

## Skeletal Dysplasia Testing Panel

Northwestern Reproductive Genetics is pleased to introduce our new and innovative *Skeletal Dysplasia Testing Panel*. In a continuing effort to provide prenatal patients with early genetic testing options, we have developed this panel as an option for pregnancies with an increased nuchal translucency in the first trimester and/or ultrasound findings suggestive of a skeletal dysplasia detected during the first or second trimester.

The *Skeletal Dysplasia Testing Panel* uses arrayed primer extension technology in combination with sequencing analysis to test for 378 mutations in eight genes known to be associated with early onset skeletal dysplasias. A summary of the genes tested and the conditions associated with mutations in each gene is included below. At this time, it is not possible to calculate an accurate detection rate for each condition included in this panel. However, the proportion of reported disease-causing mutations for each gene tested is greater than 79% and is as high as 100%, depending on the gene.

### Summary of the mutations included in the *Skeletal Dysplasia Testing Panel*:

Genes tested	Conditions associated with mutations in the gene	Number of mutations included in the panel
<b>FGFR3: 4p16.3</b>	Hypochondroplasia	19
	Thanatophoric dysplasia	11
	Achondroplasia	7
	Craniosynostosis/Muenke syndrome	2
	Acanthosis nigricans	2
	Skeletal dysplasia (undifferentiated)	1
	Crouzon syndrome	1
<b>FGFR2: 10q26</b>	Crouzon syndrome	39
	Pfeiffer syndrome	15
	Craniosynostosis (undifferentiated)	11
	Jackson-Weiss syndrome	6
	Beare Stevenson syndrome	2
	Apert syndrome	4
	Saethre Chotzen syndrome	1
<b>COL2A1: 12q13.11-q13.2</b>	Achondrogenesis type 2	14
	Achondrogenesis/Hypochondrogenesis	2
	Hypochondrogenesis	14
	Spondyloepimetaphyseal dysplasia, Strudwick type	2
	Spondylepiphyseal dysplasia	37
	Spondyloperipheral dysplasia	4
<b>SLC26A2 (DTDST): 5q32-q33.1</b>	Diastrophic dysplasia	21
	Diastrophic dysplasia/multiple epiphyseal dysplasia	1
	Achondrogenesis type 1B	9
	Atelosteogenesis type 2	7
<b>ALPL: 1p36.1-p34</b>	Hypophosphatasia (perinatal type)	86
<b>ROR2: 9q22</b>	Robinow syndrome	11
<b>ESCO2: 8p21.1</b>	Roberts syndrome	22
	SC phocomelia	5
<b>SOX9: 17q24.3-q25.1</b>	Campomelic dysplasia spectrum	22

The *Skeletal Dysplasia Testing Panel* may also be used in conjunction with the updated version of our *Increased Nuchal Translucency Testing Panel* for fetuses with increased nuchal translucency and normal karyotype. A number of studies have recently reported an association between increased nuchal translucency measurements and skeletal dysplasias; however it can be quite difficult to make a specific diagnosis based on ultrasound findings alone, especially early in the pregnancy. The *Skeletal Dysplasia Testing Panel* was designed to assist providers in making a diagnosis prenatally in order to provide patients with a more comprehensive prognosis.

**Fees:**

The self-pay fee for the *Skeletal Dysplasia 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:**

*Skeletal Dysplasia Panel:* 83891, 83894, 83896, 83900, 83901, 83904, 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.

## **References**

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- Khalil A, Pajkrt E, Chitty LS. (2011) Early prenatal diagnosis of skeletal anomalies. *Prenatal Diagnosis* **31**:115-124.
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