ARUP LABORATORIES | NATIONAL REFERENCE LABORATORY
A nonprofit enterprise of the University of Utah and its Department of Pathology

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THIS IS NOT A TEST REQUEST FORM.
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 _____ Sex ☐ F ☐ M

Physician _____ Physician Phone _____

Practice Specialty _____ Physician Fax _____

Genetic Counselor _____ Counselor Phone _____

Patient's Ethnicity (check all that apply)

☐ African-American ☐ Asian ☐ Hispanic ☐ Native American

☐ Ashkenazi Jewish ☐ Caucasian ☐ Middle Eastern ☐ Other _____

Suspected diagnosis:

☐ Achondrogenesis ☐ Diastrophic dysplasia ☐ Thanatophoric dysplasia

☐ Achondroplasia ☐ Osteogenesis imperfecta (specify type): ☐ Unknown

☐ Campomelic dysplasia _____ ☐ Other: _____

Does the patient have symptoms? ☐ No ☐ Yes (check all that apply)

☐ Abnormal ribs or small chest ☐ Extra fingers or toes ☐ Shortening of bones of arms and legs

☐ Bowed or fractured bones ☐ Hydrops ☐ Undermineralization of bones

☐ Clubfeet ☐ Irregular, thickened or thin bones ☐ Other _____

Patient is: ☐ Living ☐ Deceased; describe circumstances: _____

Does the patient have radiographic findings? ☐ No ☐ Yes (describe details below) ☐ Unknown

_____

Has the patient undergone previous molecular genetic testing for this condition? ☐ No ☐ Yes ☐ Unknown

If yes, describe the gene(s), methodology, and results: _____

Is there any relevant family history? ☐ No ☐ Yes ☐ Unknown

If yes, attach a pedigree or specify the relative's relationship to the patient. List their symptoms and age of onset:

_____

Has DNA testing been performed for the family member(s)? ☐ No ☐ Yes ☐ Unknown

If yes, attach a copy of the relative's DNA laboratory result. (REQUIRED for familial mutation testing.)

Check the test you intend to order.

Prenatal testing for skeletal dysplasias:

☐ **2012010 Skeletal Dysplasia Panel, Sequencing and Deletion/Duplication, Fetal:** Next generation sequencing and tiled custom CGH on fetal specimens.

☐ **2001980 Familial Mutation, Targeted Sequencing, Fetal:** Targeted testing for a known familial sequence variants; a copy of relative's lab result is REQUIRED.

Postnatal testing for skeletal dysplasias:

☐ **2012015 Skeletal Dysplasia Panel, Sequencing and Deletion/Duplication:** Next generation sequencing and tiled custom CGH.

☐ **2001961 Familial Mutation, Targeted Sequencing:** Targeted testing for a variant previously identified in a family member; a copy of relative's lab result is REQUIRED.

Master Label

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