Test Name & Code:

- **Test Name:** CONNEXIN26 Mutations
- **Test Code:** 3181

Related Diseases & Genes:

- Bart-Pumphrey syndrome (OMIM #: 149200) || Gene: *GJB2* (OMIM #: 121011)
- “Deafness, autosomal dominant 3A” (OMIM #: 601544)
- “Deafness, autosomal recessive 1A” (OMIM #: 220290)
- Hystrix-like ichthyosis with deafness (OMIM #: 602540)

Synonyms:

- Congenital Deafness
- GJB2 Mutations
- CX26 Mutations
- DFNB1 Mutations
- DFNA3A Mutations
- DFNB1A Mutations

Test Details:

- **Methodology:** DNA Sequencing
- **Performed:** Everyday
- **Reported:** 2-3 weeks

Specimen Details:

- **Specimen Collection:** Peripheral blood
- **Container:** Tube with EDTA
- **Specimen Volume/Amount:** > 3 ml
- **Storage/Transport Condition:** Refrigerated
- **Unacceptable Specimens/Conditions:** Serum; Frozen specimens; Hemolyzed specimens; Icterus; Lipemia

Testing Area(s):

- Dsymorphology

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For yesterday, today and *tomorrows...*