Genetic testing: skeletal dysplasia and rare bone disorders

These services may or may not be covered by your HealthPartners plan. Please see your plan documents for your specific coverage information. If there is a difference between this general information and your plan documents, your plan documents will be used to determine your coverage.

Administrative Process

Prior authorization is required for the following services:

- Genetic testing for Osteogenesis Imperfecta
- Multigene Panel Analysis for Skeletal Dysplasia or Rare Bone Disorder
- Genetic testing for Covered Skeletal Dysplasias or Rare Bone Disorders
- Testing that is associated with a procedure code listed in “Box A”, below

Tests that require prior authorization will be reviewed for medical necessity of the testing as a whole. That is, a single coverage decision will apply to all of the tests, services, and/or procedure codes associated with the genetic test, whether they are requested/billed together or separately.

Box A: Genetic testing procedure codes that require prior authorization
Molecular pathology procedures, Tier 2 or unlisted (CPT 81400-81408, 81479)
Unlisted multianalyte assays (CPT 81599)
Any other listed or unlisted laboratory/pathology CPT code when it is used in association with a genetic test.

Policy Reference Table

If available, codes are listed below for informational purposes only, and do not guarantee member coverage or provider reimbursement. The list may not be all-inclusive.

Coverage Criteria Sections	Example Tests (Labs)	Common CPT Codes	Common ICD Codes
Osteogenesis Imperfecta	Osteogenesis imperfecta COL1A1 & COL1A2 NGS Panel (HNL Genomics)	81406, 81408, 81479	Q78.0, Z82.79
	Osteogenesis Imperfecta Panel (PreventionGenetics, part of Exact Sciences)		
	Osteogenesis Imperfecta NGS Panel - Dominant & Recessive (HNL Genomics)		
Multigene Panel Analysis for Skeletal Dysplasia or Rare Bone Disorder	Skeletal Disorders Panel (Invitae)	81400, 81401, 81402, 81403, 81404, 81405, 81406, 81407, 81408, 81479	M85, Q77, Q78
	Skeletal Dysplasia Core & Extended NGS Panel (HNL Genomics)		
	Comprehensive Skeletal Dysplasias and Disorders Panel (Blueprint Genetics)		
Other Covered Skeletal Dysplasias and Rare Bone Disorders			
Other Covered Skeletal Dysplasias and Rare Bone Disorders	varies	81400, 81401, 81402, 81403, 81404, 81405, 81406, 81407, 81408, 81479	M85, Q77, Q78

Coverage

Osteogenesis Imperfecta

1. *COL1A1* and *COL1A2* variant analysis or multigene panel analysis that includes *COL1A1* and *COL1A2* to establish or confirm a diagnosis of osteogenesis imperfecta (OI) is considered **medically necessary** when:
    - A. The member has any of the following:
        - i. Fractures with minimal or no trauma in the absence of other factors, such as non-accidental trauma (NAT), or other known disorders of bone, or
        - ii. Short stature, often with bone deformity, or
        - iii. Blue/gray scleral hue, or
        - iv. Dentinogenesis imperfecta (DI), or
        - v. Progressive, postpubertal hearing loss, or
        - vi. Ligamentous laxity or other signs of connective tissue abnormality, or
        - vii. Family history of OI, or
        - viii. Fractures of varying ages and stages of healing (often of the long bones), or
        - ix. “Codfish” vertebrae, or
        - x. Wormian bones, or
        - xi. Protrusio acetabuli, or
        - xii. Low bone mass or osteoporosis.
2. *COL1A1* and *COL1A2* variant analysis or multigene panel analysis that includes *COL1A1* and *COL1A2* to establish or confirm a diagnosis of osteogenesis imperfecta is considered **investigational** for all other indications.

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Multigene Panel Analysis for Skeletal Dysplasia or Rare Bone Disorder

1. Multigene panel analysis to confirm or establish a post-natal diagnosis of a skeletal dysplasia or a rare bone disorder may be considered **medically necessary** when:
    - A. The differential diagnosis includes more than one type of skeletal dysplasia or bone disorder, **and**
    - B. The member displays one or more of the following clinical features of a skeletal dysplasia:
        - i. Prenatal ultrasound that showed shortening of the bones of the arms and legs more than 3 standard deviations below the mean, **or**
        - ii. Prenatal ultrasound that showed head circumference greater than 75th percentile, **or**
        - iii. Prenatal ultrasound that showed bone irregularities (e.g., bowed, fractured, thickened, thin, undermineralized, etc.), **or**
        - iv. Prenatal ultrasound that showed abnormal ribs or a small chest circumference, **or**
        - v. Postnatal short stature with height or length less than 3rd percentile.
2. Multigene panel analysis to confirm or establish a diagnosis of a skeletal dysplasia or a rare bone disorder is considered **investigational** for all other indications.

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Other Covered Skeletal Dysplasias and Rare Bone Disorders

1. Genetic testing to establish or confirm one of the following skeletal dysplasias or rare bone disorders to guide management is considered **medically necessary** when the member demonstrates clinical features* consistent with the disorder (the list is not meant to be comprehensive, see 2 below):
    - A. Achondroplasia Group
        - i. Achondroplasia
        - ii. Hypochondroplasia
        - iii. Thanatophoric Dysplasia
    - B. Type II Collagenopathies
        - i. Hypochondrogenesis
        - ii. Spondyloepiphyseal Dysplasia
    - C. Type XI Collagen Disorders
        - i. Fibrochondrogenesis
        - ii. Otospondylomegaepiphyseal Dysplasia (OSMED)
    - D. Sulfation Disorders
        - i. Achondrogenesis IB
        - ii. Atelosteogenesis II
        - iii. Diastrophic Dysplasia
        - iv. Chondrodysplasia with Congenital Joint Dislocations
    - E. Filamin Disorders and Similar Disorders
        - i. Atelosteogenesis Type I
        - ii. Atelosteogenesis Type III

iii. Larsen Syndrome
iv. Spondylo-Carpal-Tarsal Dysplasia

F. Short-Rib Dysplasias (with and without Polydactyly)
i. Chondroectodermal Dysplasia (Ellis-van Creveld (EVC))
ii. Short-Rib Polydactyly Syndrome I, II, III, IV including Asphyxiating Thoracic Dystrophy

G. Metaphyseal Dysplasias
i. Cartilage-Hair Hypoplasia

H. Spondylo-Epi-(Meta)-Physeal Dysplasia
i. SEMD, Short Limb Abnormal Calcification Type

I. Acromesomelic Disorders
i. Acromesomelic Dysplasia, Type Maroteaux

J. Mesomelic and Rhizo-Mesomelic Dysplasias
i. Langer Type (Homozygous Dyschondrosteosis)

K. Bent Bone Dysplasias
i. Campomelic Dysplasia
ii. Stuve-Wiedemann Dysplasia
iii. Bent Bone Dysplasia FGFR2 Type

L. Slender Bone Dysplasia
i. Microcephalic Osteodysplastic Primordial Dwarfism
ii. Osteocraniostenosis

M. Neonatal Osteosclerotic Dysplasias
i. Bloomstrand Dysplasia
ii. Caffey Disease (Infantile)
iii. Raine Dysplasia

N. Increased Bone Density Group
i. Osteopetrosis

O. Abnormal Mineralization Group
i. Hypophosphatasia

P. Multiple Epiphyseal Dysplasia and Pseudoachondroplasia Group
i. Multiple Epiphyseal Dysplasia (MED) - Autosomal Dominant
ii. Multiple Epiphyseal Dysplasia (MED) - Autosomal Recessive
iii. Stickler Syndrome

Q. Hereditary Multiple Osteochondromas

2. Genetic testing to establish or confirm the diagnosis of all other skeletal dysplasias or rare bone disorders not specifically discussed within this, or another medical policy will be evaluated by the criteria outlined in **General Approach to Genetic and Molecular Testing** (see policy for coverage criteria).

*Clinical features for a specific disorder may be outlined in resources such as GeneReviews, OMIM, National Library of Medicine, Genetics Home Reference, or other scholarly sources.

Definitions

1. **Non-accidental Trauma (NAT)** refers to injury that is purposely inflicted upon a child (e.g., child abuse). NAT often occurs as injury to the skin and soft tissue, but approximately a third of NATs are fractures.

Products

This information is for most, but not all, HealthPartners plans. Please read your plan documents to see if your plan has limits or will not cover some items. If there is a difference between this general information and your plan documents, your plan documents will be used to determine your coverage. These coverage criteria do not apply to Medicare Products. For more information regarding Medicare coverage criteria or for a copy of a Medicare coverage policy, contact Member Services at 952-883-7272 or 1-877-778-8384.

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