

## Connexin 26 GenotypR™ INFORMED CONSENT

**1. What causes hearing loss?** Normal hearing requires that all parts of the auditory pathway are working correctly. The exact location and nature of the problem in the auditory pathway determines the type and severity of a person's hearing loss. Some causes of hearing loss occur before a baby is born. These include genetic disorders and infections (such as congenital rubella or congenital syphilis). About half of all cases of hearing loss among children are thought to result from genetic factors. Sometimes these children have a syndrome of which hearing loss is only one feature. However, in most children with hearing loss that is due to a genetic cause, the hearing loss is not part of a syndrome. Mutations in the *GJB2* gene are responsible for much of the hearing loss in this latter group of children. *GJB2* encodes a protein, Connexin 26, which plays an important role in the functioning of the cochlea (the part of the ear that changes sound into nerve signals that travel to the brain).

**2. What is the purpose of this test and what are its limitations?** This test detects the presence of two mutations in the *GJB2* gene: 35delG and 167delT. Everyone has two copies of the *GJB2* gene; an individual may have two normal copies (unaffected non-carrier), two abnormal copies (affected), or one normal and one abnormal (carrier). The 35delG mutation is responsible for 10% of all deafness and 20% of all hereditary hearing loss; the 167delT is seen mostly in people of Ashkenazi Jewish population, with a frequency of 5%. If these mutations are not found by the testing procedure, it does not mean that the risk of carrying or developing hearing loss is not present. It simply means that these specific mutations have not been found, although other mutations may be present.

**3. What is required to perform this test?** You will be asked to provide 5 mL of blood, which is equal to about one tablespoon. DNA will be extracted from this blood sample and tested. The only discomfort that you may feel is the stick of the needle in your arm. You may also experience a small bruise at the site of the needle puncture. You will also be asked to provide information regarding your medical history, which is necessary for proper interpretation of your test result. In the unlikely event that you should be injured in the course of being tested, your physician will provide any necessary medical care. However, you would be expected to bear the cost of such medical care. Compensation will not be provided in the event of any injury.

**4. Is there a cost for this test?** This is a routine clinical laboratory test and the results may aid in your diagnosis; thus, you or your health insurer will be billed for this procedure.

**5. What will happen to the DNA once the test is complete?** The original blood sample will be discarded at the end of the testing process or stored not more than 60 days. The DNA will be retained for a minimum of 6 months. In some circumstances, a patient's DNA may be used anonymously as a negative or positive control sample in future testing, but, in this circumstance, all identifiers will be removed prior to re-testing and the DNA sample and results obtained will remain anonymous.

I understand and agree that my DNA remaining after testing may be stored for up to 6 months should additional testing be required. *Please initial.*

**6. How will I obtain results from this test?** DNA testing and interpretation of results are complex. The information from this test will be provided in the form of a written report to your physician who will inform you of the results. The laboratory will not provide results directly to patients. Patient may have genetic counseling prior to signing the consent form. To the extent permitted by law, all of your laboratory records and results are confidential and shall not be disclosed without your written authorization.

### Patient Attestation of Informed Consent:

My signature below indicates that I have received information about this test, **Connexin 26 GenotypR™**, and that I have read and understood the material in this document. I have been given a full opportunity to ask questions that I may have about the testing procedure and related issues. I agree to undergo this testing.

\_\_\_\_\_  
Patient Signature

\_\_\_\_\_  
Date

\_\_\_\_\_  
Signature of Parent/Guardian if Patient is a minor

\_\_\_\_\_  
Print Name of Parent/Guardian

### For the Physician:

As the referring physician, I understand the benefits and limitations of this study and have requested that the above-named patient be tested. I attest to the fact that I have provided the patient with the information contained above and fully answered any questions. I believe that the patient understands the information and is voluntarily signing this informed consent.

\_\_\_\_\_  
Signature of Physician/Health Care Professional

\_\_\_\_\_  
Print Name of Physician/Health Care Professional