Understanding "Screen Negative" Results for Permanent Hearing Loss Risk Factors

Information for Health Care Providers

What is risk factor screening for permanent hearing loss?

Ontario babies who participate in hearing screening through the **Infant Hearing Program (IHP)** are also screened for common risk factors linked to permanent hearing loss (PHL). This risk factor screening is performed by **Newborn Screening Ontario (NSO)** and uses the same dried blood spot sample that is collected by the hospital or midwife for routine newborn screening.

The specific risk factors screened include:

- **Congenital cytomegalovirus (cCMV)** – a common virus that can affect hearing in babies and children
- **Genetic risk factors** – common pathogenic variants in two specific genes (*GJB2/6* and *SLC26A4*) that are linked to PHL in babies and children

What do "screen negative" risk factor screening results for PHL mean?

Negative screening results mean that:

- Cytomegalovirus (CMV) was **not** detected
- Genetic screening for common variants in the *GJB2/6* and *SLC26A4* genes identified either one or no variants

What are the limitations of this screening?

- The risk factor screen does not detect all cases of cCMV. A baby could still have cCMV even if the screening result is negative.
    - If cCMV is suspected – diagnostic testing should be ordered (e.g. urine CMV PCR)
    - Contact NSO if a baby has confirmed cCMV infection but a negative risk factor screen.
- The genetic screening performed is **not** comprehensive. It only looks for **some** common pathogenic variants in two genes associated with PHL. A baby could have PHL related to these or other genes even if the screening result is negative. Furthermore, the screening will not tell you if a baby is a carrier of *GJB2/6* or *SLC26A4*-related PHL.
    - If there is a family history of PHL, consider referring to your local Genetics Clinic.
    - A list of the pathogenic variants included on the risk factor screen can be found on the NSO website.

How should "screen negative" risk factor screening results be interpreted?

Results should be considered **alongside the baby's physiological hearing screening results** from the IHP, as follows:

- **Physiologic hearing screen "PASS" result:** No further follow-up is needed unless other risk factors are present. If concerns about hearing come up later, parents should talk to their health care provider to arrange a hearing test.
- **Physiologic hearing screen "REFER" result:** Continue with the recommended next steps in the IHP, which could be an appointment for another screen, or an audiological assessment. A negative risk factor screen does not rule out the possibility that the baby could have or be at risk for PHL.
- **Physiologic hearing screening not yet performed:** Parents should be encouraged to complete hearing screening and can contact their local Infant Hearing Program for more information or to book an appointment. A negative risk factor screen does not rule out the possibility that the baby could have or be at risk for PHL.

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- **Permanent hearing loss has been confirmed:** Interventions and support will be offered through the IHP. A negative risk factor screen means the cause of the hearing loss wasn't found through this test. Consider referrals to Otolaryngology and Genetics for further investigation.

Need more information?

Visit newbornscreening.on.ca or ontario.ca/infanthearing

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