

# Hearing Loss mtDNA Panel

**Disorder:** Hearing loss affects about 1 in 500 newborns and a genetic etiology is suspected in two thirds of these patients. Hearing loss can be caused by mutations in many different genes which can be inherited in an autosomal dominant, autosomal recessive, X-linked or mitochondrial (maternal inheritance) manner.

Mutations in the *MTTS1* and *MTRNR1* mitochondrial genes account for approximately 1% of nonsyndromic sensorineural hearing loss in the United States.

Aminoglycoside exposure increases the risk and severity of hearing loss in carriers of *MTRNR1* mutations. Mutations in *MTTL1* and *MTTK* are associated with MELAS and MERRF which usually present with neurological symptoms. The 3243 mutation in *MTTL1* has also been associated with Maternally inherited diabetes mellitus and deafness (MIDD).

## Indications:

- Nonsyndromic hearing loss of unknown etiology
- Hearing loss in a patient with aminoglycoside exposure (or planned aminoglycoside exposure)
- Hearing loss in a patient with a family history suggestive of maternal transmission of hearing loss
- Carrier testing in a relative of a patient with a proven mitochondrial mutation

**Specimen:** At least 2 mLs whole blood in lavender top (EDTA) tube. Alternately, two cytobrushes (cheek swabs) may be collected. (Please call 1-513-636-4474 for a free cytobrush collection kit). Label each tube or brush with patient's name, birth date, and date of collection.

**Testing Methodology:** Mitochondrial specific DNA is amplified by PCR and subjected to direct sequence analysis for the following mutations: *MTRNR1* (961,1555,1494), *MTTS1* (7445, 7511), *MTTL1* (3242, 3271), and *MTTK* (8344).

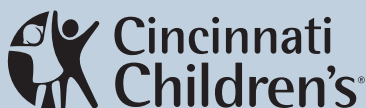
Analysis of these mitochondrial mutations is also offered as part of our **Hearing Loss Panel Tier 1** which includes analyses for additional genes that contribute to hereditary hearing loss. Please refer to our website for additional information.

**Test Sensitivity:** Approximately 70% of the mitochondrial mutations related to nonsyndromic hearing loss are detected by this targeted mutation panel.

Mutations in *MTTS1* and *MTRNR1* account for approximately 1% of nonsyndromic hearing loss in the United States.

**Turn-Around Time:** 28 days

**Cost:** Please call 1-866-450-4198 for current pricing, insurance precertification, or with any billing questions.



Human Genetics

Molecular Genetics Laboratory

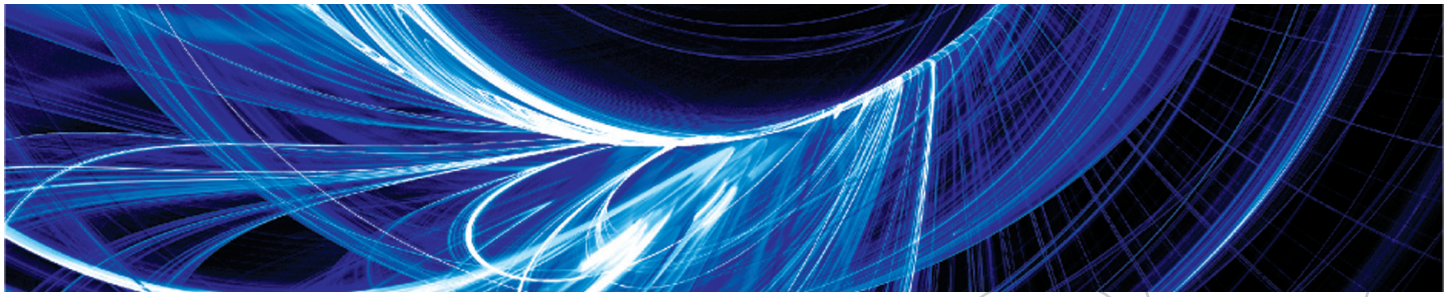
CLIA#: 36D0656333

Phone: (513) 636-4474

Fax: (513) 636-4373

Email: [moleculargenetics@cchmc.org](mailto:moleculargenetics@cchmc.org)

[www.cchmc.org/hearing-loss](http://www.cchmc.org/hearing-loss)



## CPT Codes:

### Analysis of 8 specific mitochondrial mutations

81401(x3)

### Family specific mutation analysis

81403

**Results:** Each test report includes a detailed interpretation of the genetic findings, the clinical significance of the result, and specific recommendations for clinical management and additional testing, if warranted. Results will be reported to the referring physician or health care provider as specified on the test requisition form.

## References:

Fischel-Ghodsian, N. (1999). "Mitochondrial deafness mutations reviewed." *Hum Mutat* 13(4): 261-270.

Guan, M. X. (2011). "Mitochondrial 12S rRNA mutations associated with aminoglycoside ototoxicity." *Mitochondrion* 11(2): 237-245.

Jacobs, H. T., T. P. Hutchin, et al. (2005). "Mitochondrial DNA mutations in patients with postlingual, nonsyndromic hearing impairment." *Eur J Hum Genet* 13(1): 26-33.

Kokotas, H., M. B. Petersen, et al. (2007). "Mitochondrial deafness." *Clin Genet* 71(5): 379-391.

Li, X., N. Fischel-Ghodsian, et al. (2004). "Biochemical characterization of the mitochondrial tRNA<sup>Ser</sup>(UCN) T7511C mutation associated with nonsyndromic deafness." *Nucleic Acids Res* 32(3): 867-877.

Tang, H. Y., E. Hutcheson, et al. (2002). "Genetic susceptibility to aminoglycoside ototoxicity: how many are at risk?" *Genet Med* 4(5): 336-345.

Zhao, H., R. Li, et al. (2004). "Maternally inherited aminoglycoside-induced and nonsyndromic deafness is associated with the novel C1494T mutation in the mitochondrial 12S rRNA gene in a large Chinese family." *Am J Hum Genet* 74(1): 139-152.

Additional information and test requisitions are available at: [www.cchmc.org/hearing-loss](http://www.cchmc.org/hearing-loss)

## Shipping Instructions

Please enclose a completed **test requisition, audiogram and MRI/CT report, if available** with the sample. **All information must be completed before the sample can be processed.** Place samples in Styrofoam mailer and ship at room temperature by overnight Federal Express to arrive Monday through Friday.

## Ship to:

Cytogenetics and Molecular Genetics Laboratories  
3333 Burnet Avenue NRB 1042  
Cincinnati, OH 45229  
513-636-4474