In the past, persons diagnosed with hereditary hearing loss often required multiple tests to determine the exact cause of their hearing loss. These tests could be expensive, inconvenient, and time consuming. The University of Iowa offers OtoSCOPE® - one test that is:

- **Comprehensive** – tests for 152 genes known to cause non-syndromic hearing loss and non-syndromic mimics such as Usher syndrome.
- **Pinpointed** – offers the best method of diagnosis for genomic level evaluation.
- **Easy** – patient provides one blood sample.
- **Convenient** – blood sample is taken in local doctor’s office.
- **Accurate** – 99 percent diagnostic specificity.
- **Enhancing diagnosis** – potentially providing prognosis, testing direction, and information for education and counseling.
- **Personalized** – all results are discussed at a multidisciplinary meeting which includes clinical experts, scientists, bioinformaticians, human geneticists and genetic counselors.
- **Collaborative** – MORL will work directly with your physician or genetic counselor to ensure that test results are interpreted appropriately.
- **Less expensive and more efficient** – one test versus many tests.

After an audiogram, comprehensive genetic testing is the single best test to order in the evaluation of a person with hearing loss.

---

**Molecular Otolaryngology and Renal Research Laboratories**

**Director:**
Richard J.H. Smith, M.D.

**Contact:**
Amy Weaver, Project Assistant
morl@uiowa.edu

**Website:**
http://www.medicine.uiowa.edu/morl
**BACKGROUND**

*Hearing loss is the most common sensory deficit.*

Hearing loss affects 360 million people worldwide. It is often due to genetic factors that result in damage to the inner ear. In babies, most hearing loss is due to genetic causes, and in many adults, genetic factors can be identified as well.

When genetic hearing loss is not associated with other features, it is called non-syndromic hearing loss (NSHL).

There are also over 400 syndromic causes of hearing loss (hearing loss associated with other features). Some of these syndromes resemble NSHL in infants and children; we call these “NSHL mimics.” Because there are many genes that cause NSHL and syndromic hearing loss, genetic diagnosis is difficult using conventional methods. For this reason, the Molecular Otology and Renal Research Laboratories (MORL) at the University of Iowa developed OtoSCOPE*.

**GENETIC TESTING**

Determining the cause of hearing loss by genetic testing provides important answers to many questions. Information can be given on recurrence chance for future children, prognosis (whether hearing loss will worsen over time) can be provided, courses of action can be identified (such as further testing, regular ophthalmological visits, cochlear implants or hearing aids), and results can provide peace of mind to patients and their families.

*Our lab will work directly with your physician or genetic counselor to ensure that test results are interpreted appropriately.*

**METHOD OF TESTING**

OtoSCOPE* uses custom-targeted sequence capture for DNA enrichment followed by massively parallel DNA sequencing. In this way, all genes known to cause NSHL and non-syndromic mimics are sequenced at the same time. Data analysis to examine possible causative variants using our custom variant pipeline is followed by confirmation by traditional Sanger sequencing. This comprehensive approach decreases cost and turn around time for test results.

OtoSCOPE* has a diagnostic sensitivity and specificity of greater than 99% (Shearer et al., Proc Natl Acad Sci USA, 107(49), 2010).

**CLINICAL INDICATIONS**

**NON-SYNDROMIC DEAFNESS:**
- Autosomal Dominant
- Autosomal Recessive
- Auditory Neuropathy Spectrum Disorder
- Branchiootorenal (BOR) Syndrome
- Chudley-McCullough Syndrome
- Deafness Infertility Syndrome
- Heimler syndrome
- High Myopia and Hearing Loss
- Hypoparathyroidism, Deafness and Renal Dysplasia Syndrome
- Jervell and Lange Nielsen Syndrome
- Mitochondrial Encephalomyopathy, Lactic Acidosis, and

**SYNDROMIC DEAFNESS:**
- Alport Syndrome
- Alström Syndrome
- Auditory Neuropathy with Progressive Peripheral Sensory Neuropathy
- Mohr–Tranebjaerg syndrome
- Pendred Syndrome
- Perraault Syndrome
- Pfeiffer syndrome
- Renal Tubular Acidosis with Deafness
- Sinoatrial Node Dysfunction and Deafness
- Stickler Syndrome
- Treacher Collins Syndrome
- Usher Syndrome
- Velocardiofacial Syndrome
- Waardenburg Syndrome
- Wolfram Syndrome

Our full gene list is available at: www.medicine.uiowa.edu/morl/otoscopegenes

**Is OtoSCOPE* right for you?** Use the diagnostic guide above to determine the likelihood of receiving a positive diagnosis (%). For more, see Sloan-Heggen et al., Hum Genet, 135(4), 2016.

A single genetic test for hearing loss.