

## Spondylocostal Dysostosis Testing via *LFNG* Gene Sequencing (Test #422)

**Brief Description of Disorder:** Spondylocostal Dysostosis (SCD) (OMIM 277300, 608681, 609813) is characterized by abnormal segmentation of the vertebral column. Patients have short trunks with multiple vertebral defects and rib anomalies (Turnpenny et al. J Med Genet 40:333-339, 2003). An autosomal dominant and three autosomal recessive forms (Types 1-3) of SCD are known. Type 3 is caused by mutations in the *LFNG* gene which encodes an enzyme that catalyzes the addition of N-acetylglucosamine residues to Notch receptor proteins.

**Genetics:** SCD Type 3 is an autosomal recessive disorder. To date, only one SCD patient of Lebanese ancestry has been shown to carry mutations in *LFNG* (Sparrow et al. Am J Hum Genet 78:28-37, 2006). This patient was severely affected and was homozygous for the missense mutation Phe188Leu.

**Description of This Particular Test:** This test involves bidirectional DNA sequencing of the coding regions of all 8 coding exons of *LFNG* plus about 50 bp of flanking non-coding DNA on each side. Our test includes both splicing alternates for coding exon 1.

**Indications for Test:** SCD patients that do not carry *DLL3* mutations are candidates for this test. We offer sequencing of one or two exons in parents and other family members of patients with known mutations. We also offer clinical confirmation of mutations that have been identified in research labs.

**Sensitivity of Test:** Probably < 5% of SCD patients have causative mutations in *LFNG* (Sparrow et al. 2006).

**Turn Around Time:** Maximum of 40 days, although many tests are completed in 2-3 weeks.

**SPECIMEN REQUIREMENTS:** See page 4 of the Requisition Form.

**Sequencing of the 8 coding exons of the *LFNG* gene** **\$490.**

<b>Sample Ascertainment</b>		<b>83890</b>	<b>\$ 30</b>
<b>DNA Isolation</b>		<b>83891</b>	<b>\$ 30</b>
<b>Amplification</b>	<b>x 7</b>	<b>83898</b>	<b>\$130</b>
<b>Mutation Identification by Sequencing</b>	<b>x 7</b>	<b>83904</b>	<b>\$190</b>
<b>Interpretation and Report</b>		<b>83912</b>	<b>\$110</b>

Single exon sequencing for the presence of previously identified mutations in the *LFNG* gene is also available for \$190, or two exon sequencing for \$340.

**Accreditation Info.** CLIA ID #: 52D1027685 (expires 1/18/13) (CAP#: 7185561, AU ID: 1407125 expires 12/20/12)

**Contact for info:** Ying Wang, MD, PhD, [ying.wang@preventiongenetics.com](mailto:ying.wang@preventiongenetics.com), [www.preventiongenetics.com](http://www.preventiongenetics.com)