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
- 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.



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.



MINISTRY OF CHILDREN, COMMUNITY AND SOCIAL SERVICES

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

Hearing Loss Risk Factor Screening



Newborn Blood Spot Screening

Testing for rare, treatable diseases:

- Metabolic Diseases
- **Endocrine Diseases**
- Sickle Cell Disease
- Cystic Fibrosis
- Severe Combined Immune Deficiency
- Spinal Muscular Atrophy





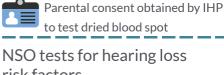




Risk factor dependent surveillance

Specialized audiology testing and/or surveillance

Consent process waived



risk factors





Evaluation including confirmation of

infection by a pediatrician required

to Infectious Diseases Clinic

and surveillance as needed

High likelihood of congenital CMV infection

If symptomatic (see below), urgent referral

Hearing status to be established by the IHP

CMV detected





- 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

- Testing of genes commonly associated with nonsyndromic hearing loss (GJB2, GJB6 and SLC26A4)
- Only common variants 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
- iaundice
- 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:

to confirm CMV infeciton Physical exam to look for petechiae, hepatosplenomegaly, microcephaly, IUGR, abnormal neurologic

Urine PCR

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

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:



asymptomatic

CMV



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:

Facilitated by standardized parent reported tool (ASQ). Cost of the tool will be covered by NSO.

Treatment for cCMV



by or in consultation with Infectious Diseases Specialist Treatment is with antiviral

Treatment decisions made

medication (oral valganciclovir, IV ganciclovir)



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



