

遺伝病のカテゴリ	小慢疾病分類 NO.	指定難病分類NO	疾患名	国際分類 ISDS (International Skeletal Dysplasia Society) Unger et al. Am J Med Genet 2023			和訳	注釈	
				グループ番号	グループ名	疾患ID (NOS)			
単一・単一遺伝子病 多因子・多因子遺伝病							出典:『骨系統疾患国際分類和訳作業ワーキンググループの活動報告』(2019) https://www.jsog.or.jp/news/pdf/s_husanki_20210226.pdf 骨系統疾患国際分類(2019)和訳		
単一/多因子	骨系統疾患1	骨・関節疾患	273 胸郭不全症候群/肋骨異常を伴う先天性側弯症	10	10: Skeletal disorders caused by abnormalities of cilia or ciliary signaling:	10-0010	Short rib-polydactyly syndrome (SRPS), DYNC2H1-related	短肋骨多指症候群	
						10-0020	Short rib-polydactyly syndrome (SRPS), IFT80-related	短肋骨多指症候群	
						10-0030	Short rib-polydactyly syndrome (SRPS), IFT81-related	短肋骨多指症候群	
						10-0040	Short rib-polydactyly syndrome (SRPS), WDR34-related	短肋骨多指症候群	
						10-0050	Short rib-polydactyly syndrome (SRPS), WDR60-related	短肋骨多指症候群	
						10-0060	Short rib-polydactyly syndrome (SRPS), DYNC2L1-related	短肋骨多指症候群	
						10-0070	Short rib-polydactyly syndrome (SRPS), NEK1-related	短肋骨多指症候群	
						10-0080	Short rib-polydactyly syndrome (SRPS), IFT122-related	短肋骨多指症候群	
						10-0090	Short rib-polydactyly syndrome (SRPS), WDR19-related	短肋骨多指症候群	
						10-0100	Short rib-polydactyly syndrome (SRPS), INTU-related	短肋骨多指症候群	
						10-0110	Short rib-polydactyly syndrome (SRPS), TRAF3IP1-related	短肋骨多指症候群	
						10-0120	Endocrine-cerebro-osteo dysplasia (ECO), CILK1-related	なし	
						10-0130	Short-rib thoracic dysplasia (formerly asphyxiating thoracic dysplasia - Jeune syndrome), DYNC2H1-related	呼吸不全性胸郭異形成症	
						10-0140	Short-rib thoracic dysplasia (formerly asphyxiating thoracic dysplasia - Jeune syndrome), DYNC2L1-related	呼吸不全性胸郭異形成症	
						10-0150	Short-rib thoracic dysplasia (formerly asphyxiating thoracic dysplasia - Jeune syndrome), WDR34-related	呼吸不全性胸郭異形成症	
						10-0160	Short-rib thoracic dysplasia (formerly asphyxiating thoracic dysplasia - Jeune syndrome), TCTEX1D2-related	呼吸不全性胸郭異形成症	
						10-0170	Short-rib thoracic dysplasia (formerly asphyxiating thoracic dysplasia - Jeune syndrome), WDR60-related	呼吸不全性胸郭異形成症	
						10-0180	Short-rib thoracic dysplasia (formerly asphyxiating thoracic dysplasia - Jeune syndrome), WDR19-related	呼吸不全性胸郭異形成症	
						10-0190	Short-rib thoracic dysplasia (formerly asphyxiating thoracic dysplasia - Jeune syndrome), IFT140-related	呼吸不全性胸郭異形成症	
						10-0200	Short-rib thoracic dysplasia (formerly asphyxiating thoracic dysplasia - Jeune syndrome), TTC21B-related	呼吸不全性胸郭異形成症	
						10-0210	Short-rib thoracic dysplasia (formerly asphyxiating thoracic dysplasia - Jeune syndrome), IFT122-related	呼吸不全性胸郭異形成症	
						10-0220	Short-rib thoracic dysplasia (formerly asphyxiating thoracic dysplasia - Jeune syndrome), WDR35-related	呼吸不全性胸郭異形成症	
						10-0230	Short-rib thoracic dysplasia (formerly asphyxiating thoracic dysplasia - Jeune syndrome), IFT43-related	呼吸不全性胸郭異形成症	
						10-0240	Short-rib thoracic dysplasia (formerly asphyxiating thoracic dysplasia - Jeune syndrome), IFT80-related	呼吸不全性胸郭異形成症	
						10-0250	Short-rib thoracic dysplasia (formerly asphyxiating thoracic dysplasia - Jeune syndrome), IFT172-related	呼吸不全性胸郭異形成症	
						10-0260	Short-rib thoracic dysplasia (formerly asphyxiating thoracic dysplasia - Jeune syndrome), IFT81-related	呼吸不全性胸郭異形成症	
						10-0270	Short-rib thoracic dysplasia (formerly asphyxiating thoracic dysplasia - Jeune syndrome), IFT52-related	呼吸不全性胸郭異形成症	
						10-0280	Short-rib thoracic dysplasia (formerly asphyxiating thoracic dysplasia - Jeune syndrome), CFAP410-related	呼吸不全性胸郭異形成症	
						10-0290	Short-rib thoracic dysplasia (formerly asphyxiating thoracic dysplasia - Jeune syndrome), CEP120-related	呼吸不全性胸郭異形成症	
						10-0300	Short-rib thoracic dysplasia (formerly asphyxiating thoracic dysplasia - Jeune syndrome), KIAA0586-related	呼吸不全性胸郭異形成症	
						10-0310	Short-rib thoracic dysplasia (formerly asphyxiating thoracic dysplasia - Jeune syndrome), GRK2-related	呼吸不全性胸郭異形成症	
						10-0320	Short-rib thoracic dysplasia (formerly asphyxiating thoracic dysplasia - Jeune syndrome), TRAF3IP1-related	呼吸不全性胸郭異形成症	
						10-0330	Short-rib thoracic dysplasia (formerly asphyxiating thoracic dysplasia - Jeune syndrome), KIAA0753-related	呼吸不全性胸郭異形成症	
						10-0340	Axial spondylometaphyseal dysplasia, CFAP410-related	軸性脊椎骨幹端異形成症	
						10-0350	Axial spondylometaphyseal dysplasia, NEK1-related	軸性脊椎骨幹端異形成症	
				36	36: Vertebral and costal dysostoses	36-0100	Currarino syndrome, MNX1-related	Currarino 症候群	
						36-0020	Spondylocostal dysostosis, DLL3-related	脊椎肋骨異骨症	
						36-0030	Spondylocostal dysostosis, MESP2-related	脊椎肋骨異骨症	
						36-0040	Spondylocostal dysostosis, LFNG-related	脊椎肋骨異骨症	
						36-0050	Spondylocostal dysostosis, HES7-related	脊椎肋骨異骨症	
						36-0060	Spondylocostal dysostosis, TBX6-related	脊椎肋骨異骨症	
						36-0070	Spondylocostal dysostosis, RIPPLY2-related	脊椎肋骨異骨症	
						36-0080	Vertebral segmentation defect (congenital scoliosis) with variable penetrance, MESP2-related	種々の浸透度を有する分節性脊椎欠損症(先天性側弯症)	
						36-0090	Vertebral segmentation defect (congenital scoliosis) with variable penetrance, HES7-related	種々の浸透度を有する分節性脊椎欠損症(先天性側弯症)	
						36-0100	Short stature, cervical segmentation defects, and developmental delay, CDK10-related	なし	
						36-0110	Klippel-Feil syndrome, GDF6-related	Klippel-Feil 症候群	
						36-0120	Klippel-Feil syndrome, MEOX1-related	Klippel-Feil 症候群	
						36-0130	Klippel-Feil syndrome, GDF3-related	Klippel-Feil 症候群	
						36-0140	Klippel-Feil syndrome, MYO18B-related	Klippel-Feil 症候群	
						36-0150	Cervico-oculo-acoustic (Wildervanck) syndrome	なし	
						36-0160	Cerebro-costo-mandibular syndrome (rib gap syndrome), SNRPB-related	脳・肋骨・下顎症候群 (rib gap 症候群)	
36-0170	Cerebro-costo-mandibular-like syndrome, COG1-related	脳・肋骨・下顎様症候群 (CDG IIg 型)							

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			" 6. 歯限局型				
単一	骨系統疾患7	代謝疾患	326 大理石骨病 1. 早発型	24 Osteopetrosis and related osteoclast disorders	24-0010 Osteopetrosis, neonatal or infantile form, TCIRG1-related 24-0020 Osteopetrosis, neonatal or infantile form, CLCN7-related 24-0030 Osteopetrosis, neonatal or infantile form, SNX10-related 24-0040 Osteopetrosis, infantile form, with nervous system involvement, OSTM1-related 24-0050 Osteopetrosis, infantile form, osteoclast-poor with immunoglobulin deficiency, TNFRSF11A-related 24-0060 Osteopetrosis, intermediate form, TCIRG1-related 24-0070 Osteopetrosis, intermediate form, TNFSF11-related 24-0080 Osteopetrosis, intermediate form, PLEKHM-related 24-0090 Osteopetrosis, intermediate form, CLCN7-related 24-0100 Osteopetrosis, late-onset, dominant form, CLCN7-related 24-0110 Osteopetrosis with renal tubular acidosis, CA2-related 24-0120 Osteopetrosis with ectodermal dysplasia and immune defect (OLEDAID), IKBKG-related 24-0130 Osteopetrosis, moderate form, SLC4A2-related 24-0140 Osteopetrosis, moderate form with defective leucocyte adhesion, FERMT3-related 24-0150 Osteopetrosis, moderate form with defective leucocyte adhesion, RASGRP2-related	低ホスファターゼ症歯限局型 大理石骨病, 重症新生児型/乳児型 大理石骨病, 重症新生児型/乳児型 大理石骨病, 重症新生児型/乳児型 大理石骨病, 乳児型, 神経系の罹患を伴う 大理石骨病, 乳児型, 免疫グロブリン欠乏を伴う破骨細胞減少型 大理石骨病, 中間型 大理石骨病, 中間型 大理石骨病, 中間型 大理石骨病, 中間型 大理石骨病, 遅発型2型 腎細管性アシドーシスを伴う大理石骨病 外胚葉異形成と免疫不全を伴う大理石骨病 なし 大理石骨病, 白血球接着不全を伴う中等症型 大理石骨病, 白血球接着不全を伴う中等症型	病名に "Osteopetrosis" が含まれているもののみを挙げた。
単一	骨系統疾患8		なし 多発性軟骨性外骨腫症	30 Disorganized development of skeletal components group	30-0010; 30-0020 1: Multiple cartilaginous exostoses, EXT1-related; 2: Multiple cartilaginous exostoses, EXT2-related	多発性軟骨性外骨腫症 病名に "exostosis" が含まれているもののみを挙げた。	
単一	骨系統疾患9		なし 内軟骨腫症	30 Disorganized development of skeletal components group	30-0140から 30-0170 14, 15: Enchondromatosis, IDH1-related (Ollier disease); Enchondromatosis, IDH2-related (Ollier disease) 16, 17: Enchondromatosis with hemangiomas, IDH1-related (Maffucci disease); Enchondromatosis with hemangiomas, IDH2-related ((Maffucci disease))	14, 15: 内軟骨腫症 (Ollier); 16, 17: 血管腫を伴う内軟骨腫症 (Maffucci) 病名に "Enchondromatosis" が含まれているもののみを挙げた。	
単一	骨系統疾患10		なし 2型コラーゲン異常症関連疾患	2 Type 2 collagen disorders	2-0010 Achondrogenesis, COL2A1-related (formerly type 2, type Langer-Saldino) 2-0020 Hypochondrogenesis, COL2A1-related 2-0030 Platypondylic dysplasia, type Torrance, COL2A1-related 2-0040 Spondyloepiphyseal dysplasia congenita (SEDC), COL2A1-related 2-0050 Spondyloepimetaphyseal dysplasia, COL2A1-related 2-0060 Kniest dysplasia, COL2A1-related 2-0070 Spondyloperipheral dysplasia, COL2A1-related 2-0080 SED with metatarsal shortening, COL2A1-related 2-0090 Stickler syndrome, COL2A1-related 2-0100 Dysplasia of the proximal femoral epiphyses, COL2A1-related	軟骨無発生症2型 軟骨低発生症 扁平椎異形成症, Torrance 型 先天性脊椎骨端異形成症 なし Kniest 骨異形成症 脊椎末梢異形成症 中足骨短縮を伴う脊椎骨端形成症 Stickler 症候群1型 大腿骨近位骨端異形成症	
単一	骨系統疾患11		なし 点状軟骨異形成症(ペルオキシソーム病を除く) X連鎖性潜性末節骨短縮型点状軟骨異形成症(CDPX1) " X染色体顕性Conradi-Hünermann型点状軟骨異形成症(CDPX2) " CHILD症候群(先天性片側異形成、魚鱗癬、四肢欠損) " Keutel症候群 " Greenberg骨異形成症 " 近位肢型点状軟骨異形成症 " 脛骨・中手骨型点状軟骨異形成症 ワルファリン胎芽症、SLE胎芽症、ビタミンK欠乏症などの非遺伝性疾患に合併する症候性の点状軟骨異形成も存在する。	23 Chondrodysplasia punctata (CDP) group	23-0010 CDP, X-linked recessive, ARSE-related (brachytelephalangic type; CDPX1) 23-0020 CDP, X-linked dominant, EBP-related (Conradi-Hu" nermann type; CDPX2) 23-0030 Congenital hemidysplasia, ichthyosis, limb defects (CHILD) syndrome, NSDHL-related 23-0040 Keutel syndrome, MGP-related 23-0050 Greenberg dysplasia, LBR-related 23-0060 Rhizomelic CDP, PEX7-related 23-0070 Rhizomelic CDP, DHPAT-related 23-0080 Rhizomelic CDP, ACP5-related 23-0090 Rhizomelic CDP, FAR1-related 23-0100 Rhizomelic CDP, PEX5-related 23-0110 CDP tibial-metacarpal type	点状軟骨異形成症, X連鎖性潜性、末節骨短縮型 (CDPX1) 点状軟骨異形成症, X染色体顕性, Conradi-Hünermann 型 (CDPX2) CHILD症候群 (先天性片側異形成, 魚鱗癬, 四肢欠損) Keutel症候群 Greenberg 骨異形成症 近位肢型点状軟骨異形成症 近位肢型点状軟骨異形成症 近位肢型点状軟骨異形成症 近位肢型点状軟骨異形成症 点状軟骨異形成症, 脛骨・中手骨型	疾患名に "CDP" を含むもののみを挙げた。 ※ 「Rhizomelic CDP」は、Peroxisome 病
単一	骨系統疾患12		なし 偽性軟骨無形性症	9 Pseudoachondroplasia and the multiple epiphyseal dysplasias	9-0010 Pseudoachondroplasia, COMP-related	偽性軟骨無形性症	
単一	骨系統疾患13		なし ラーセン症候群	6 Filamins and related disorders	6-0090 Larsen syndrome, FLNB-related	Larsen症候群	
単一	骨系統疾患14	骨・関節疾患	272 進行性骨化性線維異形成症	30 Disorganized development of skeletal components group	30-0080 Fibrodysplasia ossificans progressiva, ACVR1-related	進行性骨化性線維異形成症	
単一	骨系統疾患15		なし TRPV4異常症 変容性骨異形成症 " 脊椎骨端骨幹端異形成症Maroteaux型 " 脊椎骨幹端異形成症Kozlowski型 " 短体幹症 " 短指を伴う家族性指関節症	8 TRPV4 disorders	8-0010 から 8-0050	変容性骨異形成症 脊椎骨端骨幹端異形成症 Maroteaux型 (偽性Morquio症候群2型) 脊椎骨幹端異形成症Kozlowski型 短体幹症 (常染色体顕性型) 短指を伴う家族性指関節症	

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単一	骨系統疾患16		なし	骨硬化性疾患	24	24-0160 Osteosclerotic metaphyseal dysplasia, LRKK1-related	骨硬化性骨幹端異形成症		
					24-0170 Pyknodysostosis, CTSK-related	濃化異骨症			
					24-0180 Dysosteosclerosis, SLC29A3-related	異骨性骨硬化症			
					24-0190 Dysosteosclerosis, TNFRSF11A-related	異骨性骨硬化症			
					24-0200 Dysosteosclerosis with degenerative encephalopathy and brain malformation, CSF1R-related	なし			
					25	25:Osteosclerotic disorders	25-0010 Desmosterolosis, DHCR4-related		デスモステロール症
					25-0020 Raine dysplasia, FAM20C-related	Raine 骨異形成症			
					25-0030 Caffey disease, COL1A1-related	Caffey 病 (乳児型・寛解型)			
					25-0040 Caffey dysplasia (severe variants with prenatal onset)	Caffey 病 (出生前発症の重症型)			
					25-0050 Dysplastic cortical hyperostosis, Kozlowski-Tsuruta type	異形成性皮質骨増殖症, Kozlowski-Tsuruta型			
					25-0060 Dysplastic cortical hyperostosis, Al-Gazali type	異形成性皮質骨増殖症, Al-Gazali型			
					25-0070 Osteopoikilosis, LEMD3-related	骨斑紋症			
					25-0080 Melorheostosis with osteopoikilosis, LEMD3-related	骨斑紋症を伴う流蝕骨症			
					25-0090 Melorheostosis, MAP2K1-related	流蝕骨症 (モロレストーシス)			
					25-0100 Osteopathia striata with cranial sclerosis (OSCS), AMER1-related	頭蓋骨硬化を伴う骨線条症			
					25-0110 Pyle disease, SFRP4-related	Pyle 病			
					25-0120 Craniometaphyseal dysplasia, ANKH-related	頭蓋骨幹端異形成症			
					25-0130 Craniometaphyseal dysplasia, GJA1-related	頭蓋骨幹端異形成症			
					25-0140 Diaphyseal dysplasia Camurati-Engelmann, TGFB1-related	骨幹異形成症 Camurati-Engelmann 病			
					25-0150 Hyperostosis-Hyperphosphatemia syndrome, GALNT3-related	骨増殖症・高リン血症症候群			
					25-0160 Hyperostosis-Hyperphosphatemia syndrome, FGF23-related	骨増殖症・高リン血症症候群			
					25-0170 Hyperostosis-Hyperphosphatemia syndrome, KL-related	骨増殖症・高リン血症症候群			
					25-0180 Cerebellar hypoplasia-endosteal sclerosis, POLR3B-related	小脳低形成・骨内膜性硬化症			
					25-0190 Hematodiaphyseal dysplasia Ghosal, TBXAS1-related	血液骨幹異形成症 Ghosal			
					25-0200 Hypertrophic osteoarthropathy, HPGD-related	過形成性骨関節症			
					25-0210 Hypertrophic osteoarthropathy, SLC02A1-related	過形成性骨関節症			
					25-0220 Oculodentosseous dysplasia (ODOD), GJA1-related, dominant, mild type	眼歯骨異形成症 (ODOD) 軽症型			
25-0230 Oculodentosseous dysplasia (ODOD) GJA1-related, recessive, severe type	眼歯骨異形成症 (ODOD) 重症型								
25-0240 Osteoectasia with hyperphosphatemia (juvenile Paget disease), OPG-related	高ホスファターゼ症を伴う骨肥大症 (若年性Paget 病)								
25-0250 Osteosclerosis, LRP5-related	骨硬化症								
25-0260 Sclerosteosis, SOST-related	硬化性骨症								
25-0270 Sclerosteosis, LRP4-related	硬化性骨症								
25-0280 Endosteal hyperostosis, van Buchem type, SOST-related	骨内膜性骨増殖症, van Buchem型								
25-0290 Endosteal hyperostosis, Worth type	骨内膜性骨増殖症, Worth型								
25-0300 Craniodiaphyseal dysplasia, SOST-related	頭蓋骨幹端異形成症								
25-0310 Craniodiaphyseal dysplasia, SP7-related	頭蓋骨幹端異形成症								
25-0320 Trichodentosseous dysplasia, DLX3-related	毛髪歯骨異形成症								
25-0330 Diaphyseal medullary stenosis with malignant fibrous histiocytoma, MTAP-related	悪性繊維性組織球腫を伴う骨幹部骨髄腔狭窄症								
25-0340 Craniotubular dysplasia, TMEM53-related	なし								
25-0350 Craniometadiaphyseal dysplasia, Wormian bone type	頭蓋骨幹端骨幹異形成症, Worm骨型								
25-0360 Lenz-Majewski hyperostotic dysplasia, PTDSS1-related	Lenz-Majewski 骨増殖異形成症								
25-0370 Osteochondrodysplasia with hypertrichosis (Cantu syndrome), ABCC9-related	なし								
25-0380 Familial Paget disease of bone, SQSTM1-related	なし								
25-0390 Inclusion body myopathy, Paget disease of bone and frontotemporal dementia	なし								
25-0400 Endosteal hyperostosis, oligodontia, short stature, facial dysmorphism and intellectual disability, POLR3GL-related	なし								
25-0410 Metaphyseal dysplasia, Braun-Tinschert type	骨幹端異形成症, Braun-Tinschert型 A								
25-0420 Trichothiodystrophy with axial osteosclerosis	中央骨硬化症を伴う中間肢硫黄欠乏性毛髪異常								
単一	骨系統疾患17		なし	ピーلز症候群	31	Overgrowth (tall stature) syndromes and segmental overgrowth	31-0020 Congenital contractural arachnodactyly (Beals-Hecht syndrome), FBN2-related	先天性拘縮性くも状指症	
多因子	なし	骨・関節疾患	68	黄色靱帯骨化症					
多因子	なし	骨・関節疾患	69	後縦靱帯骨化症					
多因子	なし	骨・関節疾患	70	広範脊柱管狭窄症					
多因子	なし	骨・関節疾患	71	特発性大脳骨頭壊死症					
多因子	なし	骨・関節疾患	271	強直性脊椎炎					
多因子	なし		なし	びまん性特発性骨増殖症 (DISH: Diffuse Idiopathic Skeletal Hyperostosis)					
多因子	なし		なし	乾癬性関節炎					
多因子	なし		なし	炎症性腸疾患関連関節炎					
多因子	なし		なし	SAPHO症候群					