

**THIS IS NOT A TEST REQUEST FORM.**  
**The information below is required to perform molecular genetic testing.**  
**Please fill out this form and submit it with the test request form or electronic packing list.**

**PATIENT HISTORY FOR SKELETAL DYSPLASIA TESTING**

Patient Name \_\_\_\_\_ Date of Birth \_\_\_\_/\_\_\_\_/\_\_\_\_ Gender  F  M

Physician \_\_\_\_\_ Physician Phone (\_\_\_\_) \_\_\_\_\_ Practice Specialty \_\_\_\_\_

Genetic Counselor \_\_\_\_\_ Counselor Phone (\_\_\_\_) \_\_\_\_\_

**Patient's Ethnicity** (check all that apply)  
 African American     Ashkenazi Jewish     Asian     Caucasian  
 Hispanic     Middle Eastern     Native American     Other \_\_\_\_\_

**SUSPECTED DIAGNOSIS**

Achondroplasia     Diastrophic Dysplasia     Other \_\_\_\_\_  
 Achondrogenesis     Thanatophoric Dysplasia     Unknown  
 Campomelic dysplasia     Osteogenesis Imperfecta (specify type) \_\_\_\_\_

**Does the patient have SYMPTOMS?**  No  Yes **If yes, please check all of the following that apply:**

Shortening of bones of arms and legs     Undermineralization of bones     Clubfeet  
 Bowed or fractured bones     Abnormal ribs or small chest circumference     Hydrops  
 Irregular, thickened or thin bones     Extra fingers or toes     Other \_\_\_\_\_

**Patient is:**  Living     Deceased; describe circumstances: \_\_\_\_\_

**Does the patient have RADIOGRAPHIC FINDINGS?**  No  Yes  Unknown **If yes, please detail below:**

**Has the patient had PREVIOUS MOLECULAR GENETIC TESTING?**  Yes  No  Unknown

Gene/s \_\_\_\_\_ Methodology \_\_\_\_\_ Results \_\_\_\_\_

**Does the patient have a FAMILY HISTORY of a skeletal dysplasia?**  No  Yes  Unknown

NAME OF THE DISORDER diagnosed in the relatives: \_\_\_\_\_

RELATIONSHIP of the family members to the patient: \_\_\_\_\_

Provide the gene mutation(s) identified in the relative \_\_\_\_\_

**Circle the test you intend to order OR write the test name and number below:**

<b>Prenatal testing for skeletal dysplasias:</b>	
2012010	<b>Skeletal Dysplasia Panel, Sequencing, 39 Genes and Deletion/Duplication, 36 Genes, Fetal</b> - Next generation sequencing and tiled custom CGH on fetal specimens.
2001980	<b>Familial Mutation, Targeted Sequencing, Fetal</b> - Targeted testing for a known familial sequence variants (laboratory report from family member is REQUIRED).
<b>Postnatal testing for skeletal dysplasias:</b>	
2012015	<b>Skeletal Dysplasia Panel, Sequencing, 39 Genes and Deletion/Duplication, 36 Genes</b> - Next generation sequencing and tiled custom CGH.
2001961	<b>Familial Mutation, Targeted Sequencing</b> - targeted testing for known familial sequence variants.

**Other test not listed:** \_\_\_\_\_

**For questions, contact an ARUP genetic counselor at (800) 242-2787, ext. 2141**

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