

The University of Chicago Genetic Services Laboratories



5841 S. Maryland Ave., Rm. L035, MC 0077, Chicago, Illinois 60637
Toll Free: (888) UC GENES ♦ (888) 824 3637
Local: (773) 834 0555 ♦ FAX: (773) 834 0556
ucgslabs@genetics.uchicago.edu ♦ www.genes.uchicago.edu
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Connexin 26/GJB2 Sequencing

Thank you for your inquiry regarding *Connexin26/GJB2* sequence analysis.

Approximately 40% of prelingual, sensorineural deafness is non-syndromic and autosomal recessive. Of cases of autosomal recessive, non-syndromic hearing loss, 50% are due to mutations of the *Connexin 26* gene^{1,2}, and a single mutation, 35delG, accounts for approximately 70% of *Connexin 26* mutations³. Identification of *Connexin 26* mutations in affected children will allow for better genetic and prognostic counseling for their families by establishing a specific diagnosis and providing information about recurrence risks.

Our laboratory offers analysis for the common 35delG mutation of the *Connexin 26* gene and full gene analysis for those who are not homozygous for this mutation. In addition, samples from individuals heterozygous for a mutation in the gene will be analyzed for the 342 kb deletion, involving the *Connexin 30* gene. Mutations in these genes are known to interact and cause deafness. Sample submission paperwork and instructions are included with this fax.

Sample specifications:	5 cc of blood in a lavender top/EDTA tube
Turn-around-time:	4-6_weeks
Cost:	\$ 430
CPT codes:	83891, 83898, 83904, 83894, 83912

* Once a mutation is identified, other family members can be tested for the same mutation for a fee of \$390 per blood sample (CPT codes: 83891, 83898 x 2, 83894, 83912), and prenatal samples can be tested for a fee of \$540 per sample (CPT codes: 83891, 83898 x 2, 83894, 83912, 88235-52).

Please contact UCGS personnel if you wish to discuss the specifics of your case or if you have any questions.

1. Green GE, et al. Carrier rates in the midwestern United States for *GJB2* mutations causing inherited deafness. (1999) JAMA, 281:2211-2216.
2. Denoyelle F, et al. Clinical features of the prevalent form of childhood deafness, DFNB1, due to a connexin-26 gene defect: Implications for genetic counselling. (1999) 353:1298-1303.
3. Denoyelle F, et al. Prelingual deafness: High prevalence of a 30delG mutation in the connexin 26 gene. (1997) 6:2173-2177.
4. del Castillo I, et al. A deletion involving the Connexin 30 gene in nonsyndromic hearing impairment. NEJM (2002) 346:243-249.