

Updated Increased Nuchal Translucency Testing Panel

We have updated the *Increased Nuchal Translucency Testing Panel* as of May 1, 2011.

Northwestern Reproductive Genetics is pleased to introduce our updated version of the *Increased Nuchal Translucency Testing Panel*, a prenatal testing option for fetuses with increased nuchal translucency and normal karyotype. Based on our experience with over 270 patients, we have updated the panel to provide the most clinically significant information as quickly and as cost effectively as possible. We are also excited to report that Northwestern Reproductive Genetics will soon be offering an innovative *Skeletal Dysplasia Testing Panel* that may be requested in conjunction with the *Updated Increased Nuchal Translucency Testing Panel*. This panel is expected to be available within the coming months.

The updated panel includes mutation analysis for the following two conditions:

Noonan Syndrome

Prevalence: Approximately 1/1,000-1/2,500

Noonan syndrome (NS) is an autosomal dominant condition characterized by short stature, congenital heart defect, and developmental delay of variable degree. Other findings include broad or webbed neck, unusual chest shape, cryptorchidism, characteristic facies, varied coagulation defects, lymphatic dysplasias, and ocular abnormalities.

The *Updated Increased Nuchal Translucency Testing Panel* uses arrayed primer extension (APEX) technology to test for mutations in all five genes known to be associated with NS, including PTPN11, KRAS, SOS1, RAF1, and MEK1, with a total detection of more than 70%.

Of the first 270 fetuses tested using the Increased Nuchal Translucency Panel, 13 (4.8%) were found to have a mutation in one of these five genes associated with Noonan syndrome. Of the patients who opted to pursue parental testing following an identified mutation in the fetal sample, two fetuses were found to have paternally inherited mutations.

Spinal Muscular Atrophy

Prevalence: Approximately 1/7,000

Spinal muscular atrophy (SMA) is an autosomal recessive condition characterized by progressive muscle weakness resulting from degeneration and loss of anterior horn cells in the spinal cord and brain stem nuclei. Onset ranges from before birth to adolescence or young adulthood. Poor weight gain, sleep difficulties, pneumonia, scoliosis, and joint contractures are common complications.

The *Updated Increased Nuchal Translucency Testing Panel* uses a PCR-based assay to determine the number of SMN1 gene copies as well as tests for 29 point mutations in SMN1, with a combined detection of approximately 95%.

Of the first 270 fetuses tested using the Increased Nuchal Translucency Panel, 18 (6.7%) were found to be carriers for spinal muscular atrophy, while none were expected to be affected with the condition.

Fees:

The self-pay fee for the *Updated Increased Nuchal Translucency Testing Panel* is \$425. The self-pay fee for maternal cell contamination studies is an additional \$300, if requested. Charges will be submitted through the patient's insurance directly, and patient will not be responsible for more than the self-pay fees listed above.

Institutional billing is also available by request. Please contact our billing department for more information.

CPT Codes:

Updated Increased Nuchal Translucency Panel: 83891, 83896, 83898, 83900, 83901, 83904, 83909, 83914, 83912, 99000

Maternal cell contamination studies: 83891, 83898, 83909, 83912

Specimen and Shipping Requirements:**Prenatal Samples Accepted**

We prefer to receive one T-25 flask of confluent fetal cells. We may also be able to accept direct chorionic villi, unspun amniotic fluid, or extracted DNA. Please contact our laboratory if you would like to order testing on one of these types of samples. If maternal cell contamination testing is requested, please also include one purple top tube of maternal blood.

Additional specimen must be held for back-up culture at another facility.

Shipping Requirements

Ship overnight at ambient temperature to arrive Monday-Friday. A completed requisition form, billing form, and consent form must be included with the sample. Shipping costs are the responsibility of the sender.

Ship to:

Northwestern Reproductive Genetics, Inc.

680 N. Lake Shore Drive, Suite 1230

Chicago, IL 60611

Telephone number: 312-981-4400

Turnaround Time

Approximately 3-4 weeks.