Panelinformation Skelettdysplasi in-silico

Version: 1.0

Gen	Transkript*	% täckning > 10x **	% täckning > 20x**	x Median täckning **	Associerad fenotyp, MIM# (OMIM)	Nedärvning
ALPL	NM_000478.5	100	100	146	Hypophosphatasia, adult , 146300	AD, AR
					Hypophosphatasia, childhood, 241510	AR
					Hypophosphatasia, infantile, 241500	AR
					Odontohypophosphatasia, 146300	AD, AR
ARSE	NM_000047.2	100	100	169	Chondrodyplasia punctata, X-linked recessive, 302950	XLR
COL1A1	NM_000088.3	100	100	196	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	NM_000089.3	99	99	85	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	NM_001851.4	99	97	72	?Epiphyseal dysplasia, multiple, 6, 614135	AD
					Stickler syndrome, type IV, 614134	
COL9A2	NM_001852.3	100	100	130	?Stickler syndrome, type V, 614284	AR
					Epiphyseal dysplasia, multiple, 2, 600204	AD

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COL9A3	NM_001853.3	100	100	122	Epiphyseal dysplasia, multiple, 3, with or without myopathy, 600969 {Intervertebral disc disease, susceptibility to}, 603932	AD
COL10A1	NM_000493.3	100	100	90	Metaphyseal chondrodysplasia, Schmid type, 156500	AD
COL11A1	NM_080629.2	100	98	57	Fibrochondrogenesis 1, 228520 Marshall syndrome, 154780 Stickler syndrome, type II, 604841 {Lumbar disc herniation, susceptibility to}, 603932	AR AD AD
COL11A2	NM_080680.2	100	99	138	Deafness, autosomal dominant 13, 601868 Deafness, autosomal recessive 53, 609706 Fibrochondrogenesis 2, 614524 Otospondylomegaepiphyseal dysplasia, autosomal dominant, 184840 Otospondylomegaepiphyseal dysplasia, autosomal recessive, 215150	AD AR AD, AR AD AR
COL2A1	NM_001844.4	100	100	152	Achondrogenesis, type II or hypochondrogenesis, 200610 Avascular necrosis of the femoral head, 608805 Czech dysplasia, 609162 Epiphyseal dysplasia, multiple, with myopia and deafness, 132450 Kniest dysplasia, 156550 Legg-Calve-Perthes disease, 150600 Osteoarthritis with mild chondrodysplasia, 604864 Platyspondylic skeletal dysplasia, Torrance type, 151210 SED congenita, 183900 SEMD Strudwick type, 184250 Spondyloepiphyseal dysplasia, Stanescu type, 616583 Spondyloperipheral dysplasia Stickler sydrome, type I, nonsyndromic ocular, 271700 Stickler syndrome, type I, 108300 Vitreoretinopathy with phalangeal epiphyseal dysplasia	AD AD AD AD AD AD AD AD AD AD AD AD AD AD AD

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COMP	NM_000095.2	98	96	141	Epiphyseal dysplasia, multiple, 1, 132400 Pseudoachondroplasia, 177170	AD AD
DDR2	NM_001014796.2	100	100	104	Spondylometaepiphyseal dysplasia, short limb-hand type, 271665	AR
EBP	NM_006579.2	100	100	168	Chondrodysplasia punctata, X-linked dominant, 302960 MEND syndrome, 300960	XLD XLR
FGFR3	NM_000142.4	100	100	128	Achondroplasia, 100800 Bladder cancer, somatic, 109800 CATSHL syndrome, 610474 Cervical cancer, somatic, 603956 Colorectal cancer, somatic, 114500 Crouzon syndrome with acanthosis nigricans, 612247 Hypochondroplasia, 146000 LADD syndrome, 149730 Muenke syndrome, 602849 Nevus, epidermal, somatic SADDAN, 162900 Spermatocytic seminoma, somatic, 273300 Thanatophoric dysplasia, type I, 187600 Thanatophoric dysplasia, type II, 187601	AD AD, AR AD AD AD AD AD AD AD

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FLNA	NM_001110556.1	100	100	160	Cardiac valvular dysplasia, X-linked, 314400 Congenital short bowel syndrome, 300048 FG syndrome 2, 300321 Frontometaphyseal dysplasia 1, 305620 Heterotopia, periventricular Intestinal pseudoobstruction, neuronal, 300049 Melnick-Needles syndrome, 309350 Otopalatodigital syndrome, type I, 311300 Otopalatodigital syndrome, type II, 304120 Terminal osseous dysplasia, 300244	XLR XLR XLR XLD XLR XLD XLD XLD
FLNB	NM_001164317.1	100	100	161	Atelosteogenesis, type I, 108720 Atelosteogenesis, type III, 108721 Boomerang dysplasia, 112310 Larsen syndrome, 150250 Spondylocarpotarsal synostosis syndrome, 272460	AD AD AD AD AR
GSC	NM_173849.2	100	100	96	Short stature, auditory canal atresia, mandibular hypoplasia, skeletal abnormalities, 602471	AR
HSPG2	NM_005529.6	99	99	167	Dyssegmental dysplasia, Silverman-Handmaker type, 224410 Schwartz-Jampel syndrome, type 1, 255800	AR AR
INPPL1	NM_001567.3	99	98	132	Opsismodysplasia, 258480	AR
LBR	NM_194442.2	96	96	60	?Reynolds syndrome, 613471 Greenberg skeletal dysplasia, 215140 Pelger-Huet anomaly, 169400	AD AR AD
LIFR	NM_002310.5	87	73	57	Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome, 601559	AR
MATN3	NM_002381.4	88	84	108	?Spondyloepimetaphyseal dysplasia, 608728 Epiphyseal dysplasia, multiple, 5, 607078 {Osteoarthritis susceptibility 2}, 140600	AR AD AD

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MMP9	NM_004994.2	100	100	105	Metaphyseal anadysplasia 2, 613073	
MMP13	NM_002427.3	100	98	77	Metaphyseal anadysplasia 1, 602111 Metaphyseal dysplasia, Spahr type, 250400 Spondyloepimetaphyseal dysplasia, Missouri type, 602111	AD AR AD
NKX3-2	NM_001189.3	100	100	106	Spondylo-megaepiphyseal-metaphyseal dysplasia, 613330	AR
NSDHL	NM_001129765.1	100	100	140	CHILD syndrome, 308050 CK syndrome, 300831	XLD XLR
PEX7	NM_000288.3	100	94	76	Peroxisome biogenesis disorder 9B, 614879 Rhizomelic chondrodysplasia punctata, type 1, 215100	 AR
PTH1R	NM_000316.2	100	100	164	Chondrodysplasia, Blomstrand type, 215045 Eiken syndrome, 600002 Failure of tooth eruption, primary, 125350 Metaphyseal chondrodysplasia, Murk Jansen type, 156400	AR AR AD AD
RMRP	NR_003051.3	100	100	248	Anauxetic dysplasia 1, 607095 Cartilage-hair hypoplasia, 250250 Metaphyseal dysplasia without hypotrichosis, 250460	AR AR AR
SBDS	NM_016038.3	100	95	52	Shwachman-Diamond syndrome, 260400 {Aplastic anemia, susceptibility to}, 609135	AR
SHOX	NM_006883.2	93	93	178	Langer mesomelic dysplasia, 249700 Leri-Weill dyschondrosteosis, 127300 Short stature, idiopathic familial, 300582	AR AD
SLC26A2	NM_000112.3	100	100	71	Achondrogenesis Ib, 600972 Atelosteogenesis II, 256050 De la Chapelle dysplasia, 256050 Diastrophic dysplasia, 222600 Diastrophic dysplasia, broad bone-platyspondylic variant, 222600 Epiphyseal dysplasia, multiple, 4, 226900	AR AR AR AR AR AR

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SOX9	NM_000346.3	100	99	105	Acampomelic campomelic dysplasia, 114290 Campomelic dysplasia, 114290 Campomelic dysplasia with autosomal sex reversal, 114290	AD AD AD
TRIP11	NM_004239.4	76	54	23	Achondrogenesis, type IA, 200600	AR
TRPV4	NM_021625.4	100	100	198	?Avascular necrosis of femoral head, primary, 2, 617383 Brachyolmia type 3, 113500 Digital arthropathy-brachydactyly, familial, 606835 Hereditary motor and sensory neuropathy, type Iic, 606071 Metatropic dysplasia, 156530 Parastremmatic dwarfism, 168400 Scapuloperoneal spinal muscular atrophy, 181405 SED, Maroteaux type, 184095 Spinal muscular atrophy, distal, congenital nonprogressive, 600175 Spondylometaphyseal dysplasia, Kozlowski type, 184252 [Sodium serum level QTL 1], 613508	AD AD AD AD AD AD AD AD AD AD

* Referenstranskript enligt NCBI Reference Sequences (RefSeq). Detta transkriptnummer används som utgångspunkt för analys av horisontell täckning. Däremot görs varianttolkning från genens hela kodande sekvens, oberoende av isoform.
** Medelvärde av 32 valideringsprov

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