Ontario Infant Hearing Program and Newborn Screening Ontario

INFORMATION FOR PARENTS/GUARDIANS:
My Baby Has a Positive Genetic Risk Factor Screening Result

Risk factor screening for permanent hearing loss (PHL) was done on the dried blood sample that the hospital or midwife collected for your baby's newborn (heel prick) screen. This information sheet reviews what it means to have a positive screening result and the follow-up that is recommended for your baby.

Why are babies screened for permanent hearing loss (PHL)?
Hearing screening helps find babies who have PHL or have a higher chance of developing it in early childhood. Finding hearing loss as early as possible allows children with PHL to get the support and services they need to learn language.

What are risk factors for permanent hearing loss (PHL)?
There are many different factors that can increase a baby's chance to have PHL. Some risk factors are genetic (caused by changes in genes) and some are environmental, such as exposure to certain infections or medications.

Your baby was tested for some of the most common genetic risk factors for PHL as well as for the presence of Cytomegalovirus (CMV) infection at birth (called *congenital* CMV). Babies with these risk factors have a higher chance of having or developing PHL.

What genetic risk factors was my baby tested for?
More than half of all childhood hearing loss has a genetic cause and many different genes are known to be related to hearing loss.

Your baby was tested for some of the most common genetic differences (also called mutations) in the GJB2, GJB6 and SLC26A4 genes that can cause hearing loss. A mutation is a difference in a gene that either makes the gene not work, or causes the gene to work differently. These three genes all provide instructions to make proteins that are important to the development of the inner ear or cochlea. The SLC26A4 gene is also involved in the development of the thyroid (a gland found in the neck region which makes important hormones the body needs).

What does a "positive" genetic risk factor result mean?
A positive genetic risk factor screening result means that your baby has a high chance of having PHL. Your baby should have a hearing (audiology) assessment with an Infant Hearing Program (IHP) audiologist to find out if your baby has PHL.

What will happen at the hearing (audiology) assessment?
An IHP audiologist will do several different tests to check your baby's hearing. These tests will help determine if there is any hearing loss, and if there is:

- What sounds the hearing loss is impacting
- How much hearing loss there is
- If the hearing loss is permanent or not

You will be with your baby during the whole appointment. The hearing tests do not hurt. In fact, babies are asleep during the assessment as that is the only way to get a complete test. The assessment can take up to 2 hours from start to finish.

Do I need to do anything to prepare my baby for the hearing assessment?
Try to keep your baby awake just before, and when travelling to, the audiology appointment. You will have a chance to feed your baby before the testing starts in a comfortable, quiet room with low lighting. Do not use any lotions or creams on your baby's forehead on the day of the appointment. Please bring any special blanket, pacifier, or other soother your child uses when falling asleep.

What happens after the hearing assessment?
The IHP audiologist will explain your baby's results to you after all the tests are done. The next steps will depend on the outcome of the assessment.

- **If your baby has PHL**
  - If PHL is confirmed in your baby, the IHP audiologist will give you more information about the hearing loss and what services and supports IHP offers to help your baby.
  - Babies who are newly found to have PHL usually meet with an Ear, Nose, Throat (ENT) doctor (also called an otolaryngologist) for further assessment.
- **If your baby does not have PHL**
  - Some babies with a positive genetic risk factor screening result will not have PHL identified at their initial hearing assessment.
  - Your baby still has a chance to develop PHL.
  - Your baby will have their hearing checked more often over the next few years so that if PHL develops, it is found as early as possible.
  - A small number of babies with a positive genetic risk factor screening result will not have PHL.
- **If your baby's hearing assessment is incomplete**
  - Rarely, the IHP audiologist is not able to do all of the necessary tests during the appointment to tell whether or not a hearing loss is present in your baby. If this happens, the IHP audiologist will talk with you about next steps.

How does permanent hearing loss (PHL) happen in families?
Hearing loss can happen in families in different ways, depending on the genes involved. The genetic risk factor screen specifically looks for PHL caused by 3 genes: GJB2, GJB6 and SLC26A4. PHL caused by differences in these genes is called "recessive". This means a child must have two non-working copies of the same or related gene to have, or be at a higher risk to develop, PHL.

Your baby was found to have two genetic risk factors for PHL, which means that each parent is a "carrier" of a genetic risk factor for PHL. This means they have **only one** non-working copy of the hearing loss gene. Carriers are <u>not</u> at high risk for PHL. In fact, most babies with a genetic form of PHL are born to parents who can hear, and there is often no one else in the family with PHL.

What do these results mean for my family?
When both parents are carriers for a genetic risk factor for PHL, there is a 25% chance in **each** pregnancy for the baby to have two non-working copies of the gene and a high risk to have PHL. There is also a 50% chance for a baby to be a carrier (just like the parents), and a 25% chance for the baby to have two working copies of the gene (i.e. low risk for PHL, not a carrier).

If you have more questions about what a positive genetic risk factor screening result means for you, your child/family, or a future pregnancy, you can discuss this with a Genetic Counsellor at Newborn Screening Ontario or ask your doctor for a referral to your local Genetics Clinic.

Where can I get more information?
For more information about hearing screening and/or risk factor screening for PHL, talk to your health care provider or visit the following websites:

Ontario Infant Hearing Program: www.ontario.ca/infanthearing
Newborn Screening Ontario: www.newbornscreening.on.ca

NOTE TO PARENTS/GUARDIANS: This information is for parents whose baby has had a positive genetic risk factor screening result through Newborn Screening Ontario and the Ontario Infant Hearing Program. This fact sheet was written for information purposes only and should not replace professional medical or audiologic advice, diagnosis, or treatment.

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