Displasias esqueléticas

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RESEARCH ARTICLE

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Nosology and Classification of Genetic Skeletal Disorders: 2015 Revision

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Group/Name of Disorder	Inheritance	MIM No.	Locus or Gene	
1. FGFR3 chondrodysplasia group				
Thanatophoric dysplasia type 1 (TD1)	AD	187600	*FGFR3*	F
Thanatophoric dysplasia type 2 (TD2)	AD	187601	*FGFR3*	F
Severe achondroplasia with developmental delay and acanthosis nigricans (SADDAN)	AD	187600	*FGFR3*	F
Achondroplasia	AD	100800	*FGFR3*	F
Hypochondroplasia	AD	146000	*FGFR3*	F
Camptodactyly, tall stature and hearing loss syndrome (CATSHL)	AD	610474	*FGFR3*	F
Hypochondroplasia–like dysplasia(s)	AD, SP			
See also group 33 for craniosynostoses syndromes linked to *FGFR3* mutations, as well as LADD syndrome in group 41 for another *FGFR3*-related phenotype				
2. Type 2 collagen group				
Achondrogenesis type 2 (ACG2; Langer-Saldino)	AD	200610	*COL2A1*	T
Platyspondylic dysplasia, Torrance type	AD	151210	*COL2A1*	T
Hypochondrogenesis	AD	200610	*COL2A1*	T
Spondyloepiphyseal dysplasia congenital (SEDC)	AD	183900	*COL2A1*	T
Spondyloepimetaphyseal dysplasia (SEMD) Strudwick type	AD	184250	*COL2A1*	T
Kniest dysplasia	AD	156550	*COL2A1*	T
Spondyloperipheral dysplasia	AD	271700	*COL2A1*	T
Mild SED with premature onset arthrosis	AD		*COL2A1*	T
SED with metatarsal shortening (formerly Czech dysplasia)	AD	609162	*COL2A1*	T
Stickler syndrome type 1	AD	108300	*COL2A1*	T
3. Type 11 collagen group				
Stickler syndrome type 2	AD	604841	*COL11A1*	T
Marshall syndrome	AD	154780	*COL11A1*	T

Group/Name of Disorder	Inheritance	MIM No.	Locus or Gene
4. Sulphation disorders group			
Achondrogenesis type 1B (ACG1B)	AR	600972	*DTDST*
Atelosteogenesis type 2 (AO2)	AR	256050	*DTDST*
Diastrophic dysplasia (DTD)	AR	222600	*DTDST*
MED, autosomal recessive type (rMED; EDM4)	AR	226900	*DTDST*
SEMD, PAPSS2 type	AR	612847	*PAPSS2*
Brachyolmia, recessive type	AR	612847	*PAPSS2*
Chondrodysplasia gPAPP type (includes Catel–Manzke-like syndrome	AR	614078	*IMPAD1*
Chondrodysplasia with congenital joint dislocations, CHST3 type (recessive Larsen syndrome)	AR	608637	*CHST3*
Ehlers–Danlos syndrome, CHST14 type ("musculo-skeletal variant")	AR	601776	*CHST14*
See also group 7 and group 20 for other conditions with multiple dislocations.			
5. Perlecan group			
Dyssegmental dysplasia, Silverman–Handmaker type	AR	224410	*PLC (HSPG2)*
Dyssegmental dysplasia, Rolland–Desbuquois type	AR	224400	*PLC (HSPG2)*
Schwartz–Jampel syndrome (myotonic chondrodystrophy)	AR	255800	*PLC (HSPG2)*
6. Aggrecan group			
SED, Kimberley type	AD	608361	*AGC1*
SEMD, Aggrecan type	AR	612813	*AGC1*

7. Filamin group and related disorders			
Frontometaphyseal dysplasia	XLD	305620	*FLNA*
Osteodysplasty Melnick–Needles	XLD	309350	*FLNA*
Otopalatodigital syndrome type 1 (OPD1)	XLD	311300	*FLNA*
Otopalatodigital syndrome type 2 (OPD2)	XLD	304120	*FLNA*
Terminal osseous dysplasia with pigmentary defects (TODPD)	XLD	300244	*FLNA*
Atelosteogenesis type 1 (AO1)	AD	108720	*FLNB*
Atelosteogenesis type 3 (AO3)	AD	108721	*FLNB*
Larsen syndrome (dominant)	AD	150250	*FLNB*
Spondylo-carpal-tarsal dysplasia	AR	272460	*FLNB*
Frank-ter Haar syndrome	AR	249420	*SH3PXD2B*
(see also group 4 for recessive Larsen syndrome and group 20 for conditions with multiple dislocations)			
8. TRPV4 group			
Metatropic dysplasia	AD	156530	*TRPV4*
Spondyloepimetaphyseal dysplasia, Maroteaux type (Pseudo-Morquio syndrome type 2)	AD	184095	*TRPV4*

9. Ciliopathies with major skeletal involvement			
Chondroectodermal dysplasia (Ellis-van Creveld)	AR	225500	*EVC1* *EVC2*
Short rib–polydactyly syndrome (SRPS) type 1/3 (Saldino–Noonan/Verma–Naumoff)	AR	208500	*DYNC2H1*
		613091	*IFT80*
			WDR34
Asphyxiating thoracic dysplasia (ATD; Jeune)	AR	263510	*DYNC2H1*
			IFT80
			WDR34 *TTC21B*
			WDR19 *IFT172* *IFT140*

10. Multiple epiphyseal dysplasia and pseudoachondroplasia group			
Pseudoachondroplasia (PSACH)	AD	177170	*COMP*
Multiple epiphyseal dysplasia (MED) type 1 (EDM1)	AD	132400	*COMP*
Multiple epiphyseal dysplasia (MED) type 2 (EDM2)	AD	600204	*COL9A2*
Multiple epiphyseal dysplasia (MED) type 3 (EDM3)	AD	600969	*COL9A3*
Multiple epiphyseal dysplasia (MED) type 5 (EDM5)	AD	607078	*MATN3*
Multiple epiphyseal dysplasia (MED) type 6 (EDM6)	AD	120210	*COL9A1*
Multiple epiphyseal dysplasia (MED), other types			
Stickler syndrome, recessive type	AR	120210	*COL9A1*
Familial hip dysplasia (Beukes)	AD	142669	4q35
Multiple epiphyseal dysplasia with microcephaly and nystagmus (Lowry–Wood)	AR	226960	
See also Multiple Epiphyseal Dysplasia, recessive type (rMED; EDM4) in sulphation disorders (group 4), Familial osteochondritis dissecans in the Aggrecan group, as well as ASPED in the Acromelic group			
11. Metaphyseal dysplasias			
Metaphyseal dysplasia, Schmid type (MCS)	AD	156500	*COL10A1*
Cartilage-hair hypoplasia (CHH; metaphyseal dysplasia, McKusick type)	AR	250250	*RMRP*
Metaphyseal dysplasia, CHH-like, POP1 type	AR		*POP1*
Metaphyseal dysplasia, Jansen type	AD	156400	*PTHR1*

12. Spondylometaphyseal dysplasias (SMD)			
Spondyloenchondrodysplasia (SPENCD)	AR	271550	*ACP5*
Odontochondrodysplasia (ODCD)	AR	184260	
SMD, Sutcliffe type or corner fractures type	AD	184255	
SMD with cone-rod dystrophy	AR	608940	*PCYT1A*
SMD with retinal degeneration, axial type	AR	602271	
See also SMD Kozlowski (group TRPV4) as well as SMD Sedaghatian type in group 14; there are many individual reports of SMD variants.			
13. Spondylo-epi-(meta)-physeal dysplasias (SE(M)D)			
Dyggve–Melchior–Clausen dysplasia (DMC)	AR	223800	*DYM*
		615222	*RAB33B*
Immuno-osseous dysplasia (Schimke)	AR	242900	*SMARCAL1*

14. Severe spondylodysplastic dysplasias			
Achondrogenesis type 1A (ACG1A)	AR	200600	*TRIP11*
Schneckenbecken dysplasia	AR	269250	*SLC35D1*
Spondylometaphyseal dysplasia, Sedaghatian type	AR	250220	*GPX4*
Severe spondylometaphyseal dysplasia (SMD Sedaghatian-like)	AR		*SBDS*
Opsismodysplasia	AR	258480	*INPPL1*
MAGMAS related skeletal dysplasia	AR		*MAGMAS*
See also: Thanatophoric dysplasia, types 1 and 2 (group 1); ACG2 and Torrance dysplasia (group 2); Fibrochondrogenesis (group 3); Achondrogenesis type 1B (group 4); and Metatropic Dysplasia (group 8)			

TABLE I. (*Continued*)

Group/Name of Disorder	Inheritance	MIM No.	Locus or Gene
15. Acromelic dysplasias			
Tricho-rhino-phalangeal dysplasia types 1/3	AD	190350	*TRPS1*
Tricho-rhino-phalangeal dysplasia type 2 (Langer–Giedion)	AD	150230	*TRPS1 and EXT1*

16. Acromesomelic dysplasias			
Acromesomelic dysplasia type Maroteaux (AMDM)	AR	602875	*NPR2*
Grebe dysplasia	AR	200700	*GDF5*
Fibular hypoplasia and complex brachydactyly (Du Pan)	AR	228900	*GDF5*
Acromesomelic dysplasia with genital anomalies	AR	609441	*BMPR1B*
Acromesomelic dysplasia, Osebold-Remondini type	AD	112910	
17. Mesomelic and rhizo-mesomelic dysplasias			
Dyschondrosteosis (Leri–Weill)	Pseudo-AD	127300	*SHOX*
Langer type (homozygous dyschondrosteosis)	Pseudo-AR	249700	*SHOX*

18. Campomelic dysplasia and related disorders			
Campomelic dysplasia (CD)	AD	114290	*SOX9*
Stüve–Wiedemann dysplasia	AR	601559	*LIFR*
Kyphomelic dysplasia, several forms		211350	
See also group 33 for craniosynostoses syndromes linked to FGFR2			
19. Slender bone dysplasia group			
3-M syndrome	AR	273750	*CUL7*
		612921	*OBSL1*
		614205	*CCDC8*
Kenny–Caffey dysplasia	AR	244460	*TBCE*

20. Dysplasias with multiple joint dislocations			
Desbuquois dysplasia (with accessory ossification centre in digit 2)	AR	251450	*CANT1*
Desbuquois dysplasia with short metacarpals and elongated phalanges (Kim type)	AR	251450	*CANT1*
Desbuquois dysplasia type 2	AR	615777	*XYLT1*
Pseudodiastrophic dysplasia	AR	264180	
SEMD with joint laxity (SEMD-JL) leptodactylic or Hall type	AD	603546	*KIF22*
SEMD with joint laxity (SEMD-JL) Beighton type	AR	271640	*B3GALT6*
See also: SED with congenital dislocations, CHST3 type (group 4); Atelosteogenesis type 3 and Larsen syndrome (group 7)			
21. Chondrodysplasia punctata (CDP) group			
CDP, X-linked dominant, Conradi-Hünermann type (CDPX2)	XLD	302960	*EBP*
CDP, X-linked recessive, brachytelephalangic type (CDPX1)	XLR	302950	*ARSE*
CHILD (congenital hemidysplasia, ichthyosis, limb defects)	XLD	308050	*NSDHL*
Keutel syndrome	AR	245150	*MGP*
Greenberg dysplasia	AR	215140	*LBR*
Rhizomelic CDP type 1	AR	215100	*PEX7*
Rhizomelic CDP type 2	AR	222765	*DHPAT*

22. Neonatal osteosclerotic dysplasias			
Blomstrand dysplasia	AR	215045	*PTHR1*
Desmosterolosis	AR	602398	*DHCR24*
Caffey disease (including prenatal, infantile and attenuated forms)	AD	114000	*COL1A1*
Caffey dysplasia (severe variants with prenatal onset)	AR	114000	
Raine dysplasia (lethal and non-lethal forms)	AR	259775	*FAM20C*
See also Astley–Kendall dysplasia and CDPs in group 21			
23. Osteopetrosis and related disorders			
Osteopetrosis, severe neonatal or infantile forms (OPTB1)	AR	259700	*TCIRG1*
Osteopetrosis, severe neonatal or infantile forms (OPTB4)	AR	611490	*CLCN7*
Osteopetrosis, severe neonatal or infantile forms (OPTB8)	AR	615085	*SNX10*
Osteopetrosis, infantile form, with nervous system involvement (OPTB5)	AR	259720	*OSTM1*

24. Other sclerosing bone disorders			
Craniometaphyseal dysplasia, autosomal dominant type	AD	123000	*ANKH*
Diaphyseal dysplasia Camurati–Engelmann	AD	131300	*TGFB1*
Hematodiaphyseal dysplasia Ghosal	AR	231095	*TBXAS1*
Hypertrophic osteoarthropathy	AR	259100	*HPGD*
Pachydermoperiostosis (hypertrophic osteoarthropathy, primary, autosomal dominant)	AD	167100	
Oculo-dento-osseous dysplasia (ODOD) mild type	AD	164200	*GJA1*
Oculo-dento-osseous dysplasia (ODOD) severe type	AR	257850	*GJA1*
Osteoectasia with hyperphosphatasia (juvenile Paget disease)	AR	239000	*OPG*
Sclerosteosis	AR,AD	269500, 614305	*SOST*, *LRP4*
Endosteal hyperostosis, van Buchem type	AR	239100	*SOST*
Trichodentoosseous dysplasia	AD	190320	*DLX3*
Craniometaphyseal dysplasia, autosomal recessive type	AR	218400	*GJA1*
Diaphyseal medullary stenosis with malignant fibrous histiocytoma	AD	112250	
Craniodiaphyseal dysplasia	AD	122860	*SOST*
Craniometadiaphyseal dysplasia, Wormian bone type	AR	615118	
Endosteal sclerosis with cerebellar hypoplasia	AR	213002	
Lenz-Majewski hyperostotic dysplasia	SP	151050	*PTDSS1*
Metaphyseal dysplasia, Braun–Tinschert type	AD	605946	
Pyle disease	AR	265900	
25. Osteogenesis imperfecta and decreased bone density group			
For comments the classification of Osteogenesis imperfecta, please refer to the text			

26. Abnormal mineralization group			
Hypophosphatasia, perinatal lethal, infantile and juvenile forms	AR	241500	*ALPL*
Hypophosphatasia, juvenile and adult forms	AD	146300	*ALPL*
Hypophosphatemic rickets, X-linked dominant	XLD	307800	*PHEX*
Hypophosphatemic rickets, autosomal dominant	AD	193100	*FGF23*
Hypophosphatemic rickets, autosomal recessive, type 1 (ARHR1)	AR	241520	*DMP1*
Hypophosphatemic rickets, autosomal recessive, type 2 (ARHR2)	AR	613312	*ENPP1*
Hypophosphatemic rickets with hypercalciuria, X-linked recessive	XLR	300554	*CICN5*
Hypophosphatemic rickets with hypercalciuria, autosomal recessive (HHRH)	AR	241530	*SLC34A3*
Neonatal hyperparathyroidism, severe form	AR	239200	*CASR*
Familial hypocalciuric hypercalcemia with transient neonatal hyperparathyroidism	AD	145980	*CASR*
Calcium pyrophosphate deposition disease (familial chondrocalcisnosis) type 2	AD	118600	*ANKH*
See also Jansen dysplasia and Eiken dysplasia			
27. Lysosomal Storage Diseases with Skeletal Involvement (Dysostosis Multiplex group)			
Mucopolysaccharidosis type 1H/1S (Hurler, Hurler–Scheie, Scheie)	AR	607014	*IDA*
Mucopolysaccharidosis type 2 (Hunter)	XLR	309900	*IDS*
Mucopolysaccharidosis type 3A (Sanfilippo A)	AR	252900	*HSS*
Mucopolysaccharidosis type 3B (Sanfilippo B)	AR	252920	*NAGLU*
Mucopolysaccharidosis type 3C (Sanfilippo C)	AR	252930	*HSGNAT*

28. Osteolysis group			
Familial expansile osteolysis	AD	174810	*RANK (TNFRSF11A)*
Mandibuloacral dysplasia type A	AD	248370	*LMNA*
Mandibuloacral dysplasia type B	AR	608612	*ZMPSTE24*
Progeria, Hutchinson–Gilford type	AD	176670	*LMNA*
Torg–Winchester syndrome	AR	259600	*MMP2*
Hajdu–Cheney syndrome	AD	102500	*NOTCH2*
Multicentric carpal-tarsal osteolysis with and without nephropathy	AD	166300	*MAFB*
See also Pycnodysostosis, cleidocranial dysplasia, Keutel and Singleton–Merten syndrome. Note: several neurologic conditions may cause acroosteolysis			
29. Disorganized development of skeletal components group			
Multiple cartilaginous exostoses 1	AD	133700	*EXT1*
Multiple cartilaginous exostoses 2	AD	133701	*EXT2*
Multiple cartilaginous exostoses 3	AD	600209	

30. Overgrowth (tall stature) syndromes with skeletal involvement			
Weaver syndrome	SP/AD	277590	*EZH2*
Sotos syndrome	AD	117550	*NSD1*
Sotos-like syndrome	AD		*SETD2*
Marshall–Smith syndrome	SP	602535	*NFIX*
Proteus syndrome	SP	176920	*AKT1*
CLOVES	SP	612918	*PIK3CA*
Marfan syndrome	AD	154700	*FBN1*
Congenital contractural arachnodactyly	AD	121050	*FBN2*

31. Genetic inflammatory/rheumatoid-like osteoarthropathies			
Progressive pseudorheumatoid dysplasia (PPRD; SED with progressive arthropathy)	AR	208230	*WISP3*
Chronic infantile neurologic cutaneous articular syndrome (CINCA)/neonatal onset multisystem inflammatory disease (NOMID)	AD	607115	*CIAS1*
Sterile multifocal osteomyelitis, periostitis, and pustulosis (CINCA/NOMID-like)	AR	147679	*IL1RN*
Chronic recurrent multifocal osteomyelitis with congenital dyserythropoietic anemia (CRMO with CDA; Majeed syndrome)	AR	609628	*LPIN2*
Hyperostosis/hyperphosphatemia syndrome	AR	610233	*GALNT3*
Hyaline fibromatosis syndrome	AR	236490	*ANTXR2*
32. Cleidocranial dysplasia and related disorders			
Cleidocranial dysplasia	AD	119600	*RUNX2*
CDAGS syndrome (craniosynostosis, delayed fontanel closure, parietal foramina, imperforate anus, genital anomalies, skin eruption)	AR	603116	
Yunis–Varon dysplasia	AR	216340	*FIG4*
Parietal foramina (isolated)	AD	168500	*ALX4* *MSX2*

33. Craniosynostosis syndromes			
Pfeiffer syndrome (FGFR1-related)	AD	101600	*FGFR1,*

TABLE I. (*Continued*)

Group/Name of Disorder	**Inheritance**	**MIM No.**	**Locus or Gene**
			FGFR2
Apert syndrome	AD	101200	*FGFR2*
Craniosynostosis with cutis gyrata (Beare–Stevenson)	AD	123790	*FGFR2*
Crouzon syndrome	AD	123500	*FGFR2*
Bent bone dysplasia	AD	614592	*FGFR2*
Crouzon-like craniosynostosis with acanthosis nigricans (Crouzonodermoskeletal syndrome)	AD	612247	*FGFR3*
Craniosynostosis, Muenke type	AD	602849	*FGFR3*

34. Dysostoses with predominant craniofacial involvement			
Mandibulo-facial dysostosis (Treacher Collins, Franceschetti–Klein)	AD, AD, AR	154500	*TCOF1, POLR1D, POLR1C*
Oral-facial-digital syndrome type I (OFD1)	XLR	311200	*CXORF5*
Weyers acrofacial (acrodental) dysostosis	AD	193530	*EVC1 EVC2*
Endocrine-cerebro-osteodysplasia (ECO)	AR	612651	*ICK*
Craniofrontonasal syndrome	XLD	304110	*EFNB1*
Frontonasal dysplasia, type 1	AR	136760	*ALX3*

35. Dysostoses with predominant vertebral with and without costal involvement			
Currarino triad	AD	176450	*HLXB9*
Spondylocostal dysostosis type 1 (SCD01), type 2 (SCD02), type 3(SCD03), type 4 (SCD04),	AR	277300	*DLL3*
		608681	*MESP2*
		609813	*LFNG*
		613686	*HES7*
type 5 (SCD05)	AD	122600	*TBX6*
Spondylothoracic Dyostosis (STD)	AR		*MESP2*
Vertebral segmentation defect (congenital scoliosis) with variable penetrance	AD		*MESP2*
			HES7
Klippel–Feil anomaly with laryngeal malformation	AD	148900	*GDF6*
		613702	*GDF3*
	AR	214300	*MEOX1*
Cerebro-costo-mandibular syndrome (rib gap syndrome)	AD	117650	*SNRPB*
Cerebro-costo-mandibular-like syndrome with vertebral defects	AR	611209	*COG1*
Diaphanospondylodysostosis	AR	608022	*BMPER*
Spondylo-megaepiphyseal-metaphyseal dysplasia (SMMD)	AR	613330	*NKX3-2*
See also Spondylocarpotarsal dysplasia in group 7			
36. Patellar dysostoses			
Ischiopatellar dysplasia (small patella syndrome)	AD	147891	*TBX4*
Nail-patella syndrome	AD	161200	*LMX1B*
Genitopatellar syndrome	AR?	606170	*KAT6B*
Ear-patella-short stature syndrome (Meier–Gorlin)	AR	224690	*ORC1*

37. Brachydactylies (without extraskeletal manifestations)			
Brachydactyly type A1	AD	112500	*IHH*
Brachydactyly type A1	AD		
Brachydactyly type A2	AD	112600	*BMPR1B*
Brachydactyly type A2	AD	112600	*BMP2*
Brachydactyly type A2	AD	112600	*GDF5*
Brachydactyly type B	AD	113000	*ROR2*
Brachydactyly type B2	AD	611377	*NOG*
Brachydactyly type C	AD, AR	113100	*GDF5*
Brachydactyly type D	AD	113200	*HOXD13*
Brachydactyly type E	AD	113300	*PTHLH*
Brachydactyly type E	AD	113300	*HOXD13*
Brachydactyly with anonychia (Cooks syndrome)	AD	106995	*SOX9*
38. Brachydactylies (with extraskeletal manifestations)			
Brachydactyly-mental retardation syndrome	AD	600430	*HDAC4*
Hyperphosphatasia with mental retardation, brachytelephalangy, and distinct face	AR		*PIGV*
Brachydactyly-hypertension syndrome (Bilginturan)	AD	112410	*PDE3A*
Microcephaly-oculo-digito-esophageal-duodenal syndrome (Feingold syndrome)	AD	164280	*MYCN*
Hand-foot-genital syndrome	AD	140000	*HOXA13*
Rubinstein–Taubi syndrome	AD	180849	*CREBBP*

39. Limb hypoplasia–reduction defects group			
Ulnar-mammary syndrome	AD	181450	*TBX3*
de Lange syndrome	AD	122470	*NIPBL*
	XL	300590	*SMC1A*
	AD	619759	*SMC3*
	AD	614701	*RAD21*
	XL	300882	*HDAC8*
Fanconi anemia *(see note below)*	AR	227650	*(several)*
Thrombocytopenia-absent radius (TAR)	AR	274000	*RBM8A*
Thrombocythemia with distal limb defects	AD		*THPO*
Holt-Oram syndrome	AD	142900	*TBX5*
Okihiro syndrome (Duane–radial ray anomaly)	AD	607323	*SALL4*
Cousin syndrome	AR	260660	*TBX15*
Roberts syndrome	AR	268300	*ESCO2*
Split-hand-foot malformation with long bone deficiency (SHFLD3)	AD	612576	*BHLHA9*
Tibial hemimelia	?	275220	
Tibial hemimelia-polysyndactyly-triphalangeal thumb	AD	188740	*SHH-ZRS*
Acheiropodia	AR	200500	*LMBR1*

40. Ectrodactyly with and without other manifestations			
Ankyloblepharon-ectodermal dysplasia-cleft lip/palate (AEC)	AD	106260	*P63 (TP63)*
Ectrodactyly-ectodermal dysplasia cleft-palate syndrome Type 3 (EEC3)	AD	604292	*P63 (TP63)*
Ectrodactyly-ectodermal dysplasia cleft-palate syndrome type 1 (EEC1)	AD	129900	
Ectrodactyly-ectodermal dysplasia-macular dystrophy syndrome (EEM)	AR	225280	*CDH3*
Limb-mammary syndrome (including ADULT syndrome)	AD	603273	*P63 (TP63)*
Split hand-foot malformation, isolated form, type 4 (SHFM4)	AD	605289	*P63 (TP63)*
Split hand-foot malformation, isolated form, type 1 (SHFM1)	AD	183600	*DLX5 DLX6*
Split hand-foot malformation, isolated form, type 3 (SHFM3)	AD	246560	10q
Split hand-foot malformation, isolated form, type 5 (SHFM5)	AD AR	606708	*WNT10B*
Hartsfield syndrome	AD	615465	*FGFR1*
41. Polydactyly-Syndactyly-Triphalangism group			
Preaxial polydactyly type 1 (PPD1)	AD	174400	*SHH-ZRS*
Postaxial polydactyly type A	AD	174200	*GLI3*
Postaxial polydactyly type B	Complex		
Triphalangeal thumb (TPT)-polydactyly syndrome	AD	174500	*SHH-ZRS*
Preaxial polydactyly type 3 (PPD3)	AD	174600	
Preaxial polydactyly type 4 (PPD4)	AD	174700	*GLI3*
Greig cephalopolysyndactyly syndrome	AD	175700	*GLI3*
Pallister–Hall syndrome	AD	146510	*GLI3*

42. Defects in joint formation and synostoses			
Multiple synostoses syndrome type 3	AD	612961	*FGF9*
Proximal symphalangism type 1	AD	185800	*NOG*
Proximal symphalangism type 2	AD	185800	*GDF5*
Radio-ulnar synostosis with amegakaryocytic thrombocytopenia	AD	605432	*HOXA11*
Liebenberg syndrome	AD	186550	*PITX1*
Congenital club foot	AD	119800	*PITX1*