

# CPT codes: Pediatric genetics



Chromosomal developmental disorders	CPT code(s)
Invitae Chromosomal Microarray Analysis	81229
Follow-up FISH for Developmental Disorders CMA	88230, 88271, 88273, 88291
Invitae Karyotype (Chromosome Analysis)	88230, 88262, 88291
Fragile X Syndrome	81243TSO, 99001
Chronic pancreatitis	CPT code(s)
Invitae Chronic Pancreatitis Panel	81223, 81405, 81404
Ciliopathies	CPT code(s)
Invitae Bardet-Biedl Syndrome Panel	81404, 81406, 81408
Invitae Ciliopathies Panel	81405, 81406, 81407, 81408
Invitae Joubert and Meckel-Gruber Syndromes Panel	81405, 81407, 81408
Invitae Neonatal Respiratory Distress Panel	81479
Invitae Nephronophthisis Panel	81405, 81407, 81408
Invitae Perinatal Lethal Skeletal Dysplasia and Skeletal Ciliopathies Panel	81479
Invitae Primary Ciliary Dyskinesia Panel	81479
Connective tissue	CPT code(s)
Invitae Connective Tissue Disorders Panel	81408
Congenital heart defects	CPT code(s)
Invitae Congenital Heart Defects and Heterotaxy Panel	81210, 81311, 81407, 81408
Cystic fibrosis and chronic pancreatitis	CPT code(s)
Invitae Cystic Fibrosis Test	81220
Deafness	CPT code(s)
Invitae Comprehensive Deafness Panel	81406, 81407
Invitae Stickler Syndrome Panel	81479
Invitae Usher Syndrome Panel	81406, 814071, 81408
Developmental disorders	CPT code(s)
Invitae Alagille Syndrome Panel	81406, 81479
Invitae Alpha Thalassemia X-linked Intellectual Disability Test	81479
Invitae Baraitser-Winter Cerebrofrontofacial Syndrome Panel	81479
Invitae Carpenter Syndrome Panel	81479
Invitae Coffin-Lowry Syndrome Test	81479
Invitae Cohen Syndrome Test	81408
Invitae Cornelia de Lange Syndrome Test	81479
Invitae Hypogonadotropic Hypogonadism Panel	81407, 81479
Invitae Kabuki Syndrome Test	81479
Invitae KBG Syndrome Test	81405
Invitae Oculo-Facio-Cardio-Dental Syndrome Test	81479
Invitae Rubinstein-Taybi Syndrome Panel	81406, 81479
Invitae Simpson-Golabi-Behmel Syndrome Test	81479
Invitae van der Woude Syndrome Panel	81479
Invitae Weaver Syndrome Test	81479

Disorders of sex development/endocrinology	CPT code(s)
Invitae Androgen Insensitivity Panel	81405, 81479
Invitae Disorders of Female Sex Development Test	81400
Invitae Disorders of Sex Development Panel	81479
Invitae Hypogonadotropic Hypogonadism Panel	81407, 81479
Epilepsy seizures and developmental brain abnormalities	CPT code(s)
Invitae Baraitser-Winter Cerebrofrontofacial Syndrome Test	81479
Invitae Brain Malformations Panel	81404, 81408
Invitae Cerebral Cavernous Malformations Panel	81479
Invitae Cerebral Palsy Spectrum Disorders Panel	81406, 81408
Invitae Epilepsy Panel	81302, 81321, 81401, 81404, 81405, 81406
Invitae Holoprosencephaly Panel	81479
Invitae Leukodystrophy and Genetic Leukoencephalopathy Panel	81479
Invitae Rett/Angelman and Related Disorders Panel	81302, 81404, 81405, 81406
Invitae Septo-optic Dysplasia Panel	81479
Invitae Tuberous Sclerosis Complex Panel	81406
Eye disorders	CPT code(s)
Invitae Alport Syndrome Panel	81407, 81408
Invitae Bardet-Biedl Syndrome Panel	81404, 81406, 81408
Invitae Cataracts Panel	81479
Invitae Early-Onset Glaucoma Panel	81479
Invitae Glaucoma Panel	81479
Invitae Inherited Retinal Disorders Panel	81479
Invitae Macular Dystrophy Panel	81408
Invitae Microphthalmia/Anophthalmia Disorders Panel	81479
Invitae Oculocutaneous Albinism Panel	81479
Invitae Oculo-Facio-Cardio-Dental Syndrome Test	81479
Invitae Septo-optic Dysplasia Panel	81479
Invitae Stickler Syndrome Panel	81479
Hypophosphatemia	CPT code(s)
Invitae Hypophosphatemia Panel	81404, 81405, 81406
Invitae X-Linked Hypophosphatemia Test	81404, 81405, 81406
Kidney disorders	CPT code(s)
Invitae Congenital Anomalies of Kidney and Urinary Tract (CAKUT) Panel	81479
Invitae Cystic Kidney Disease Panel	81479
Invitae Nephrotic Syndrome and Focal Segmental Glomerulosclerosis (FSGS) Panel	81408

Overgrowth syndromes	CPT code(s)
Invitae Overgrowth Syndromes Panel	81479
Invitae Overgrowth and Macrocephaly Syndromes Panel	81408, 81321
Invitae Simpson-Golabi-Behmel Syndrome Test	81479
Invitae Weaver Syndrome Test	81479
Parental follow-up for chromosomal developmental disorders	CPT code(s)
Invitae Parental Chromosomal Microarray Analysis for Developmental Disorders	81229
Invitae Parental FISH Analysis for Developmental Disorders	88230, 88271, 88273, 88291
Invitae Parental Karyotype (Chromosome Analysis) for Developmental Disorders	88230, 88262, 88291
Pediatric microarray	CPT code(s)
Invitae Parental Chromosomal Microarray Analysis for Pregnancy Loss	81229
Fragile X Syndrome	81243TSO, 99001
Invitae Neurofibromatosis Type 1 Test	81408
Invitae Neurofibromatosis Type 2 Test	81405
Pediatric oncology	CPT code(s)
Invitae Pediatric Hematologic Malignancies Panel	81292, 81294, 81295, 81297, 81298, 81300, 81317, 81319
Invitae Pediatric Nervous System/Brain Tumors Panel	81292, 81294, 81295, 81297, 81298, 81300, 81317, 81319
Invitae Pediatric Solid Tumors Panel	81408
Rasopathies (noonan spectrum disorders)	CPT code(s)
Invitae Cardio-Facio-Cutaneous Syndrome Panel	81210, 81406, 81405
Invitae Legius Syndrome Test	81405
Invitae Neurofibromatosis Type 1 Test	81408
Invitae RASopathies and Noonan Spectrum Disorders Panel	81442
Skeletal disorders	CPT code(s)
Invitae Adams-Oliver Syndrome Panel	81407, 81479
Invitae Antley-Bixler syndrome Test	81479
Invitae ARSE-Related Chondrodysplasia Punctata Test	81479
Invitae Campomelic Dysplasia Test	81479
Invitae Craniosynostosis Panel	81404, 81479
Invitae Ellis van Creveld and Weyers Acrofacial Dysostosis Panel	81479
Invitae Hereditary Multiple Osteochondromas Panel	81479
Invitae Limb and Digital Malformations Panel	81408
Invitae Osteogenesis Imperfecta and Bone Fragility Panel	81479
Invitae Perinatal Lethal Skeletal Dysplasia and Skeletal Ciliopathies Panel	81479
Invitae Stickler Syndrome Panel	81479
Invitae Skeletal Disorders Panel	81479
Invitae Spondylocostal Dysostosis Panel	81479
Invitae Trichorhinophalangeal Syndrome Panel	81479

## CPT codes: Pediatric genetics (continued)



Skin disorders	CPT code(s)
Invitae Adams-Oliver Syndrome Panel	81407, 81479
Invitae Cardio-Facio-Cutaneous Syndrome Panel	81210, 81406, 81405
Invitae Congenital Ichthyosis Panel	81479
Invitae Ectodermal Dysplasia with or without Tooth Agenesis Panel	81479
Invitae Epidermolysis Bullosa and Palmoplantar Keratoderma Panel	81479
Invitae Legius Syndrome Test	81405
Invitae Oculocutaneous Albinism Panel	81479
Invitae van der Woude Syndrome Test	81479
Invitae Xeroderma Pigmentosum Panel	81479

The CPT codes included in this publication are in accordance with Current Procedural Terminology, a publication of the American Medical Association, as of January 2021. CPT codes are provided here for the convenience of our clients; however, correct coding often varies from one carrier to another, and Invitae may bill specific carriers using codes other than those shown. Clients who bill for services should verify the code(s) with the applicable payer to confirm that their use is appropriate in each case.