Panelinformation Skelettdysplasi in-silico

Version: 2.0

Gen	Associerad fenotyp, MIM# (OMIM)	Nedärvning
ALPL	Hypophosphatasia, adult , 146300	AD, AR
	Hypophosphatasia, childhood, 241510	AR
	Hypophosphatasia, infantile, 241500	AR
	Odontohypophosphatasia, 146300	AD, AR
ARSB	Mucopolysaccharidosis type VI (Maroteaux-Lamy), 253200	AR
ARSE	Chondrodyplasia punctata, X-linked recessive, 302950	XLR
COL1A1	Caffey disease, 114000	AD
	Ehlers-Danlos syndrome, classic, 130000	AD
	Ehlers-Danlos syndrome, type VIIA, 130060	AD
	Osteogenesis imperfecta, type I, 166200	AD
	Osteogenesis imperfecta, type II, 166210	AD
	Osteogenesis imperfecta, type III, 259420	AD
	Osteogenesis imperfecta, type IV, 166220	AD
	{Bone mineral density variation QTL, osteoporosis}, 166710	AD
COL1A2	Ehlers-Danlos syndrome, cardiac valvular form, 225320	AR
	Ehlers-Danlos syndrome, type VIIB, 130060	AD
	Osteogenesis imperfecta, type II, 166210	AD
	Osteogenesis imperfecta, type III, 259420	AD
	Osteogenesis imperfecta, type IV, 166220	AD
	{Osteoporosis, postmenopausal}, 166710	AD
COL9A1	?Epiphyseal dysplasia, multiple, 6, 614135	AD
	Stickler syndrome, type IV, 614134	
COL9A2	?Stickler syndrome, type V, 614284	AR
	Epiphyseal dysplasia, multiple, 2, 600204	AD
COL9A3	Epiphyseal dysplasia, multiple, 3, with or without myopathy, 600969	AD
	{Intervertebral disc disease, susceptibility to}, 603932	
COL10A1	Metaphyseal chondrodysplasia, Schmid type, 156500	AD
COL11A1	Fibrochondrogenesis 1, 228520	AR
	Marshall syndrome, 154780	AD
	Stickler syndrome, type II, 604841	AD
	{Lumbar disc herniation, susceptibility to}, 603932	
COL11A2	Deafness, autosomal dominant 13, 601868	AD
	Deafness, autosomal recessive 53, 609706	AR
	Fibrochondrogenesis 2, 614524	AD, AR
	Otospondylomegaepiphyseal dysplasia, autosomal dominant, 184840	AD
	Otospondylomegaepiphyseal dysplasia, autosomal recessive, 215150	AR

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COL2A1	Achondrogenesis, type II or hypochondrogenesis, 200610	AD
	Avascular necrosis of the femoral head, 608805	AD
	Czech dysplasia, 609162	AD
	Epiphyseal dysplasia, multiple, with myopia and deafness, 132450	AD
	Kniest dysplasia, 156550	AD
	Legg-Calve-Perthes disease, 150600	AD
	Osteoarthritis with mild chondrodysplasia, 604864	AD
	Platyspondylic skeletal dysplasia, Torrance type, 151210	AD
	SED congenita, 183900	AD
	SEMD Strudwick type, 184250	AD
	Spondyloepiphyseal dysplasia, Stanescu type, 616583	AD
	Spondyloperipheral dysplasia	AD
	Stickler sydrome, type I, nonsyndromic ocular, 271700	AD
	Stickler syndrome, type I, 108300	AD
	Vitreoretinopathy with phalangeal epiphyseal dysplasia	
COMP	Epiphyseal dysplasia, multiple, 1, 132400	AD
	Pseudoachondroplasia, 177170	AD
DDR2	Spondylometaepiphyseal dysplasia, short limb-hand type, 271665	AR
EBP	Chondrodysplasia punctata, X-linked dominant, 302960	XLD
	MEND syndrome, 300960	XLR
FGFR3	Achondroplasia, 100800	AD
	Bladder cancer, somatic, 109800	
	CATSHL syndrome, 610474	AD, AR
	Cervical cancer, somatic, 603956	
	Colorectal cancer, somatic, 114500	
	Crouzon syndrome with acanthosis nigricans, 612247	AD
	Hypochondroplasia, 146000	AD
	LADD syndrome, 149730	AD
	Muenke syndrome, 602849	AD
	Nevus, epidermal, somatic	
	SADDAN, 162900	AD
	Spermatocytic seminoma, somatic, 273300	
	Thanatophoric dysplasia, type I, 187600	AD
	Thanatophoric dysplasia, type II, 187601	AD

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FLNA	Cardiac valvular dysplasia, X-linked, 314400	XLR
	Congenital short bowel syndrome, 300048	XLR
	FG syndrome 2, 300321	
	Frontometaphyseal dysplasia 1, 305620	XLR
	Heterotopia, periventricular	XLD
	Intestinal pseudoobstruction, neuronal, 300049	XLR
	Melnick-Needles syndrome, 309350	XLD
	Otopalatodigital syndrome, type I, 311300	XLD
	Otopalatodigital syndrome, type II, 304120	XLD
	Terminal osseous dysplasia, 300244	
FLNB	Atelosteogenesis, type I, 108720	AD
	Atelosteogenesis, type III, 108721	AD
	Boomerang dysplasia, 112310	AD
	Larsen syndrome, 150250	AD
	Spondylocarpotarsal synostosis syndrome, 272460	AR
GALNS	Mucopolysaccharidosis IVA, 253000	AR
GLB1	GM1-gangliosidosis, type I, 230500	AR
	GM1-gangliosidosis, type II, 230600	AR
	GM1-gangliosidosis, type III, 230650	AR
	Mucopolysaccharidosis type IVB (Morquio), 253010	AR
GSC	Short stature, auditory canal atresia, mandibular hypoplasia, skeletal abnormalities, 602471	AR
GUSB	Mucopolysaccharidosis VII, 253220	AR
HGSNAT	Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930	AR
	Retinitis pigmentosa 73, 616544	AR
HSPG2	Dyssegmental dysplasia, Silverman-Handmaker type, 224410	AR
	Schwartz-Jampel syndrome, type 1, 255800	AR
HYAL1	?Mucopolysaccharidosis type IX, 601492	AR
IDS	Mucopolysaccharidosis II, 309900	XLR
IDUA	Mucopolysaccharidosis Ih, 607014	AR
	Mucopolysaccharidosis Ih/s, 607015	AR
	Mucopolysaccharidosis Is, 607016	AR
INPPL1	Opsismodysplasia, 258480	AR
LBR	?Reynolds syndrome, 613471	AD
	Greenberg skeletal dysplasia, 215140	AR
	Pelger-Huet anomaly, 169400	AD
LIFR	Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome, 601559	AR
MATN3	?Spondyloepimetaphyseal dysplasia, 608728	AR
	Epiphyseal dysplasia, multiple, 5, 607078	AD
	{Osteoarthritis susceptibility 2}, 140600	AD

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MMP9	Metaphyseal anadysplasia 2, 613073	
MMP13	Metaphyseal anadysplasia 1, 602111	AD
	Metaphyseal dysplasia, Spahr type, 250400	AR
	Spondyloepimetaphyseal dysplasia, Missouri type, 602111	AD
NAGLU	?Charcot-Marie-Tooth disease, axonal, type 2V, 616491	AD
	Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920	AR
NKX3-2	Spondylo-megaepiphyseal-metaphyseal dysplasia, 613330	AR
NSDHL	CHILD syndrome, 308050	XLD
	CK syndrome, 300831	XLR
PEX7	Peroxisome biogenesis disorder 9B, 614879	
	Rhizomelic chondrodysplasia punctata, type 1, 215100	AR
PTH1R	Chondrodysplasia, Blomstrand type, 215045	AR
	Eiken syndrome, 600002	AR
	Failure of tooth eruption, primary, 125350	AD
	Metaphyseal chondrodysplasia, Murk Jansen type, 156400	AD
RMRP	Anauxetic dysplasia 1, 607095	AR
	Cartilage-hair hypoplasia, 250250	AR
	Metaphyseal dysplasia without hypotrichosis, 250460	AR
SBDS	Shwachman-Diamond syndrome, 260400	AR
	{Aplastic anemia, susceptibility to}, 609135	
SGSH	Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900	AR
SHOX	Langer mesomelic dysplasia, 249700	AR
	Leri-Weill dyschondrosteosis, 127300	AD
	Short stature, idiopathic familial, 300582	
SLC26A2	Achondrogenesis Ib, 600972	AR
	Atelosteogenesis II, 256050	AR
	De la Chapelle dysplasia, 256050	AR
	Diastrophic dysplasia, 222600	AR
	Diastrophic dysplasia, broad bone-platyspondylic variant, 222600	AR
	Epiphyseal dysplasia, multiple, 4, 226900	AR
SOX9	Acampomelic campomelic dysplasia, 114290	AD
	Campomelic dysplasia, 114290	AD
	Campomelic dysplasia with autosomal sex reversal, 114290	AD
TRIP11	Achondrogenesis, type IA, 200600	AR

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TRPV4	?Avascular necrosis of femoral head, primary, 2, 617383	AD
	Brachyolmia type 3, 113500	AD
	Digital arthropathy-brachydactyly, familial, 606835	AD
	Hereditary motor and sensory neuropathy, type Iic, 606071	AD
	Metatropic dysplasia, 156530	AD
	Parastremmatic dwarfism, 168400	AD
	Scapuloperoneal spinal muscular atrophy, 181405	AD
	SED, Maroteaux type, 184095	AD
	Spinal muscular atrophy, distal, congenital nonprogressive, 600175	AD
	Spondylometaphyseal dysplasia, Kozlowski type, 184252	AD
	[Sodium serum level QTL 1], 613508	
VPS33A	Mucopolysaccharidosis-plus syndrome, 617303	AR

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