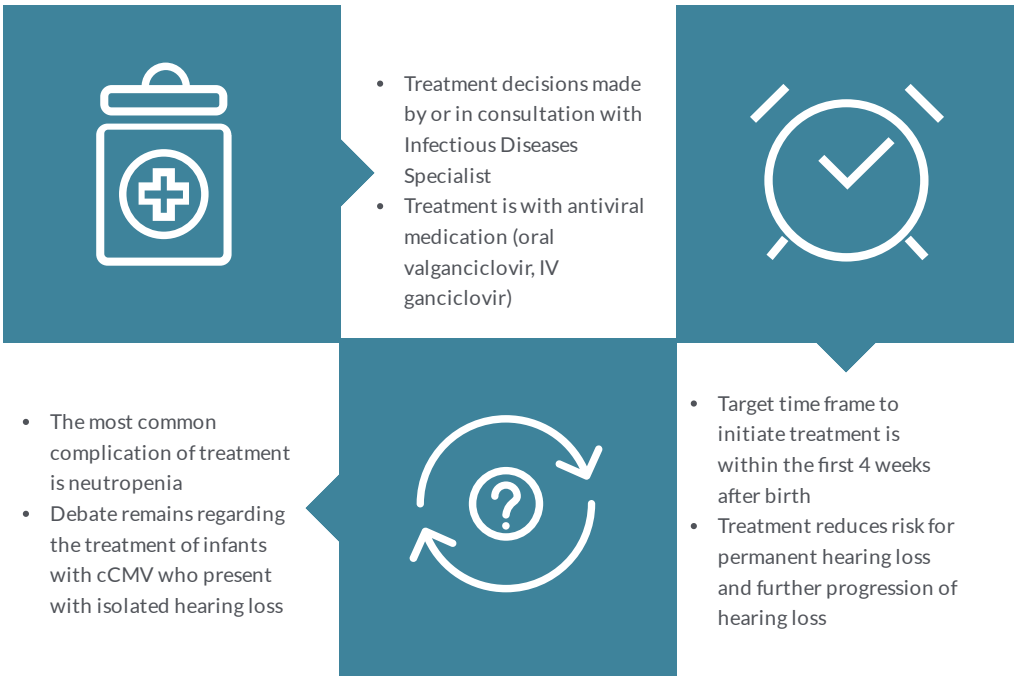
cCMV associated hearing loss can:

- be congenital
- affect one or both ears
- affect some or all pitches
- be mild to severe
- be progressive and fluctuating

Developmental surveillance by pediatrician:

- 3 times a year until the age of 6 years facilitated by standardized parent reported tools (ASQ and Looksee Checklist). Cost of the tools will be covered by NSO.

Treatment for cCMV



- Treatment decisions made by or in consultation with Infectious Diseases Specialist
- Treatment is with antiviral medication (oral valganciclovir, IV ganciclovir)

- Target time frame to initiate treatment is within the first 4 weeks after birth
- Treatment reduces risk for permanent hearing loss and further progression of hearing loss

- The most common complication of treatment is neutropenia
- Debate remains regarding the treatment of infants with cCMV who present with isolated hearing loss