INVITAE

Discover Dysplasias™
No-Charge Genetic Test for Skeletal Dysplasias

ORDER ID
For Invitae internal use only

Requisition Form
Discover Dysplasias TRF965-2

This requisition form can be used to submit an order for the **Discover Dysplasias™** program, a no-charge sponsored testing program for genetic disorders brought to you by **BioMarin Pharmaceutical Inc**. and **Invitae Corporation**.

INSTRUCTIONS: Review the ordering options and then complete all sections of this form. Your ordering option will be indicated in the test selection section.

ORDERING OPTIONS

1. DISCOVER DYSPLASIAS™ PROGRAM

For individuals that meet the eligibility criteria below and wish to receive the program specific genetic testing panels.

REQUIRED: You must select below the appropriate eligibility criteria for this patient.

This program is available to patients in the US and Canada with one or more of the following signs and symptoms suggestive of or consistent with a diagnosis of skeletal dysplasia (select one or more):

- ◯ Skeletal abnormalities suggestive of skeletal dysplasia
- ◯ Short stature
- ◯ Disproportionate growth
- ◯ Dysmorphic facial features
- ◯ Other signs or symptoms suggestive of skeletal dysplasia

2. GENE-SPECIFIC FAMILY FOLLOW-UP TESTING

For relatives of program participants who received a Pathogenic/Likely Pathogenic result or approved VUS who want to receive gene specific family follow-up testing at no additional cost. Relatives do not need to meet the eligibility criteria listed above. Learn more at www.invitae.com/family.

PATIENT INFORMATION

First name	MI	Last name
Date of birth (MM/DD/YYYY)	Biological sex ◯ M ◯ F	MRN (medical record number)

Ancestry ◯ Asian ◯ Black/African American ◯ White/Caucasian ◯ Ashkenazi Jewish ◯ Hispanic ◯ Native American ◯ Pacific Islander ◯ French Canadian ◯ Sephardic Jewish ◯ Mediterranean ◯ Other: _____

Phone	Email address (report access after clinician releases)	
Address		City
State/Prov	ZIP/Postal code	Country

Ship a saliva kit to this patient (to submit, fax this form to Client Services at 415-276-4164)
- ◯ Ship kit to address above
- ◯ Ship kit to alternate address: _____

SPECIMEN INFORMATION

Specimen type: Blood (3-mL purple EDTA) -OR- Saliva (Oragene™) -OR- Assisted Saliva -OR- DNA source: _____

We are unable to accept blood/saliva from patients with:
- *Allogeneic bone marrow transplants*
- *Blood transfusion <2 weeks prior to specimen collection*

Specimen collection date (MM/DD/YYYY): *If not provided, the day before specimen receipt will be used* ☐☐ ☐☐ ☐☐☐☐

Special cases: ◯ History of/current hematologic malignancy in patient

INVITAE PARTNER CODE

SKEL

CLINICIAN INFORMATION

Organization name	
Phone	Fax
Address	City
State/Prov	ZIP/Postal code / Country

Primary clinical contact name (if different from ordering provider)	NPI
Specialty	
Primary clinical contact email address (for report access)	

Ordering provider (select one ordering provider by marking the checkbox before the name)

	Name	NPI	Specialty	Email address (for report access)
◯				
◯				
◯				
◯				
◯				

Additional clinical or laboratory contacts (optional, to share access to order online)

◯ **Share this order with the primary clinical contact's default clinical team, manage at invitae.com**

Name	Email address (for report access)
Name	Email address (for report access)

CLINICAL HISTORY

FAMILY HISTORY

Is there a family history of disease for which the patient is being tested? ◯ Yes ◯ No If yes, describe below and attach pedigree and/or clinical notes.

Relative's relationship to this patient	Maternal or paternal	Diagnosed condition	Age at diagnosis	Relative's relationship to this patient	Maternal or paternal	Diagnosed condition	Age at diagnosis

PERSONAL HISTORY

Is/was this patient affected or symptomatic?† ◯ Yes ◯ No

Provide details in the required clinical history questions (if applicable).

† Symptomatic means this patient has features or signs known or suspected to be related to the genetic testing being ordered and could include findings on physical examination, laboratory tests, or imaging.

REQUIRED CLINICAL HISTORY

When was the first sign or symptom of skeletal dysplasia noted?

◯ **Prenatally**

◯ **At Birth**

◯ **Age in Years** _____

Medical History and Physical Examination (select all that apply)

Growth
- ◯ Short stature
- ◯ Tall stature
- ◯ Intrauterine growth retardation
- ◯ Postnatal growth retardation
- ◯ Asymmetric bone growth
- ◯ Normal stature and/or growth

Development
- ◯ Developmental delay
- ◯ Developmental regression
- ◯ Intellectual disability
- ◯ Progressive cognitive impairment
- ◯ Normal intellect
- ◯ Normal development

Head and neck
- ◯ Macrocephaly
- ◯ Microcephaly
- ◯ Craniosynostosis
- ◯ Cloverleaf skull
- ◯ Wormian bones
- ◯ Large fontanelles
- ◯ Wide cranial sutures
- ◯ Hydrocephalus
- ◯ Frontal bossing
- ◯ Decreased calvarial ossification
- ◯ Short neck
- ◯ Normal head size/shape

Facial features
- ◯ Coarse facial features
- ◯ Midface hypoplasia
- ◯ Depressed nasal bridge
- ◯ Prominent nasal bridge
- ◯ Anteverted nares
- ◯ Hypertelorism
- ◯ Downslanted palpebral fissures
- ◯ Long philtrum
- ◯ Micrognathia
- ◯ Proptosis
- ◯ Other dysmorphic facial features
- ◯ No noted facial dysmorphism

Oral/dental
- ◯ Cleft lip
- ◯ Cleft palate
- ◯ High arched palate
- ◯ Bifid uvula
- ◯ Multiple oral frenula
- ◯ Natal teeth
- ◯ Supernumerary teeth
- ◯ Dentinogenesis imperfecta
- ◯ Hypoplasia of dental cementum

Eyes and ears
- ◯ Congenital cataract
- ◯ Corneal clouding
- ◯ Severe myopia
- ◯ Microspherophakia
- ◯ Ectopia lentis
- ◯ Retinal degeneration or detachment
- ◯ Low-set ears
- ◯ Posteriorly rotated ears
- ◯ Microtia
- ◯ Cystic swelling of the pinnae
- ◯ Sensorineural hearing loss
- ◯ Conductive hearing loss

Limbs
- ◯ Rhizomelia
- ◯ Mesomelia
- ◯ Acromelia
- ◯ Acromesomelia
- ◯ Micromelia
- ◯ Disproportionately long limbs
- ◯ Bowing of the long bones
- ◯ Coxa vara
- ◯ Genu varum
- ◯ Genu valgum
- ◯ Talipes equinovarus
- ◯ Radial ray defects
- ◯ Absence of a limb
- ◯ Patellar hypoplasia or aplasia

Hands and feet
- ◯ Brachydactyly
- ◯ Arachnodactyly
- ◯ Preaxial polydactyly
- ◯ Postaxial polydactyly
- ◯ Syndactyly
- ◯ Clinodactyly
- ◯ Camptodactyly
- ◯ Ectrodactyly
- ◯ Hitchhiker thumb
- ◯ Trident hand
- ◯ Broad or spatulate fingers/toes
- ◯ Pes planus
- ◯ Nail hypoplasia or dysplasia

Thorax
- ◯ Thoracic hypoplasia
- ◯ Short thorax
- ◯ Long thorax
- ◯ Narrow chest
- ◯ Bell-shaped chest
- ◯ Barrel-shaped chest
- ◯ Pectus carinatum
- ◯ Pectus excavatum
- ◯ Abnormal ribs
- ◯ Hypoplastic clavicles
- ◯ Hypoplastic scapulae

Spine
- ◯ Scoliosis
- ◯ Kyphosis
- ◯ Kyphoscoliosis
- ◯ Lumbar lordosis
- ◯ Platyspondyly
- ◯ Other abnormal form of the vertebral bodies
- ◯ Atlanto-axial instability
- ◯ Cervical spine instability
- ◯ Cervical medullary compression
- ◯ Craniovertebral junction stenosis
- ◯ Spinal stenosis

Joints
- ◯ Joint laxity
- ◯ Joint hypermobility
- ◯ Congenital joint contractures
- ◯ Progressive joint contractures
- ◯ Limb joint contracture
- ◯ Multiple joint dislocations
- ◯ Joint stiffness
- ◯ Joint pain
- ◯ Osteoarthritis
- ◯ Carpal tunnel syndrome in a child

Bone structure
- ◯ Fracture of the long bones
- ◯ Recurrent fractures
- ◯ Thickened long bones
- ◯ Thin long bones
- ◯ Delayed bone age
- ◯ Advanced bone age
- ◯ Decreased bone mineralization

Skin and hair
- ◯ Redundant skin folds
- ◯ Ichthyosiform erythroderma
- ◯ Acanthosis nigricans
- ◯ Aberrant mongolian spots
- ◯ Thickened skin
- ◯ Soft and/or thin skin
- ◯ Skin hyperextensibility
- ◯ Thickened coarse hair
- ◯ Brittle hair
- ◯ Hirsutism
- ◯ Sparse or absent hair

Heart and lungs
- ◯ Cardiomegaly
- ◯ Atrial septal defect or single atrium
- ◯ Patent ductus arteriosus
- ◯ Transposition of the great vessels
- ◯ Cardiac valve prolapse
- ◯ Valvular stenosis
- ◯ Aortic root dilation
- ◯ Heart conduction abnormalities
- ◯ Lung hypoplasia
- ◯ Spontaneous pneumothorax

Neuromuscular
- ◯ Abnormal gait
- ◯ Frequent falls
- ◯ Hypotonia
- ◯ Hypertonia
- ◯ Muscle weakness and/or atrophy
- ◯ Spasticity
- ◯ Neuropathy or impaired sensation
- ◯ Reduced or absent deep tendon reflexes
- ◯ Increased deep tendon reflexes

Other Systems
- ◯ Hepatomegaly
- ◯ Splenomegaly
- ◯ Genital abnormalities
- ◯ Renal abnormalities
- ◯ Inguinal or umbilical hernia
- ◯ Autoimmune disease

CLINICAL HISTORY (continued)

Does this patient have a clinical or suspected diagnosis of a specific skeletal dysplasia? If so, please provide diagnosis.

◯ Clinical Diagnosis ◯ Suspected Diagnosis

Radiographic Findings – Please provide information about relevant radiographic findings not addressed in the checklist above.

Laboratory Findings – Please provide information about relevant laboratory findings, including biochemical testing results.

OPTIONAL - REQUESTED VARIANTS FOR THIS PATIENT'S REPORT, IF KNOWN

*To have the presence or absence of specific variants commented on in this patient's report, provide the details below. For gene-specific family follow-up see **Note** under Test Selection.*

Was the proband (individual with variant) tested at Invitae? ◯ Yes, Invitae Order ID: RQ# _____ ◯ No: Attach copy of lab results (*required*)

Variant(s) *(e.g., GENE c.2200A>T (p.Thr734Ser) NM_00012345) If left blank, all variants identified in the proband will be commented on.*		**This patient's relationship to proband:**
		◯ Parent ◯ Sibling ◯ Grandchild
		◯ Child ◯ Self ◯ Other: _____

TEST SELECTION – Select option 1 or 2 below:

◯ 1. DISCOVER DYSPLASIAS™ PROGRAM – Indicate test(s) to be performed below:

Test code	Test name	# of genes	Gene list
◯ 89100	Invitae Expanded Skeletal Dysplasia Panel	109	ACP5, ADAMTS10, AGPS, ALPL, ANKH, ARSB, ARSE, B3GALT6, B3GAT3, BGN, BMP2, BMPR1B, CFAP410, CANT1, CDC45, CDC6, CDT1, CHST14, CHST3, COL10A1, COL11A1*, COL2A1, COL9A1, COL9A2, COL9A3, COMP, CUL7, DDR2, DLL3, DVL1, DVL3, DYM, EBP, EIF2AK3, EVC, EVC2, FBN1, FGFR1, FGFR2, FGFR3, FLNA, FLNB, FN1*, GALNS, GDF5, GLB1, GMNN, GNPAT, GNPTAB, GNPTG, GNS, GORAB, GPC6, GUSB, HES7, HGSNAT, HSPG2, HYAL1, ICK, IDS*, IDUA, IFT172, IHH, IMPAD1, INPPL1, KAT6B, KIF22, LBR, LEMD3, LFNG, LIFR, LMX1B, MAP3K7, MATN3, MESP2, MGP, MMP13, MMP2, MMP9, NAGLU, NKX3-2, NPR2, OBSL1, ORC1, ORC4, ORC6, PAPSS2, PEX5, PEX7, PTH1R, RIPPLY2, RMRP, ROR2, RSPRY1, RUNX2, SGSH, SH3PXD2B, SLC26A2, SLC39A13, SMAD4, SMARCAL1, SOX9, TBCE, TBX6, TRPV4, WDR35, WISP3, WNT5A, XYLT1

**Denotes gene offered with limitation.*

◯ 2. GENE-SPECIFIC FAMILY FOLLOW-UP TESTING *For relatives of a program participant ('proband') who received a Pathogenic/Likely Pathogenic result or approved VUS.*

Proband's Invitae Order ID: RQ# _____	**This patient's relationship to proband:** ◯ Parent ◯ Sibling ◯ Grandchild ◯ Child ◯ Other: _____	**Gene(s) to be tested in this patient:**

***NOTE**: The presence or absence of all variants identified in the proband for the gene(s) ordered for gene-specific family follow-up will be commented on in this patient's report unless a limited selection is specified in the **Requested Variants** section above. Invitae will report any Pathogenic/Likely Pathogenic variants found in this patient for the gene(s) ordered.*

Invitae continually updates its panels based on the most recent evidence. If an order is placed using an outdated test requisition form, Invitae reserves the right to upgrade ordered tests to their current versions. Test IDs containing add-on codes will include the original panel as well as the add-on.

By signing this form, the medical professional acknowledges that the individual/family member authorized to make decisions for the individual (collectively, the "Patient") has been supplied information regarding and consented to undergo genetic testing, and has reviewed and signed Invitae's Patient Consent for Sponsored Genetic Testing (www.invitae.com/forms). The medical professional will retain this signed Consent and will provide it to Invitae upon request. In connection with the Discover Dysplasias™ Program, the Patient has been informed that Invitae may notify the Patient of clinical updates related to genetic test results (in consultation with the ordering medical professional as indicated). The medical professional warrants that he/she will not seek reimbursement for this no-cost test from any third party, including but not limited to federal healthcare programs. The medical professional also hereby acknowledges that organization and clinician contact information provided in the order may be shared with third party sponsors of the Discover Dysplasias™ Program, and that such third parties that may contact the medical professional directly in connection with the Discover Dysplasias™ Program. The medical professional is under no obligation to order or recommend any products that may be made available by third party sponsors. The medical professional further acknowledges that he/she has made the Patient aware that third party sponsors of the Discover Dysplasias™ Program may contact the Patient's medical professional regarding de-identified information gathered through the Discover Dysplasias™ Program. The Patient has been informed that his/her personal information and specimen will be transferred to and processed in the United States and that de identified Patient data may be used and shared for research purposes in the United States. In addition to the above, I attest that I am authorized under applicable law to order this test.

Medical professional signature (required)	**Date (MM/DD/YYYY)**