

THIS IS NOT A TEST REQUEST FORM. Please complete and submit with the test request form or electronic packing list.

PATIENT HISTORY FOR SKELETAL DYSPLASIA TESTING

Patient Name: _____ **Date of Birth:** _____ **Sex:** Female Male
Ordering Provider: _____ **Provider's Phone:** _____
Practice Specialty: _____ **Provider's Fax:** _____
Genetic Counselor: _____ **Counselor Phone:** _____

Patient's Ethnicity/Ancestry (check all that apply)

African American/Black Asian Hispanic White Other: _____

List country of origin (if known): _____

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.