Newborn Screening Ontario/Ontario Infant Hearing Program
Screen Negative Risk Factor Results for Permanent Hearing Loss: Information for Health Care Providers

Infants who have physiologic hearing screening through the Ministry of Children, Community and Social Services Infant Hearing Program (IHP) in Ontario are offered the addition of screening for common risk factors associated with permanent hearing loss (PHL). Specifically, risk factor screening for congenital cytomegalovirus (cCMV) and selected mutations (pathogenic variants) in three genes associated with congenital or childhood-onset PHL can be done. Risk factor screening is performed by Newborn Screening Ontario (NSO) using the same sample collected by the hospital or midwife for newborn blood spot screening. Parent/guardian consent for risk factor screening occurs at the time of IHP hearing screening or booking of the IHP hearing screening appointment. Due to the COVID-19pandemic, all dried blood spot samples from babies born on or after March 26, 2020 will be screened for CMV and genetic risk factors for PHL without the need for additional consent by the IHP.

What do negative risk factor screening results for PHL mean?
Negative screening results mean that:

- cCMV was **not** detected
- The common mutations tested in the GJB2, GJB6, and SLC26A4 genes were **not** detected
    - Negative genetic risk factor screening results do not provide information regarding a child's carrier status for the mutations tested. Carrier results are available by request from NSO.

What are the limitations of the risk factor screen for PHL?

- The risk factor screen cannot rule out cCMV infection as the sensitivity of the test is approximately 80%
    - An infant could have a cCMV infection that is not detected through the risk factor screen (i.e. false negative)
    - If an infant has concerns suggestive of possible cCMV infection, diagnostic testing should be considered
    - Please contact NSO about any infant in your care who has a suspected or confirmed cCMV infection and negative risk factor screening results
- The risk factor screen looks for some, but not all, mutations in the GJB2, GJB6 and SLC26A4 genes
    - An infant may have other mutations in these genes that are not detected through the risk factor screen
    - If an infant is at high risk of childhood-onset PHL based on family history, a referral to your local Genetics clinic should be considered
    - A list of mutations included on the risk factor screen can be found on NSO's website at www.newbornscreening.on.ca

How should negative risk factor screening results be interpreted?
Screen negative results should be interpreted along with the results of a child's IHP hearing screen and/or audiology assessment. Please contact your local IHP Lead Agency to request results of a child's hearing screen or audiology assessment. Contact information for IHP Lead Agencies can be found at www.ontario.ca/infanthearing

- A child who passes IHP hearing screening and has a negative result on their risk factor screen does not require any further follow-up, unless additional risk factors were identified by the IHP. If any concerns arise in the future regarding a child's hearing, parents/guardians should follow-up with their child's health care provider.
- A child who has received a refer result on IHP hearing screening and has a negative result on their risk factor screen should continue to their next step in the IHP screening pathway (e.g. community screen or audiology assessment).
- A child who has not had their hearing screened – parents/guardians should follow-up with the Infant Hearing Program in their region for recommendations regarding hearing screening. Negative risk factor screening results should be interpreted with caution and do not rule out the possibility of hearing loss or risk for hearing loss.
- A child with confirmed PHL will be offered services and support through the IHP. A negative risk factor screening result means that the screen did not identify a cause for the child's hearing loss.
    - Referrals to Otolaryngology and Genetics, and additional investigations to identify the potential etiology of a child's hearing loss should be considered.
    - NSO can provide additional interpretation, and potentially further testing, for children who have PHL and negative risk factor screening results. Please contact NSO for more details.

For more information, please visit www.newbornscreening.on.ca.

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