HNL Genomics (CTGT)
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TEST REQUISITION FORM WITH TARGETED VARIANT TESTING

Please print clearly and provide all requested information. CTGT cannot initiate testing unless this information is provided.

PATIENT / SPECIMEN INFORMATION

PATIENT NAME – LAST, FIRST, MI	M F	MRN	DATE OF BIRTH (MM-DD-YYYY)
ADDRESS		PHONE	ETHNICITY
CITY, STATE, ZIP		NAME OF LEGAL GUARDIAN IF PATIENT IS A MINOR	
TYPE OF SPECIMEN	DATE OF COLLECTION	DATE AND TIME OF RECEIPT (TO BE COMPLETED BY CTGT)	

REPORTING INFORMATION

REFERRAL SOURCE

REFERRED BY	NPI NUMBER	GENETIC COUNSELOR	
INSTITUTION		PHONE	FAX
ADDRESS		E-MAIL	
CITY, STATE, ZIP		**SIGNATURE (REQUIRED** – By signing, you agree to the Terms on page 10 of this form)	

ADDITIONAL REPORTS

REFERRING LAB	CONTACT PERSON	
ADDRESS	PHONE	FAX
CITY, STATE, ZIP	REFERRING LAB ID#	

PAYMENT INFORMATION

☐ **INSTITUTIONAL BILLING**

FACILITY NAME	CONTACT PERSON	
ADDRESS	PHONE	FAX
CITY, STATE, ZIP	E-MAIL	

☐ **SELF PAY (ALSO REQUIRED FOR ALL INSURANCE CASES – PLEASE SEE "INSURANCE" BELOW.)**

☐ CHECK ☐ M.O.	Please make check or money order payable to **Health Network Laboratories**.	CARD HOLDER NAME	BILLING ZIP CODE
☐ MC ☐ VISA	ACCOUNT NUMBER	EXPIRATION DATE	3 DIGIT SECURITY CODE (on back of card)
The total cost of testing is $_____. I agree that CTGT, LLC. shall bill this amount to my credit card.		**SIGNATURE OF CARDHOLDER (REQUIRED)**	

☐ **INSURANCE -** In addition to completing the information below, be sure to provide a clear copy of both the front and back of your insurance card, and sign below. In order to provide all necessary information to the insurance company for both benefit investigation and prior authorization the Prior Authorization Information packet must be completed and submitted to HNL Genomics (CTGT) via fax 484-244-2904 or emailed to HNLPriorAuth@hnl.com. This form can be found on our website at

NAME OF INSURED		RELATIONSHIP TO PATIENT
INSURANCE ID NUMBER		GROUP NUMBER
PRE-AUTHORIZATION NUMBER	DATE(S) AUTHORIZATION VALID	INSURANCE COMPANY PHONE NUMBER
SIGNATURE OF INSURED (REQUIRED)		**DATE/MM/YY:**

ICD-10 CODES (REQUIRED):

CLINICAL DIAGNOSIS:	AGE AT INITIAL PRESENTATION:

PLEASE CONTACT OUR OFFICE PRIOR TO SENDING SPECIMEN IF THERE ARE ANY QUESTIONS.

LABORATORY TEST REQUISITION, PAGE 2

NGS - DEL/DUP - NGS & DEL/DUP FOR ANY SINGLE GENE IN A NGS PANEL

☐ 5184 *NGS*	☐ 2044 *Del / Dup*	☐ 5287 *NGS & Del / Dup*	Gene:

NEXT GENERATION SEQUENCING PANELS

NGS panel	Genes	Seq	Del/Dup	Seq & Del/Dup
Abnormal mineralization disorders	*ALPL, ANKH, CASR, CLCN5, CYP27B1, DMP1, ENPP1, FAH, FGF23, OCRL, PHEX, SLC34A1, SLC34A3, SLC9A3R1, VDR*	☐ 5082	☐ 5083	☐ 5084
Achondrogenesis	*COL2A1, SLC26A2, TRIP11*	☐ 5139	☐ 5140	☐ 5141
Adams-Oliver syndrome	*ARHGAP31, DLL4, DOCK6, EOGT, NOTCH1, RBPJ*	☐ 1933	☐ 1934	☐ 1935
Alagille syndrome	*ATP8B1, JAG1, NOTCH2*	☐ 5157	☐ 5158	☐ 5159
Alport syndrome	*COL4A3, COL4A4, COL4A5, COL4A6*	☐ 5142	☐ 5143	☐ 5144
Amelogenesis imperfecta and related disorders	*AMELX, C4ORF26, DLX3, DSPP, ENAM, FAM20A, FAM83H, GPR68, ITGB6, KLK4, LAMA3, LAMB3, MMP20, SLC24A4, SMOC2, WDR72*	☐ 5197	☐ 5198	☐ 5199
Amyotrophic lateral sclerosis and related disorders	*ALS2, ANG, ARHGEF28, CHCHD10, CHMP2B, ERBB4, FIG4, FUS, HNRNPA1, HNRNPA2B1, MATR3, OPTN, PFN1, SETX, SIGMAR1, SOD1, SPG11, SQSTM1, TARDBP, TBK1, TUBA4A, UBQLN2, VAPB, VCP*	☐ 5235	☐ 5236	☐ 5237
Arterial calcification, generalized, of infancy	*ABCC6, ENPP1*	☐ 1438	☐ 1478	☐ 1479
Atrial fibrillation	*ABCC9, GATA6, GJA5, GLA, HCN4, KCNA5, KCND3, KCNE1, KCNE2, KCNH2, KCNJ2, KCNQ1, LMNA, MYL4, NPPA, NUP155, SCN1B, SCN2B, SCN3B, SCN4B, SCN5A, TBX5*	☐ 5309	☐ 5310	☐ 5311
Atrioventricular block	*DES, EMD, GAA, GLA, LMNA, NKX2-5, SCN1B, SCN5A, TRPM4*	☐ 5312	☐ 5313	☐ 5314
Atypical hemolytic uremic syndrome susceptibility and related disorders	*ADAMTS13, C3, CD46, CFB, CFH, CFHR1, CFHR2, CFHR3, CFHR5, CFI, DGKE, MMACHC,THBD*	☐ 5145	☐ 5146	☐ 5147
Auriculocondylar syndrome	*EDN1, GNAI3, PLCB4*	☐ 5352	☐ 5353	☐ 5354
Axial spondylometaphyseal dysplasia	*C21orf2, NEK1*	☐ 5254	☐ 5255	☐ 5256
Bartter syndrome	*BSND, CASR, CLCNKA, CLCNKB, GNA11, KCNJ1, MAGED2, SLC12A1, SLC12A3*	☐ 5391	☐ 5392	☐ 5393
Basal cell nevus syndrome	*PTCH1, PTCH2, SUFU*	☐ 2220	☐ 2221	☐ 2222
Bethlem myopathy & Ullrich congenital muscular dystrophy	*COL6A1, COL6A2, COL6A3, COL12A1*	☐ 1059	☐ 1486	☐ 1487
Blepharocheilodontic syndrome	*CDH1, CTNND1*	☐ 5346	☐ 5347	☐ 5348
Brittle cornea syndrome	*PRDM5, ZNF469*	☐ 5247	☐ 5248	☐ 5249
Brugada syndrome and related disorders	*ABCC9, ANK2, CACNA1C, CACNA2D1, CACNB2, CAV3, FGF12, GAA, GLA, GPD1L, HCN4, KCND2, KCND3, KCNE3, KCNE5, KCNH2, KCNJ8, PKP2, RANGRF, SCN1B, SCN2B, SCN3B, SCN5A, SCN10A, SEMA3A, SLMAP, TRPM4*	☐ 5315	☐ 5316	☐ 5317
CADASIL	*HTRA1, NOTCH3*	☐ 5415	☐ 5416	☐ 5417
Cantu syndrome	*ABCC9, KCNJ8*	☐ 5160	☐ 5161	☐ 5162
Cardiac channelopathy	*ABCC9, AKAP9, ANK2, CACNA1C, CACNA2D1, CACNB2, CALM1, CALM2, CALM3, CASQ2, CAV3, FGF12, GPD1L, HCN4, KCND2, KCND3, KCNE1, KCNE2, KCNE3, KCNE5, KCNH2, KCNJ2, KCNJ5, KCNJ8, KCNQ1, PKP2, NOS1AP, RANGRF, RYR2, SCN1B, SCN2B, SCN3B, SCN4B, SCN5A, SCN10A, SEMA3A, SLMAP, SNTA1, TECRL, TRDN, TRPM4*	☐ 5318	☐ 5319	☐ 5320
Catecholaminergic polymorphic ventricular tachycardia	*ANK2, CALM1, CALM2, CALM3, CASQ2, KCNJ2, RYR2, TECRL, TRDN*	☐ 5321	☐ 5322	☐ 5323
Cerebral cavernous malformations	*KRIT1, CCM2, PDCD10*	☐ 1502	☐ 1503	☐ 1504
Cerebral small vessel disease	*COL4A1, COL4A2, CTC1, GLA, HTRA1, NOTCH3, TREX1*	☐ 5418	☐ 5419	☐ 5420
Cerebrooculofacioskeletal syndrome	*ERCC1, ERCC2, ERCC5, ERCC6*	☐ 5364	☐ 5365	☐ 5366
Charcot Marie Tooth disease	*AARS, AIFM1, BSCL2, C12ORF65, COX6A1, DHTKD1, DNM2, DYNC1H1, EGR2, FGD4, FIG4, GARS, GDAP1, GJB1, GNB4, HADHB, HSPB1, HSPB8, KARS, KIF1B, LITAF, LMNA, LRSAM1, MED25, MFN2, MPZ, MTMR2, NDRG1, NEFL, PDK3, PLEKHG5, PMP22, PRPS1, PRX, RAB7A, SBF1, SBF2, SH3TC2, TFG, TRIM2, TRPV4, YARS*	☐ 5130	☐ 5131	☐ 5132
Cholestasis	*ATP8B1, ABCB11, ABCB4, TJP2*	☐ 2262	☐ 2263	☐ 2264
Chondrodysplasia punctata and related disorders	*AGPS, ARSL, EBP, FAR1, GNPAT, LBR, MGP, NSDHL, PEX5, PEX7*	☐ 2024	☐ 2025	☐ 2026
Cleft lip, cleft palate and related disorders	*BMP4, COL2A1, COL11A1, COL11A2, COL9A1, COL9A2, COL9A3, FOXE1, GRHL3, IRF6, MSX1, SUMO1, NECTIN1, SATB2, TBX22, TGDS, TP63*	☐ 5291	☐ 5292	☐ 5293
Cockayne syndrome	*ERCC6, ERCC8*	☐ 5367	☐ 5368	☐ 5369
Cole-Carpenter syndrome	*P4HB, SEC24D*	☐ 5188	☐ 5189	☐ 5190
Congenital contracture syndrome extended	*ADCY6, ADGRG6, CHRNA1, CHRND, CHRNG, CNTNAP1, DNM2, DOK7, ECEL1, ERBB3, FBN2, GLDN, GLE1, LGI4, LMNA, MUSK, MYBPC1, MYH3, MYH8, NALCN, NEK9, PIEZO2, PIP5K1C, RAPSN, TNNI2, TNNT3, TPM2, VIPAS39, VPS33B, ZBTB42, ZMPSTE24*	☐ 5294	☐ 5295	☐ 5296
Congenital heart disease	*CHD7, ELN, GATA4, GATA6, GDF1, JAG1, NKX2-5, NKX2-6, NOTCH1, NOTCH2, NR2F2, TAB2, TBX1, TBX5, TBX20, ZIC3*	☐ 5148	☐ 5149	☐ 5150
Connective tissue disorder NGS panel	*ACTA2, ADAMTS2, AEBP1, ATP7A, B3GALT6, B4GALT7, BGN, C1R, C1S, CBS, CHST14, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, COL12A1, DCHS1, DSE, FBN1, FBN2, FKBP14, FLCN, FLNA, FOXE3, LOX, LTBP3, MAT2A, MFAP5, MED12, MYH11, MYLK, NOTCH1, PRKG1, PLOD1, PRDM5, SKI, SLC2A10, SLC39A13, SMAD2, SMAD3, SMAD4, TGFB2, TGFB3, TGFBR1, TGFBR2, ZNF469*	☐ 5433	☐ 5434	☐ 5435
Cornelia de Lange syndrome and related disorders	*AFF4, ANKRD11, HDAC8, KMT2A, NIPBL, RAD21, SMC1A, SMC3*	☐ 5181	☐ 5182	☐ 5183
Craniosynostosis core	*FGFR1, FGFR2, FGFR3, TCF12, TWIST1*	☐ 5194	☐ 5195	☐ 5196
Craniosynostosis	*CDC45, CYP26B1, EFNB1, ERF, FGFR1, FGFR2, FGFR3, FREM1, GLI3, IFT43, IFT122, IL11RA, MEGF8, MSX2, POR, RAB23, RECQL4, SKI, SLC25A24, TCF12, TGFBR1, TGFBR2, TWIST1, WDR19, WDR35, ZIC1*	☐ 5085	☐ 5086	☐ 5087
Cutis laxa	*ALDH18A1, ATP6V0A2, ATP6V1A, ATP6V1E1, EFEMP2, ELN, FBLN5, LTBP4, PYCR1*	☐ 1712	☐ 1713	☐ 1714

NGS panel	Genes	Seq	Del/Dup	Seq & Del/Dup
Dense bone dysplasia	*ANKH, COL1A1, DLX3, GJA1, HPGD, LRP4, MTAP, PTDSS1, SLCO2A1, SOST, TBXAS1, TGFB1, TNFRSF11B, TYROBP*	☐ 5088	☐ 5089	☐ 5090
Desbuquois dysplasia core	*CANT1, CSGALNACT1, IMPAD1, XYLT1*	☐ 1854	☐ 1855	☐ 1856
Desbuquois dysplasia and related disorders	*B3GALT6, B3GAT3, CANT1, CHST3, CSGALNACT1, FLNB, GZF1, IMPAD1, KIF22, SLC26A2, XYLT1*	☐ 5124	☐ 5125	☐ 5126
Diamond-Blackfan anemia	*GATA1, RPL11, RPL15, RPL26, RPL27, RPL31, RPL35A, RPL5, RPS10, RPS17, RPS19, RPS24, RPS26, RPS27, RPS28, RPS29, RPS7, TSR2*	☐ 5288	☐ 5289	☐ 5290
Distal arthrogryposes	*ECEL1, FBN2, MYBPC1, MYH3, MYH8, NALCN, PIEZO2, TNNI2, TNNT3, TPM2*	☐ 5133	☐ 5134	☐ 5135
Distal hereditary motor neuropathy and related disorders	*ATP7A, BICD2, BSCL2, DCAF8, DCTN1, DNAJB2, DYNC1H1, FBXO38, GAN, GARS, GJB1, HARS, HINT1, HSPB1, HSPB3, HSPB8, IGHMBP2, PDK3, PLEKHG5, REEP1, SETX, SIGMAR1, SLC5A7, TRPV4*	☐ 5200	☐ 5201	☐ 5202
Distal myopathy	*ANO5, BAG3, CAV3, CRYAB, DES, DNAJB6, DNM2, DYSF, FHL1, FLNC, GNE, LDB3, MATR3, MYH7, MYOT, SQSTM1, TCAP, TIA1, TTN, VCP*	☐ 5238	☐ 5239	☐ 5240
Dyggve-Melchior-Clausen disease	*DYM, RAB33B*	☐ 5297	☐ 5298	☐ 5299
Dyskeratosis congenita	*ACD, CTC1, DKC1, NHP2, NOP10, PARN, RTEL1, TERC, TERT, TINF2, USB1, WRAP53*	☐ 5203	☐ 5204	☐ 5205
Ectodermal dysplasia	*EDA, EDAR, EDARADD, GJB6, HOXC13, KDF1, KREMEN1, KRT74, KRT85, MSX1*	☐ 5206	☐ 5207	☐ 5208
Ectopia lentis	*ADAMTSL4, FBN1*	☐ 5163	☐ 5164	☐ 5165
Ehlers-Danlos syndrome - Dominant	*C1R, C1S, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, COL12A1, FLNA*	☐ 5064	☐ 5065	☐ 5066
Ehlers-Danlos syndrome - Dominant & Recessive	*ADAMTS2, AEBP1, ATP7A, B3GALT6, B4GALT7, C1R, C1S, CHST14, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, COL12A1, DSE, FKBP14, FLNA, PLOD1, PRDM5, SLC39A13, ZNF469*	☐ 5067	☐ 5068	☐ 5069
Ehlers-Danlos syndrome - Recessive	*ADAMTS2, AEBP1, ATP7A, B3GALT6, B4GALT7, CHST14, COL12A1, DSE, FKBP14, PLOD1, PRDM5, SLC39A13, ZNF469*	☐ 5070	☐ 5071	☐ 5072
Ehlers-Danlos syndrome, arthrochalasia type NGS panel	*COL1A1, COL1A2*	☐ 1138	☐ 1139	☐ 1538
Ehlers-Danlos syndrome, classic type	*COL5A1, COL5A2*	☐ 1134	☐ 1535	☐ 1536
Ehlers-Danlos syndrome core	*COL3A1, COL5A1, COL5A2*	☐ 5209	☐ 5210	☐ 5211
Ehlers-Danlos syndrome, periodontal type	*C1R, C1S*	☐ 5250	☐ 5251	☐ 5252
Ehlers-Danlos syndrome, spondylodysplastic type	*B3GALT6, B4GALT7, SLC39A13*	☐ 2035	☐ 2036	☐ 2037
Ellis-van Creveld syndrome & Weyers acrofacial dysostosis	*EVC, EVC2*	☐ 1140	☐ 1141	☐ 1539
Epidermolysis bullosa	*CAST, CDSN, CHST8, COL17A1, COL7A1, CSTA, DSP, DST, EXPH5, FERMT1, ITGA3, ITGA6, ITGB4, JUP, KLHL24, KRT5, KRT14, LAMA3, LAMB3, LAMC2, PKP1, PLEC, SERPINB8, TGM5*	☐ 5073	☐ 5074	☐ 5075
Erythrokeratodermias and related disorders	*DSP, GJA1, GJB3, GJB4, KDSR, KRT83, LOR*	☐ 5373	☐ 5374	☐ 5375
Exudative vitreoretinopathy	*CAPN5, FZD4, KIF11, LRP5, NDP, TSPAN12, ZNF408*	☐ 1399	☐ 1548	☐ 1549
Familial hypercholesterolemia	*APOB, LDLR, LDLRAP1, PCSK9*			☐ 7000
Fibrillinopathy	*CBS, FBN1, FBN2*	☐ 5003	☐ 5012	☐ 5013
Fibrochondrogenesis	*COL11A1, COL11A2*	☐ 1694	☐ 1695	☐ 1696
Frontometaphyseal dysplasia	*FLNA, MAP3K7, TAB2*	☐ 5212	☐ 5213	☐ 5214
Frontonasal dysplasia and craniofrontonasal syndrome	*ALX3, ALX4, ALX1, EFNB1, SIX2*	☐ 1918	☐ 1919	☐ 1920
Glomuvenous malformations / Cutaneomucosal venous malformations	*GLMN, TEK*	☐ 1736	☐ 1737	☐ 1738
Hereditary hemorrhagic telangiectasia	*ACVRL1, ENG, GDF2, RASA1, SMAD4*	☐ 1721	☐ 1722	☐ 1723
Hyperphosphatemic familial tumoral calcinosis	*FGF23, GALNT3, KL*	☐ 2369	☐ 2370	☐ 2371
Hyperuricemic nephropathy, familial juvenile	*REN, SEC61A1, UMOD*	☐ 5400	☐ 5401	☐ 5402
Hypotrichosis	*APCDD1, CDSN, DSG4, HR, KRT25, KRT71, KRT74, LIPH, LPAR6, RPL21, SNRPE*	☐ 5306	☐ 5307	☐ 5308
Ichthyosis	*ABCA12, ALOX12B, ALOXE3, CASP14, CAST, CDSN, CERS3, CHST8, CSTA, CYP4F22, FLG, FLG2, GJA1, GJB3, GJB4, KDSR, KRT1, KRT10, KRT2, KRT83, LIPN, LOR, MBTPS2, NIPAL4, PNPLA1, POMP, SERPINB8, ST14, STS, SULT2B1, TGM1, TGM5*	☐ 5091	☐ 5092	☐ 5093
Insulin-like growth factor I deficiency and Insulin-like growth factor I, resistance to	*IGF1, IGF1R*	☐ 2093	☐ 2094	☐ 2095
Joubert syndrome and related disorders	*AHI1, ARL13B, ARMC9, B9D1, B9D2, C2CD3, C5orf42, CC2D2A, CEP104, CEP120, CEP290, CEP41, CSPP1, INPP5E, KIAA0556, KIAA0586, KIF14, KIF7, MKS1, NPHP1, NPHP3, OFD1, PDE6D, PIBF1, RPGRIP1L, SUFU, TMEM107, CTN1, TCTN2, TCTN3, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TTC21B, ZNF423*	☐ 5136	☐ 5137	☐ 5138
Kabuki syndrome	*HNRNPK, KDM6A, KMT2D, RAP1A, RAP1B*	☐ 5427	☐ 5428	☐ 5429
Kenny-Caffey syndrome	*FAM111A, TBCE*	☐ 1906	☐ 1907	☐ 1908
Keratoconus and related disorders	*MIR184, PRDM5, VSX1, ZNF469*	☐ 5421	☐ 5422	☐ 5423
Klippel-Feil syndrome	*GDF3, GDF6, MEOX1, MYO18B, RIPPLY2*	☐ 1963	☐ 1964	☐ 1965
Leber congenital amaurosis and related disorders	*AIPL1, ALMS1, CABP4, CEP290, CLUAP1, CNGA3, CRB1, CRX, DTHD1, GDF6, GUCY2D, IFT140, IMPDH1, INPP5E, IQCB1, KCNJ13, LCA5, LRAT, MERTK, MYO7A, NMNAT1, OTX2, PRPH2, RD3, RDH5, RDH12, ROM1, RPE65, RPGRIP1, SNRNP200, SPATA7, TULP1*	☐ 5268	☐ 5269	☐ 5270
Lethal congenital contractual syndrome and related disorders	*ADCY6, ADGRG6, CHRNA1, CHRND, CHRNG, CNTNAP1, DNM2, DOK7, ERBB3, GLDN, GLE1, LGI4, LMNA, MUSK, MYBPC1, NEK9, PIP5K1C, RAPSN, VIPAS39, VPS33B, ZBTB42, ZMPSTE24*	☐ 5300	☐ 5301	☐ 5302
Limb girdle muscular dystrophy	*ANO5, CAPN3, CAV3, DAG1, DES, DNAJB6, DYSF, EMD, FHL1, FKRP, FKTN, FLNC, GAA, GMPPB, HNRNPDL, ISPD, LARGE1, LIMS2, LMNA, MYOT, PLEC, POMGNT1, POMK, POMT1, POMT2, SGCA, SGCB, SGCD, SGCG, TCAP, TNPO3, TOR1AIP1, TRAPPC11, TRIM32, TTN*	☐ 5215	☐ 5216	☐ 5217
Lissencephaly core	*ARX, CDK5, DCX, KATNB1, LAMB1, NDE1, PAFAH1B1, RELN, TMTC3,TUBA1A*	☐ 5403	☐ 5404	☐ 5405
Lissencephaly and related disorders	*ACTB, ACTG1, ADGRG1, ARX, ATP6V0A2, B3GALNT2, B4GAT1, CDK5, DAG1, DCX, DYNC1H1, FKRP, FKTN, GMPPB, ISPD, KATNB1, KIF2A, KIF5C, LAMA2, LAMB1, LARGE1, NDE1, PAFAH1B1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, RELN, RXYLT1, SNAP29, SRD5A3, TMTC3, TUBA1A, TUBA8, TUBB, TUBB2A, TUBB2B, TUBB3, TUBG1, VLDLR, WDR62*	☐ 5406	☐ 5407	☐ 5408
Loeys-Dietz syndrome core	*TGFBR1, TGFBR2*	☐ 1181	☐ 1571	☐ 1572

NGS panel	Genes	Seq	Del/Dup	Seq & Del/Dup
Loeys-Dietz syndrome	*SMAD3, TGFBR1, TGFBR2, TGFB2, TGFB3*	☐ 2208	☐ 2209	☐ 2210
Long QT syndrome	*AKAP9, ANK2, CACNA1C, CALM1, CALM2, CALM3, CAV3, KCNE1, KCNE2, KCNH2, KCNJ2, KCNJ5, KCNQ1, NOS1AP, RYR2, SCN4B, SCN5A, SNTA1, TRDN*	☐ 5324	☐ 5325	☐ 5326
Marfan syndrome, type I / II	*FBN1, TGFBR2*	☐ 1191	☐ 1579	☐ 1580
Marfan syndrome and Loeys-Dietz syndrome core	*FBN1, TGFBR1, TGFBR2*	☐ 5260	☐ 5261	☐ 5262
Marfan syndrome and Loeys-Dietz syndrome	*FBN1, SMAD3, TGFBR1, TGFBR2, TGFB2, TGFB3*	☐ 1192	☐ 1581	☐ 1582
Marfan Syndrome, Loeys-Dietz Syndrome, Familial Thoracic Aortic Aneurysms & Dissections, and Related Disorders	*ACTA2, BGN, CBS, COL3A1, COL5A1, COL5A2, FBN1, FBN2, FLNA, FOXE3, LOX, LTBP3, MAT2A, MED12, MFAP5, MYH11, MYLK, NOTCH1, PRKG1, SKI, SLC2A10, SMAD2, SMAD3, SMAD4, TGFB2, TGFB3, TGFBR1 TGFBR2*	☐ 5076	☐ 5077	☐ 5078
Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome	*AKT3, CCND2, PIK3R2*	☐ 5409	☐ 5410	☐ 5411
Meier-Gorlin syndrome	*CDC45, CDC6 , CDT1, GMNN, MCM5, ORC1, ORC4, ORC6*	☐ 1875	☐ 1876	☐ 1877
Metaphyseal anadysplasia	*MMP13, MMP9*	☐ 1202	☐ 1587	☐ 1588
Microcephalic primordial dwarfism	*ATR, ATRIP, CDC45, CDC6, CDT1, CENPJ, CEP63, CEP152, DNA2, DONSON, GMNN, LIG4, NIN, ORC1, ORC4, ORC6, PCNT, RBBP8, RNU4ATAC, TRAIP, XRCC4*	☐ 5166	☐ 5167	☐ 5168
Mucopolysaccharidosis	*ARSB, GALNS, GLB1, GNPTAB, GNPTG, GNS, GUSB, HGSNAT, HYAL1, IDUA, IDS, MCOLN1, NAGLU, SGSH*	☐ 5241	☐ 5242	☐ 5243
Multiple epiphyseal dysplasia (MED)	*CANT1, COL2A1, COL9A1, COL9A2, COL9A3, COMP, MATN3, SLC26A2*	☐ 5094	☐ 5095	☐ 5096
Multiple exostoses	*EXT1, EXT2*	☐ 1146	☐ 1542	☐ 1543
Multiple pterygium syndrome, lethal type	*CHRNA1, CHRND, CHRNG*	☐ 2274	☐ 2275	☐ 2276
Myofibrillar myopathy and related disorders	*ACTA1, BAG3, CRYAB, DES, DNAJB6, FHL1, FLNC, KY, LDB3, MYOT, PYROXD1*	☐ 5218	☐ 5219	☐ 5220
Nephrolithiasis and related disorders	*ADCY10, AGXT, AP2S1, APRT, ATP6V0A4, ATP6V1B1, CA2, CASR, CLCN5, CLDN16, CLDN19, CYP24A1, FAM20A, GNA11, GRHPR, HNF4A, HOGA1, HPRT1, KCNJ1, OCRL, SLC2A9, SLC3A1, SLC4A1, SLC7A9, SLC9A3R1, SLC12A1, SLC22A12, SLC26A1, SLC34A1, SLC34A3, XDH*	☐ 5333	☐ 5334	☐ 5335
Nephronophthisis and related disorders	*AHI1, ANKS6, CC2D2A, CEP164, CEP290, CEP41, CEP83, DCDC2, FAN1, GLIS2, INVS, IQCB1, MAPKBP1, NEK8, NPHP1, NPHP3, NPHP4, OFD1, PKHD1, RPGRIP1L, SDCCAG8, TMEM138, TMEM216, TMEM67, TRAF3IP1, TTC21B, WDR19, XPNPEP3, ZNF423*	☐ 5394	☐ 5395	☐ 5396
Nephrotic syndrome and related disorders	*ACTN4, ANLN, ARHGAP24, ARHGDIA, CD2AP, COL4A3, COL4A4, COL4A5, COL4A6, COQ2, COQ6, COQ8B, CRB2, CUBN, DGKE, EMP2, FN1, INF2, ITGA3, ITGB4, KANK2, MAGI2, MEFV, LAMB2, LMX1B, MYO1E, NPHS1, NPHS2, NUP107, NUP205, NUP93, PAX2, PDSS2, PLCE1, PTPRO, SCARB2, SGPL1, SMARCAL1, TRPC6, TTC21B, WDR73, WT1*	☐ 5221	☐ 5222	☐ 5223
Neu-Laxova syndrome	*PHGDH, PSAT1*	☐ 2171	☐ 2172	☐ 2173
Neurofibromatosis and related disorders	*MLH1, MSH2, MSH6, NF1, NF2, SPRED1*	☐ 5191	☐ 5192	☐ 5193
Noonan spectrum disorders	*A2ML1, ACTB, ACTG1, BRAF, CABIN1, CBL, HRAS, KRAS, LZTR1, MAP2K1, MAP2K2, NF1, NF2, NRAS, NSUN2, PPP1CB, PTPN11, RAF1, RASA2, RIT1, RRAS, SHOC2, SOS1, SOS2, SPRED1*	☐ 5079	☐ 5080	☐ 5081
Noonan syndrome core	*BRAF, CBL, HRAS, KRAS, LZTR1, MAP2K1, MAP2K2, NRAS, PTPN11, RAF1, RIT1, SHOC2, SOS1, SOS2*	☐ 5349	☐ 5350	☐ 5351
Oligodontia - Selective tooth agenesis	*AXIN2, EDA, LRP6, LTBP3, MSX1, PAX9, PTH1R, WNT10A, WNT10B*	☐ 5169	☐ 5170	☐ 5171
Omodysplasia	*FZD2, GPC6*	☐ 1213	☐ 1214	☐ 1595
Opitz GBBB syndrome	*MID1, SPECC1L*	☐ 5355	☐ 5356	☐ 5357
Osteogenesis imperfecta *COL1A1* & *COL1A2*	*COL1A1, COL1A2*	☐ 1216	☐ 1599	☐ 1600
Osteogenesis imperfecta core	*COL1A1, COL1A2, IFITM5*	☐ 5232	☐ 5233	☐ 5234
Osteogenesis imperfecta - Dominant	*ALPL, ANO5, COL1A1, COL1A2, IFITM5, P4HB, PLS3*	☐ 5097	☐ 5098	☐ 5099
Osteogenesis imperfecta - Dominant & Recessive	*ALPL, ANO5, BMP1, COL1A1, COL1A2, CREB3L1, CRTAP, FKBP10, IFITM5, LRP5, MBTPS2, P3H1, P4HB, PLOD2, PLS3, PPIB, SEC24D, SERPINF1, SERPINH1, SP7, SPARC, TAPT1, TMEM38B, WNT1, XYLT2*	☐ 5100	☐ 5101	☐ 5102
Osteogenesis imperfecta - Recessive	*ALPL, BMP1, CREB3L1, CRTAP, FKBP10, LRP5, MBTPS2, P3H1, PLOD2, PLS3, PPIB, SEC24D, SERPINF1, SERPINH1, SP7, SPARC, TAPT1, TMEM38B, WNT1, XYLT2*	☐ 5103	☐ 5104	☐ 5105
Osteopetrosis core	*TCIRG1, CLCN7, OSTM1*	☐ 1253	☐ 1621	☐ 1622
Osteopetrosis	*AMER1, CA2, CLCN7, CTSK, FAM20C, FERMT3, LEMD3, LRP5, OSTM1, PLEKHM1, SNX10, TCIRG1, TNFRSF11A, TNFSF11*	☐ 5106	☐ 5107	☐ 5108
Osteopetrosis & Dense bone dysplasia	*AMER1, ANKH, CA2, CLCN7, COL1A1, CTSK, DLX3, FAM20C, FERMT3, GJA1, HPGD, LEMD3, LRP4, LRP5, MTAP, OSTM1, PLEKHM1, PTDSS1, SLCO2A1, SNX10, SOST, TBXAS1, TCIRG1, TGFB1, TNFRSF11A, TNFRSF11B, TNFSF11, TYROBP*	☐ 5109	☐ 5110	☐ 5111
Overgrowth syndrome	*EED, EZH2, GPC3, NFIX, NSD1, OFD1, PIGA*	☐ 1984	☐ 1985	☐ 1986
Paget disease of bone and related disorders	*HNRNPA1, HNRNPA2B1, SQSTM1, TNFRSF11A, TNFRSF11B, VCP, ZNF687*	☐ 2384	☐ 2385	☐ 2386
Peeling skin syndrome	*CAST, CDSN, CHST8, CSTA, FLG2, SERPINB8, TGM5*	☐ 5376	☐ 5377	☐ 5378
Platelet bleeding disorders	*ACTN1, ANO6, AP3B1, BLOC1S3, BLOC1S6, CD36, DTNBP1, GBA, GFI1B, GP1BA, GP1BB, GP6, GP9, HPS1, HPS3, HPS4, HPS5, HPS6, ITGA2B, ITGB3, LYST, MYH9, NBEAL2, P2RY12, PLAU, PRKACG, RASGRP2, SLFN14, SMPD1, TBXA2R, TBXAS1, VWF, WAS, WIPF1*	☐ 5154	☐ 5155	☐ 5156
Polycystic kidney disease and related disorders	*ALG9, ANKS6, ATP6V0A4, BICC1, GANAB, GLIS3, HNF1B, INVS, LRP5, NOTCH2, NPHP3, OFD1, PKD1, PKD2, PKHD1, SEC61A1, TMEM231, TSC1, TSC2, UMOD, ZNF423*	☐ 5336	☐ 5337	☐ 5338
Polycystic liver disease	*GANAB, LRP5, PKD1, PKD2, PKHD1, PRKCSH, SEC63*	☐ 5339	☐ 5340	☐ 5341
Popliteal pterygium syndrome	*IRF6, RIPK4*	☐ 5412	☐ 5413	☐ 5414
Porencephaly	*COL4A1 COL4A2*	☐ 2189	☐ 2190	☐ 2191
Progeroid syndromes and related disorders	*AGPAT2, ALDH18A1, B3GALT6, B4GALT7, BANF1, BSCL2, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC8, FBN1, LMNA, PDGFRB, POLD1, PYCR1, RECQL4, SLC25A24, WRN, ZMPSTE24*	☐ 5379	☐ 5380	☐ 5381
Progressive familial heart block type I	*SCN5A, TRPM4*	☐ 5327	☐ 5328	☐ 5329
Pseudoxanthoma elasticum	*ABCC6, GGCX*	☐ 2104	☐ 2105	☐ 2106
Pulmonary hypertension	*ACVRL1, BMPR1B, BMPR2, CAV1, EIF2AK4, ENG, FOXF1, GDF2, KCNA5, KCNK3, SMAD4, SMAD9*	☐ 2116	☐ 2117	☐ 2118
Restrictive dermopathy, lethal	*LMNA, ZMPSTE24*	☐ 5382	☐ 5383	☐ 5384
Renal tubular dysgenesis	*ACE, AGT, AGTR1, REN*	☐ 5397	☐ 5398	☐ 5399
Robinow syndrome	*DVL1, DVL3, FZD2, NXN, ROR2, WNT5A*	☐ 2127	☐ 2128	☐ 2129
Rubinstein-Taybi syndrome	*CREBBP, EP300*	☐ 2298	☐ 2299	☐ 2300
Short QT syndrome	*CACNA1C, CACNA2D1, CACNB2, KCNH2, KCNJ2, KCNQ1*	☐ 5330	☐ 5331	☐ 5332
Short stature with endocrinopathy	*BTK, GH1, GHR, GHRHR, GHSR, HESX1, IGF1, IGF1R, LHX3, LHX4, OTX2, POU1F1, PROP1, SOX3*	☐ 5358	☐ 5359	☐ 5360
Skeletal dysplasia ciliopathy	*C21orf2, C2CD3, CEP120, DYNC2H1, DYNCLI1, EVC, EVC2, ICK, IFT43, IFT52, IFT80, IFT81, IFT122, IFT140, IFT172, KIAA0586, KIAA0753, TCTEX1D2, NEK1, TTC21B, WDR19, WDR34, WDR35, WDR60*	☐ 5112	☐ 5113	☐ 5114
Skeletal dysplasia core	*ALPL, COL1A1, COL1A2, COL2A1, FGFR3, INPPL1, NKX3-2, SLC26A2, SOX9, TRIP11*	☐ 5115	☐ 5116	☐ 5117

NGS panel	Genes	Seq	Del/Dup	Seq & Del/Dup
Skeletal dysplasia core & extended	*ALPL, ARSL, COL1A1, COL1A2, COL2A1, COL10A1, COL11A1, COL11A2, DDR2, EBP, FGFR3, FLNB, HSPG2, INPPL1, LBR, LIFR, MMP9, MMP13, NKX3-2, NSDHL, PEX7, PTH1R, RMRP, SBDS, SLC26A2, SLC35D1, SOX9, TRIP11, TRPV4*	☐ 5118	☐ 5119	☐ 5120
Skeletal dysplasia extended	*ARSL, COL10A1, COL11A1, COL11A2, DDR2, EBP, FLNB, HSPG2, LBR, LIFR, MMP9, MMP13, NSDHL, PEX7, PTH1R, RMRP, SBDS, SLC35D1, TRPV4*	☐ 5121	☐ 5122	☐ 5123
Skeletal dysplasia and skeletal ciliopathy NGS panel	*ALPL, ARSL, C21orf2, C2CD3, CEP120, COL1A1, COL1A2, COL2A1, COL10A1, COL11A1, COL11A2, DDR2, DYNC2H1, DYNC2LI1, EBP, EVC, EVc2, FGFR3, FLNB, HSPG2, ICK, IFT43, IFT52, IFT80, IFT81, IFT122, IFT140, IFT172, INPPL1, KIAA0586, KIAA0753, LBR, LIFR, MMP9, MMP13, NEK1, NKX3-2, NSDHL, PEX7, PTH1R, RMRP, SBDS, SLC26A2, SLC35D1, SOX9, TCTEX1D2, TRIP11, TRPV4, TTC21B, WDR19, WDR34, WDR35, WDR60*	☐ 5430	☐ 5431	☐ 5432
Smith-McCort dysplasia	*DYM, RAB33B*	☐ 5257	☐ 5258	☐ 5259
SOTOS syndrome and related disorders	*APC2, EZH2, NFIX, NSD1*	☐ 5424	☐ 5425	☐ 5426
Spondylocostal dysostosis	*DLL3, HES7, LFNG, MESP2, RIPPLY2, TBX6*	☐ 1781	☐ 1782	☐ 1783
Spondylo-Epi-Metaphyseal dysplasias	*ACAN, ACP5, B3GALT6, B3GAT3, BGN, C21orf2, CANT1, CHST3, COL2A1, COL9A1, COL9A2, COL9A3, COL10A1, COL11A1, COL11A2, COMP, DYM, DDR2, EIF2AK3, FGFR3, FLNB, FN1, GPX4, HSPG2, IDUA, IHH, IMPAD1, INPPL1, KIF22, LIFR, LONP1, MATN3, MMP9, MMP13, NANS, NEK1, NKX3-2, NPR2, PAM16, PAPSS2, PCYT1A, PTH1R, RAB33B, RMRP, RNU4ATAC, RSPRY1, RUNX2, SBDS, SLC26A2, SMARCAL1, TRAPPC2, TRPV4, WISP3, XYLT1*	☐ 5185	☐ 5186	☐ 5187
Spondyloepimetaphyseal dysplasia with joint laxity	*B3GALT6, KIF22*	☐ 2069	☐ 2070	☐ 2071
Stickler syndrome	*COL2A1, COL9A1, COL9A2, COL9A3, COL11A1, COL11A2, LOXL3, VCAN*	☐ 5127	☐ 5128	☐ 5129
Stickler syndrome - Recessive	*COL9A1, COL9A2, COL9A3*	☐ 1330	☐ 1660	☐ 1661
Stickler syndrome core	*COL2A1, COL11A1, COL11A2*	☐ 1337	☐ 1665	☐ 1666
Three M syndrome	*CCDC8, CUL7, OBSL1*	☐ 5172	☐ 5173	☐ 5174
Thrombocytopenia	*ADAMTS13, ANKRD26, AP3B1, CYCS, ETV6, FLI1, FYB, GATA1, GBA, HOXA11, ITGA2B, ITGB3, LYST, MASTL, MECOM, MPL, MYH9, NBEAL2, PRKACG, RBM8A, RUNX1, SLFN14, SMPD1, SRC, TBXAS1, TUBB1, WAS, WIPF1*	☐ 5224	☐ 5225	☐ 5226
Treacher Collins syndrome core	*POLR1C, POLR1D, TCOF1*	☐ 5303	☐ 5304	☐ 5305
Treacher Collins syndrome and related disorders	*DHODH, EDNRA, EFTUD2, POLR1A, POLR1C, POLR1D, SF3B4, TCOF1, TXNL4A*	☐ 5175	☐ 5176	☐ 5177
Trichothiodystrophy	*ERCC2, ERCC3, GTF2E2, GTF2H5, MPLKIP, RNF113A*	☐ 5385	☐ 5386	☐ 5387
Tuberous sclerosis	*TSC1, TSC2*	☐ 5342	☐ 5343	☐ 5344
Van der Woude syndrome	*GRHL3, IRF6*	☐ 2313	☐ 2314	☐ 2315
Vascular malformations	*ACVRL1, BMPR2, CAV1, CCM2, EIF2AK4, ELMO2, ENG, FOXF1, GATA2, GDF2, GLMN, KCNK3, KRIT1, PDCD10, PTEN, RASA1, SMAD4, SMAD9, TEK*	☐ 5178	☐ 5179	☐ 5180
Vitreoretinopathy	*BEST1, CAPN5, FZD4, COL2A1, COL9A1, COL9A2, COL9A3, COL11A1, COL11A2, COL18A1, KCNJ13, KIF11, LRP5, NDP, NR2E3, RS1, TSPAN12, VCAN, ZNF408*	☐ 5227	☐ 5228	☐ 5229
Waardenburg syndrome	*EDN3, EDNRB, MITF, PAX3, SNAI2, SOX10, TYR*	☐ 5244	☐ 5245	☐ 5246
Weaver and Cohen-Gibson syndrome	*EED, EZH2*	☐ 5265	☐ 5266	☐ 5267
Weill-Marchesani syndrome	*ADAMTS10, ADAMTS17, FBN1, LTBP2*	☐ 1730	☐ 1731	☐ 1732
Xeroderma pigmentosum	*DDB2, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, POLH, XPA, XPC*	☐ 5388	☐ 5389	☐ 5390
Zimmermann-Laband syndrome	*KCNH1, ATP6V1B2*	☐ 2204	☐ 2205	☐ 2206

SINGLE GENE AND SANGER PANEL TESTS

Disorder	Genes	Seq	Del/ Dup	Seq & Del/Dup
Ablepharon-macrostomia syndrome	*TWIST2*	☐ 2138	☐ 2139	☐ 2140
Achondrogenesis, type IA	*TRIP11*	☐ 1001	☐ 1002	☐ 1444
Achondrogenesis, type IB	*SLC26A2*	☐ 1003	☐ 1004	☐ 1445
Achondrogenesis, type II / Hypochondrogenesis	*COL2A1*	☐ 1005	☐ 1006	☐ 1446
Achondroplasia / Hypochondroplasia	*FGFR3*	☐ 1007	☐ 1756	☐ 1757
	FGFR3 Ex 3,5, 7,9,10,13,15	☐ 1008		
Acrofacial dysostosis 1, Nager type	*SF3B4*	☐ 1988	☐ 1989	☐ 1990
Acromelic frontonasal dysostosis	*ZSWIM6*	☐ 2006	☐ 2007	☐ 2008
Acromesomelic dysplasia, Hunter-Thompson type	*GDF5*	☐ 1842	☐ 1843	☐ 1844
Acromesomelic dysplasia, Maroteaux type	*NPR2*	☐ 1010	☐ 1011	☐ 1447
Acromicric dysplasia	*FBN1* Ex 41,42	☐ 1012		
Adams-Oliver syndrome 1	*ARHGAP31*	☐ 1013	☐ 1014	☐ 1448
Adams-Oliver syndrome 2	*DOCK6*	☐ 1449	☐ 1450	☐ 1451
Adams-Oliver syndrome 3	*RBPJ*	☐ 1452	☐ 1453	☐ 1454
Adams-Oliver syndrome 4	*EOGT*	☐ 1455	☐ 1456	☐ 1457
Adams-Oliver syndrome 5	*NOTCH1*	☐ 1936	☐ 1937	☐ 1938
Adams-Oliver syndrome 6	*DLL4*	☐ 2352	☐ 2353	☐ 2354
Alagille syndrome 1	*JAG1*	☐ 1015	☐ 1016	☐ 1461
Alagille syndrome 2	*NOTCH2*	☐ 1390	☐ 1391	☐ 1462
Alagille syndrome, ATP8B1 related	*ATP8B1*	☐ 2141	☐ 2142	☐ 2143
Alopecia universalis congenita & Atrichia with papular lesions	*HR*	☐ 2451	☐ 2452	☐ 2453
Alport syndrome, X-linked	*COL4A5*	☐ 1017	☐ 1018	☐ 1463
Amelogenesis imperfecta, type IV	*DLX3*	☐ 1912	☐ 1913	☐ 1914
Anauxetic dysplasia	*RMRP*	☐ 1019	☐ 1893	☐ 1894
Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps	*COL4A1*	☐ 2144	☐ 2145	☐ 2146
Aortic aneurysm, familial thoracic 3	*TGFBR2*	☐ 1020	☐ 1021	☐ 1464
Aortic aneurysm, familial thoracic 4	*MYH11*	☐ 1023	☐ 1024	☐ 1467
Aortic aneurysm, familial thoracic 5	*TGFBR1*	☐ 1025	☐ 1026	☐ 1468
Aortic aneurysm, familial thoracic 6	*ACTA2*	☐ 1029	☐ 1030	☐ 1473
Aortic aneurysm, familial thoracic 7	*MYLK*	☐ 1031	☐ 5273	☐ 5274
Aortic aneurysm, familial thoracic 8	*PRKG1*	☐ 1739	☐ 1740	☐ 1741
Aortic aneurysm, familial thoracic 9	*MFAP5*	☐ 2009	☐ 2010	☐ 2011
Aortic valve disease 1	*NOTCH1*	☐ 1939	☐ 1940	☐ 1941
Arterial calcification, generalized, of infancy, 1	*ENPP1*	☐ 1036	☐ 1037	☐ 1476
Arterial calcification, generalized, of infancy, 2	*ABCC6*	☐ 1436	☐ 1437	☐ 1477
Arterial tortuosity syndrome	*SLC2A10*	☐ 1038	☐ 1039	☐ 1480
Atelosteogenesis, type I / III	*FLNB*	☐ 1048	☐ 2390	☐ 2391
	FLNB Ex 2, 3, 4, 27, 28, 29	☐ 1049		
Atelosteogenesis, type II	*SLC26A2*	☐ 1051	☐ 1052	☐ 1485
Avascular necrosis of femoral head, primary	*COL2A1*	☐ 1053	☐ 1835	☐ 1836
Barber-Say syndrome	*TWIST2*	☐ 2147	☐ 2148	☐ 2149
Basal cell nevus syndrome	*PTCH1*	☐ 2211	☐ 2212	☐ 2213
Basal cell nevus syndrome	*PTCH2*	☐ 2214	☐ 2215	☐ 2216
Basal cell nevus syndrome	*SUFU*	☐ 2217	☐ 2218	☐ 2219
Benign chronic pemphigus	*ATP2C1*	☐ 5361	☐ 5362	☐ 5363

Disorder	Genes	Seq	Del/ Dup	Seq & Del/Dup
Bent bone dysplasia syndrome	*FGFR2*	☐ 1909	☐ 1910	☐ 1911
Birt-Hogg-Dube syndrome	*FLCN*	☐ 2223	☐ 2224	☐ 2225
Boomerang dysplasia	*FLNB*	☐ 1060	☐ 2392	☐ 2393
	FLNB Ex 2, 3, 4, 27, 28, 29	☐ 1061		
Brachyolmia type 3	*TRPV4*	☐ 1063	☐ 1760	☐ 1761
Brain small vessel disease with or without ocular anomalies	*COL4A1*	☐ 2150	☐ 2151	☐ 2152
Branchiooculofacial syndrome	*TFAP2A*	☐ 2078	☐ 2079	☐ 2080
Brittle cornea syndrome 1	*ZNF469*	☐ 1064	☐ 1065	☐ 1488
Brittle cornea syndrome 2	*PRDM5*	☐ 1066	☐ 1067	☐ 1489
Bruck syndrome 2	*PLOD2*	☐ 1068	☐ 1692	☐ 1693
Buschke-Ollendorff syndrome	*LEMD3*	☐ 1069	☐ 1070	☐ 1490
Caffey disease	*COL1A1*	☐ 1071	☐ 2449	☐ 2450
Campomelic dysplasia	*SOX9*	☐ 1072	☐ 1073	☐ 1491
Camptodactyly, tall stature, and hearing loss syndrome	*FGFR3*	☐ 2316	☐ 2317	☐ 2318
Camurati-Engelmann disease	*TGFB1*	☐ 1074	☐ 2437	☐ 2438
Cantu syndrome	*ABCC9*	☐ 1784	☐ 1785	☐ 1786
Cantu syndrome, KCNJ8 related	*KCNJ8*	☐ 2226	☐ 2227	☐ 2228
Capillary malformation arteriovenous malformation	*RASA1*	☐ 1075	☐ 1076	☐ 1492
Cardiac valvular dysplasia, X-linked	*FLNA*	☐ 1077	☐ 5275	☐ 5276
Cartilage-hair hypoplasia	*RMRP*	☐ 1078	☐ 1895	☐ 1896
Catel-Manzke syndrome	*TGDS*	☐ 2012	☐ 2013	☐ 2014
Cerebral arteriopathy, with subcortical infarcts and leukoencephalopathy 1	*NOTCH3*	☐ 1991	☐ 2136	☐ 2137
Cerebral cavernous malformations 1	*KRIT1*	☐ 1493	☐ 1494	☐ 1495
Cerebral cavernous malformations 2	*CCM2*	☐ 1496	☐ 1497	☐ 1498
Cerebral cavernous malformations 3	*PDCD10*	☐ 1499	☐ 1500	☐ 1501
CHARGE syndrome	*CHD7*	☐ 2229	☐ 2230	☐ 2231
Cholestasis, benign recurrent intrahepatic, 1	*ATP8B1*	☐ 2232	☐ 2233	☐ 2234
Cholestasis, benign recurrent intrahepatic, 2	*ABCB11*	☐ 2235	☐ 2236	☐ 2237
Cholestasis, intrahepatic, of pregnancy, 1	*ATP8B1*	☐ 2241	☐ 2242	☐ 2243
Cholestasis, intrahepatic, of pregnancy, 3	*ABCB4*	☐ 2244	☐ 2245	☐ 2246
Cholestasis, progressive familial intrahepatic, 1	*ATP8B1*	☐ 2250	☐ 2251	☐ 2252
Cholestasis, progressive familial intrahepatic, 2	*ABCB11*	☐ 2253	☐ 2254	☐ 2255
Cholestasis, progressive familial intrahepatic, 3	*ABCB4*	☐ 2256	☐ 2257	☐ 2258
Cholestasis, progressive familial intrahepatic, 4	*TJP2*	☐ 2259	☐ 2260	☐ 2261
Chondrocalcinosis 2	*ANKH*	☐ 1079	☐ 1505	☐ 1506
Chondrodysplasia, Blomstrand type	*PTH1R*	☐ 2015	☐ 2016	☐ 2017
Chondrodysplasia, Grebe type	*GDF5*	☐ 1845	☐ 1846	☐ 1847
Chondrodysplasia punctata 1, X-linked recessive	*ARSL*	☐ 2018	☐ 2019	☐ 2020
Chondrodysplasia punctata 2, X-linked dominant	*EBP*	☐ 2021	☐ 2022	☐ 2023
Chondrodysplasia with joint dislocations, GPAPP type	*IMPAD1*	☐ 1848	☐ 1849	☐ 1850
Cleidocranial dysplasia	*RUNX2*	☐ 1080	☐ 1392	☐ 1507
Cohen syndrome	*VPS13B*	☐ 2454	☐ 2455	☐ 2456
Cole-Carpenter syndrome 1	*P4HB*	☐ 2081	☐ 2082	☐ 2083
Cole-Carpenter syndrome 2	*SEC24D*	☐ 2084	☐ 2085	☐ 2086
Congenital contractural arachnodactyly	*FBN2*	☐ 1081	☐ 1082	☐ 1508
Cornelia de Lange syndrome 1	*NIPBL*	☐ 2424	☐ 2425	☐ 2426
Craniodiaphyseal dysplasia, autosomal dominant	*SOST*	☐ 1787	☐ 1788	☐ 1789
Cranioectodermal dysplasia 2	*WDR35*	☐ 1083	☐ 1084	☐ 1509
Cranioectodermal dysplasia 4	*WDR19*	☐ 1085	☐ 1086	☐ 1510
Craniofrontonasal syndrome	*EFNB1*	☐ 1930	☐ 1931	☐ 1932
Craniometaphyseal dysplasia, autosomal dominant	*ANKH*	☐ 1087	☐ 1511	☐ 1512
CRANIOSYNOSTOSIS				
Apert syndrome	*FGFR2 Ex 8*	☐ 1088		
Beare-Stevenson cutis gyrata syndrome	*FGFR2* Ex 8, 11b	☐ 1091		
Craniosynostosis, type 1	*TWIST1*	☐ 1092	☐ 1093	☐ 1513
Crouzon Syndrome	*FGFR2* Ex 8, 10	☐ 1094		
Crouzon syndrome with acanthosis nigricans	*FGFR3* Ex 10	☐ 1096		
Jackson-Weiss syndrome	*FGFR2* Ex 8, 10	☐ 1097		
Muenke syndrome	*FGFR3* Ex 7	☐ 1098		
Pfeiffer syndrome	*FGFR1* Ex 10 & *FGFR2* Ex 8,10	☐ 1099		
Saethre-Chotzen syndrome	*TWIST1 & FGFR3 Ex 7*	☐ 1101	☐ 1102	☐ 5345
FGFR1, FGFR2 & FGFR3 related craniosynostosis panel	*FGFR1* Ex 10, *FGFR2* Ex 8,10 *FGFR3* Ex 7,10	☐ 1103		
FGFR1, FGFR2, FGFR3 & TWIST1 related craniosynostosis panel	*FGFR1* Ex 10 *FGFR2* Ex 8,10 *FGFR3* Ex 7,10 *TWIST1*	☐ 1104		
FGFR2 related craniosynostosis	*FGFR2*	☐ 1105	☐ 1106	☐ 1514
Cutaneomucosal venous malformations	*TEK*	☐ 1733	☐ 1734	☐ 1735
Cutis laxa, autosomal dominant 1	*ELN*	☐ 1515	☐ 1516	☐ 1517
Cutis laxa, autosomal dominant 2 & autosomal recessive, type IA	*FBLN5*	☐ 1107	☐ 1108	☐ 1518
Cutis laxa, autosomal recessive, type IB	*EFEMP2*	☐ 1109	☐ 1110	☐ 1523
Cutis laxa, autosomal recessive, type IC	*LTBP4*	☐ 1697	☐ 1698	☐ 1699
Cutis laxa, autosomal recessive, type IIA	*ATP6V0A2*	☐ 1700	☐ 1701	☐ 1702
Cutis laxa, autosomal recessive, type IIB & type IIIB	*PYCR1*	☐ 1703	☐ 1704	☐ 1705
Cutis laxa, autosomal recessive, type IIIA	*ALDH18A1*	☐ 1706	☐ 1707	☐ 1708
Czech dysplasia	*COL2A1*	☐ 2319	☐ 2320	☐ 2321
Darier-White disease	*ATP2A2*	☐ 5370	☐ 5371	☐ 5372
Desbuquois dysplasia 1	*CANT1*	☐ 1113	☐ 1114	☐ 1524
Desbuquois dysplasia 2	*XYLT1*	☐ 1851	☐ 1852	☐ 1853
Diaphanospondylodysostosis	*BMPER*	☐ 1115	☐ 1525	☐ 1526
Diastrophic dysplasia	*SLC26A2*	☐ 1116	☐ 1117	☐ 1527
Digital arthropathy-brachydactyly, familial	*TRPV4*	☐ 1118	☐ 1762	☐ 1763
Dyggve-Melchior-Clausen disease	*DYM*	☐ 1119	☐ 1120	☐ 1528
Dyssegmental dysplasia, Silverman-Handmaker type	*HSPG2*	☐ 1121	☐ 1122	☐ 1529
Ectodermal dysplasia 1, hypohidrotic, X-linked	*EDA*	☐ 2029	☐ 2030	☐ 2031
Ectopia lentis, isolated, autosomal dominant	*FBN1*	☐ 1123	☐ 1124	☐ 1530
Ectopia lentis, isolated, autosomal recessive	*ADAMTSL4*	☐ 1125	☐ 1126	☐ 1531
Ehlers-Danlos syndrome, classic type, 1	*COL5A1*	☐ 1130	☐ 1131	☐ 1533
Ehlers-Danlos syndrome, classic type 2	*COL5A2*	☐ 1132	☐ 1133	☐ 1534
Ehlers-Danlos syndrome, kyphoscoliotic type, 1	*PLOD1*	☐ 1137	☐ 2433	☐ 2434
Ehlers-Danlos syndrome, musculocontractural type, 1	*CHST14*	☐ 1127	☐ 1128	☐ 1532

Disorder	Genes	Seq	Del/ Dup	Seq & Del/Dup
Ehlers-Danlos syndrome, spondylodysplastic type, 1	*B4GALT7*	☐ 1951	☐ 1952	☐ 1953
Ehlers-Danlos syndrome, spondylodysplastic type, 2	*B3GALT6*	☐ 2032	☐ 5271	☐ 5272
Ehlers-Danlos syndrome, spondylodysplastic type, 3	*SLC39A13*	☐ 1129	☐ 1754	☐ 1755
Ehlers-Danlos syndrome, vascular type	*COL3A1*	☐ 1135	☐ 1136	☐ 1537
Eiken syndrome	*PTH1R*	☐ 2038	☐ 2039	☐ 2040
Epidermolysis bullosa dystrophica, autosomal dominant & recessive	*COL7A1*	☐ 1790	☐ 1791	☐ 1792
Exostoses, multiple, types I	*EXT1*	☐ 1142	☐ 1143	☐ 1540
Exostoses, multiple, types II	*EXT2*	☐ 1144	☐ 1145	☐ 1541
Exudative vitreoretinopathy 1	*FZD4*	☐ 1393	☐ 1394	☐ 1544
Exudative vitreoretinopathy 2, X-linked	*NDP*	☐ 1395	☐ 1396	☐ 1545
Exudative vitreoretinopathy 4	*LRP5*	☐ 1147	☐ 1148	☐ 1546
Exudative vitreoretinopathy 5	*TSPAN12*	☐ 1397	☐ 1398	☐ 1547
Fabry disease	*GLA*	☐ 2463	☐ 2464	☐ 2465
Failure of tooth eruption, primary	*PTH1R*	☐ 2041	☐ 2042	☐ 2043
Familial transthyretin amyloidosis	*TTR*	☐ 7001		
Fibrochondrogenesis 1	*COL11A1*	☐ 1149	☐ 1150	☐ 1550
Fibrochondrogenesis 2	*COL11A2*	☐ 1151	☐ 1152	☐ 1551
Fibrodysplasia ossificans progressiva	*ACVR1*	☐ 1796	☐ 1797	☐ 1798
Fibular hypoplasia and complex brachydactyly	*GDF5*	☐ 1857	☐ 1858	☐ 1859
Focal dermal hypoplasia	*PORCN*	☐ 1153	☐ 1154	☐ 1552
Frontometaphyseal dysplasia	*FLNA*	☐ 1155	☐ 5277	☐ 5278
Frontonasal dysplasia 1	*ALX3*	☐ 1921	☐ 1922	☐ 1923
Frontonasal dysplasia 2	*ALX4*	☐ 1924	☐ 1925	☐ 1926
Frontonasal dysplasia 3	*ALX1*	☐ 1927	☐ 1928	☐ 1929
Frontonasal dysplasia, SIX2 related	*SIX2*	☐ 2357	☐ 2358	☐ 2359
Geleophysic dysplasia 1	*ADAMTSL2*	☐ 1156	☐ 1157	☐ 1553
Geleophysic dysplasia 2	*FBN1* Ex 41, 42	☐ 1158		
Genitopatellar syndrome	*KAT6B*	☐ 1799	☐ 1800	☐ 1801
Glass syndrome	*SATB2*	☐ 2153	☐ 2154	☐ 2155
Glomuvenous malformations	*GLMN*	☐ 1159	☐ 1160	☐ 1554
Gnathodiaphyseal dysplasia	*ANO5*	☐ 2156	☐ 2157	☐ 2158
Gracile bone dysplasia	*FAM111A*	☐ 1802	☐ 1803	☐ 1804
Greig cephalopolysyndactyly syndrome	*GLI3*	☐ 1161	☐ 1162	☐ 1555
Hajdu-Cheney syndrome	*NOTCH2*	☐ 1400	☐ 1401	☐ 1556
Hereditary hemorrhagic telangiectasia type 1	*ENG*	☐ 1404	☐ 1405	☐ 1557
Hereditary hemorrhagic telangiectasia type 2	*ACVRL1*	☐ 1406	☐ 1407	☐ 1558
Hereditary hemorrhagic telangiectasia type 5	*GDF2*	☐ 1718	☐ 1719	☐ 1720
Hereditary motor and sensory neuropathy, type IIC	*TRPV4*	☐ 1163	☐ 1764	☐ 1765
Homocystinuria	*CBS*	☐ 1164	☐ 1165	☐ 1563
Hyperostosis corticalis generalisata	*SOST*	☐ 1805	☐ 1806	☐ 1807
Hyperostosis corticalis generalisata, benign form of worth, with torus palatinus	*LRP5*	☐ 1166	☐ 1167	☐ 1564
Hyperparathyroidism, neonatal severe	*CASR*	☐ 1742	☐ 1743	☐ 1744
Hyperphosphatemic familial tumoral calcinosis	*FGF23*	☐ 2363	☐ 2364	☐ 2365
Hyperphosphatemic familial tumoral calcinosis	*GALNT3*	☐ 2366	☐ 2435	☐ 2436
Hypocalcemia, autosomal dominant 1	*CASR*	☐ 1745	☐ 1746	☐ 1747
Hypocalciuric hypercalcemia, familial, type 1	*CASR*	☐ 1751	☐ 1752	☐ 1753
Hypophosphatasia, infantile, childhood, or adult type	*ALPL*	☐ 1168	☐ 1169	☐ 1565
Inclusion body myopathy with early-onset Paget disease with or without frontotemporal dementia 1	*VCP*	☐ 2400	☐ 2401	☐ 2402
Insulin-like growth factor I deficiency	*IGF1*	☐ 2087	☐ 2088	☐ 2089
Insulin-like growth factor I, resistance to	*IGF1R*	☐ 2090	☐ 2091	☐ 2092
Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome	*SMAD4*	☐ 1410	☐ 1411	☐ 1566
KBG syndrome	*ANKRD11*	☐ 1966	☐ 1967	☐ 1968
Kenny-Caffey syndrome, type 1	*TBCE*	☐ 1903	☐ 1904	☐ 1905
Kenny-Caffey syndrome, type 2	*FAM111A*	☐ 1808	☐ 1809	☐ 1810
Klippel-Feil syndrome 1	*GDF6*	☐ 1954	☐ 1955	☐ 1956
Klippel-Feil syndrome 2	*MEOX1*	☐ 1957	☐ 1958	☐ 1959
Klippel-Feil syndrome 3	*GDF3*	☐ 1960	☐ 1961	☐ 1962
Klippel-Feil syndrome, RIPPLY2 related	*RIPPLY2*	☐ 2324	☐ 2348	☐ 2349
Kniest dysplasia	*COL2A1*	☐ 1170	☐ 1171	☐ 1567
Knobloch syndrome 1	*COL18A1*	☐ 1942	☐ 1943	☐ 1944
Langer mesomelic dysplasia	*SHOX*	☐ 2327	☐ 2328	☐ 2329
Larsen syndrome, autosomal dominant	*FLNB*	☐ 1172	☐ 2394	☐ 2395
	FLNB Ex 2, 3, 4, 27, 28, 29	☐ 1173		
Larsen syndrome, autosomal recessive	*CHST3*	☐ 1175	☐ 1176	☐ 1568
Lateral meningocele syndrome	*NOTCH3*	☐ 2096	☐ 2097	☐ 2098
Leri-Weill dyschondrosteosis	*SHOX*	☐ 2330	☐ 2331	☐ 2332
Loeys-Dietz syndrome 1	*TGFBR1*	☐ 1177	☐ 1178	☐ 1569
Loeys-Dietz syndrome 2	*TGFBR2*	☐ 1179	☐ 1180	☐ 1570
Loeys-Dietz syndrome 3	*SMAD3*	☐ 1182	☐ 1183	☐ 1573
Loeys-Dietz syndrome 4	*TGFB2*	☐ 1184	☐ 1185	☐ 1574
Loeys-Dietz syndrome 5	*TGFB3*	☐ 2133	☐ 2134	☐ 2135
Lysosomal acid lipase deficiency	*LIPA*	☐ 2403	☐ 2404	☐ 2405
Mandibulofacial dysostosis, Guion-Almeida type	*EFTUD2*	☐ 2159	☐ 2160	☐ 2161
Marfan syndrome, type I	*FBN1*	☐ 1187	☐ 1188	☐ 1577
Marfan syndrome, type II	*TGFBR2*	☐ 1189	☐ 1190	☐ 1578
Marshall-Smith syndrome	*NFIX*	☐ 1969	☐ 1970	☐ 1971
Marshall syndrome	*COL11A1*	☐ 1193	☐ 1194	☐ 1583
Meier-Gorlin syndrome 1	*ORC1*	☐ 1860	☐ 1861	☐ 1862
Meier-Gorlin syndrome 2	*ORC4*	☐ 1863	☐ 1864	☐ 1865
Meier-Gorlin syndrome 3	*ORC6*	☐ 1866	☐ 1867	☐ 1868
Meier-Gorlin syndrome 4	*CDT1*	☐ 1869	☐ 1870	☐ 1871
Meier-Gorlin syndrome 5	*CDC6*	☐ 1872	☐ 1873	☐ 1874
Melnick-Needles syndrome	*FLNA*	☐ 1195	☐ 5279	☐ 5280
Menkes disease	*ATP7A*	☐ 1196	☐ 1197	☐ 1584
Metaphyseal anadysplasia 1	*MMP13*	☐ 1198	☐ 1199	☐ 1585
Metaphyseal anadysplasia 2	*MMP9*	☐ 1200	☐ 1201	☐ 1586
Metaphyseal chondrodysplasia, Jansen type	*PTH1R*	☐ 2045	☐ 2046	☐ 2047
Metaphyseal chondrodysplasia, Schmid type	*COL10A1*	☐ 1203	☐ 2072	☐ 2073
Metaphyseal dysplasia, Spahr type	*MMP13*	☐ 1839	☐ 1840	☐ 1841
Metaphyseal dysplasia with maxillary hypoplasia with or without brachydactyly	*RUNX2*	☐ 1412	☐ 1413	☐ 1589
Metaphyseal dysplasia without hypotrichosis	*RMRP*	☐ 1204	☐ 1897	☐ 1898
Metatropic dysplasia	*TRPV4*	☐ 1205	☐ 1766	☐ 1767
Microcephaly-capillary malformation syndrome	*STAMBP*	☐ 2162	☐ 2163	☐ 2164
Mitral valve prolapse 2	*DCHS1*	☐ 2333	☐ 2334	☐ 2335
Mowat-Wilson syndrome	*ZEB2*	☐ 2336	☐ 2337	☐ 2338
Multiple epiphyseal dysplasia	*COMP*	☐ 1206	☐ 2074	☐ 2075

Disorder	Genes	Seq	Del/ Dup	Seq & Del/Dup
Multiple joint dislocations, short stature, craniofacial dysmorphism, and congenital heart defects	*B3GAT3*	☐ 2048	☐ 2049	☐ 2050
Multiple pterygium syndrome, lethal type	*CHRNA1*	☐ 2265	☐ 2266	☐ 2267
Multiple pterygium syndrome, lethal type	*CHRND*	☐ 2268	☐ 2269	☐ 2270
Multiple pterygium syndrome, lethal type & Escobar variant	*CHRNG*	☐ 2271	☐ 2272	☐ 2273
Multiple self-healing squamous epithelioma, susceptibility to	*TGFBR1*	☐ 1590	☐ 1591	☐ 1592
Myhre syndrome	*SMAD4*	☐ 1414	☐ 1415	☐ 1416
Nail-Patella syndrome	*LMX1B*	☐ 1210	☐ 5230	☐ 5231
Neu-Laxova syndrome 1	*PHGDH*	☐ 2165	☐ 2166	☐ 2167
Neu-Laxova syndrome 2	*PSAT1*	☐ 2168	☐ 2169	☐ 2170
Neurofibromatosis type I	*NF1*	☐ 2427	☐ 2428	☐ 2429
Neutropenia, severe congenital, X-linked	*WAS*	☐ 2406	☐ 2407	☐ 2408
NKX2-5 related heart malformations	*NKX2-5*	☐ 2409	☐ 2410	☐ 2411
Norrie disease	*NDP*	☐ 1417	☐ 1418	☐ 1593
Occipital horn syndrome	*ATP7A*	☐ 1211	☐ 1212	☐ 1594
Oculodentodigital dysplasia	*GJA1*	☐ 2412	☐ 2413	☐ 2414
Odontoonychodermal dysplasia	*WNT10A*	☐ 1994	☐ 1995	☐ 1996
Ohdo syndrome, SBBYS variant	*KAT6B*	☐ 1811	☐ 1812	☐ 1813
Opsismodysplasia	*INPPL1*	☐ 1596	☐ 1597	☐ 1598
Orofaciodigital syndrome I	*OFD1*	☐ 2280	☐ 2281	☐ 2282
Osteoarthritis with mild chondrodysplasia	*COL2A1*	☐ 1215	☐ 1837	☐ 1838
Osteogenesis imperfecta, types I / II / III / IV	*COL1A1*	☐ 1601	☐ 1217	☐ 1602
Osteogenesis imperfecta, types I / II / III / IV	*COL1A2*	☐ 1603	☐ 1218	☐ 1604
Osteogenesis imperfecta, type V	*IFITM5*	☐ 1219	☐ 1220	☐ 1605
Osteogenesis imperfecta, type VI	*SERPINF1*	☐ 1221	☐ 1222	☐ 1606
Osteogenesis imperfecta, type VII	*CRTAP*	☐ 1223	☐ 1224	☐ 1607
Osteogenesis imperfecta, type VIII	*P3H1*	☐ 1225	☐ 1226	☐ 1608
Osteogenesis imperfecta, type IX	*PPIB*	☐ 1227	☐ 1228	☐ 1609
Osteogenesis imperfecta, type X	*SERPINH1*	☐ 1229	☐ 1230	☐ 1610
Osteogenesis imperfecta, type XI	*FKBP10*	☐ 1231	☐ 1232	☐ 1611
Osteogenesis imperfecta, type XII	*SP7*	☐ 1233	☐ 1234	☐ 1612
Osteogenesis imperfecta, type XIII	*BMP1*	☐ 1235	☐ 1236	☐ 1613
Osteogenesis imperfecta, type XVI	*CREB3L1*	☐ 1878	☐ 1879	☐ 1880
Osteogenesis imperfecta, type XVII	*SPARC*	☐ 2174	☐ 2175	☐ 2176
Osteogenesis imperfecta, PLS3 related	*PLS3*	☐ 1899	☐ 1900	☐ 1901
Osteopathia striata with cranial sclerosis	*AMER1*	☐ 1814	☐ 1815	☐ 1816
Osteopetrosis, autosomal dominant 1	*LRP5*	☐ 1238	☐ 1239	☐ 1614
Osteopetrosis, autosomal dominant 2 & autosomal recessive 4	*CLCN7*	☐ 1240	☐ 1241	☐ 1615
Osteopetrosis, autosomal recessive 1	*TCIRG1*	☐ 1242	☐ 1243	☐ 1616
Osteopetrosis, autosomal recessive 2	*TNFSF11*	☐ 1244	☐ 1245	☐ 1617
Osteopetrosis, autosomal recessive 5	*OSTM1*	☐ 1248	☐ 1249	☐ 1619
Osteopetrosis, autosomal recessive 6	*PLEKHM1*	☐ 1250	☐ 2447	☐ 2448
Osteopetrosis, autosomal recessive 7	*TNFRSF11A*	☐ 1251	☐ 1252	☐ 1620
Osteopetrosis with renal tubular acidosis	*CA2*	☐ 1254	☐ 1255	☐ 1623
Osteoporosis-pseudoglioma syndrome	*LRP5*	☐ 1256	☐ 1257	☐ 1624
Otopalatodigital syndrome, type I/II	*FLNA*	☐ 1258	☐ 5281	☐ 5282
Otospondylomegaepiphyseal dysplasia	*COL11A2*	☐ 1259	☐ 1260	☐ 1625
Paget disease of bone 2	*TNFRSF11A*	☐ 2372	☐ 2373	☐ 2374
Paget disease of bone 3	*SQSTM1*	☐ 2375	☐ 2376	☐ 2377
Paget disease of bone 5	*TNFRSF11B*	☐ 2378	☐ 2379	☐ 2380
Paget disease of bone 6	*ZNF687*	☐ 2381	☐ 2382	☐ 2383
Pallister-Hall syndrome	*GLI3*	☐ 1261	☐ 1262	☐ 1626
Parastremmatic dwarfism	*TRPV4*	☐ 1263	☐ 1768	☐ 1769
Parkes Weber syndrome	*RASA1*	☐ 1264	☐ 1265	☐ 1627
Periventricular nodular heterotopia 1	*FLNA*	☐ 1267	☐ 5283	☐ 5284
Phosphoglycerate dehydrogenase deficiency	*PHGDH*	☐ 2177	☐ 2178	☐ 2179
Phosphoserine aminotransferase deficiency	*PSAT1*	☐ 2180	☐ 2181	☐ 2182
Platyspondylic lethal skeletal dysplasia, Torrance type	*COL2A1*	☐ 1268	☐ 1269	☐ 1628
	COL2A1 Ex 51, 52, 53, 54	☐ 1270		
Pneumothorax, primary spontaneous	*FLCN*	☐ 2283	☐ 2284	☐ 2285
Polydactyly, preaxial IV & postaxial, type A1	*GLI3*	☐ 1272	☐ 1273	☐ 1629
Popliteal pterygium syndrome	*IRF6*	☐ 2286	☐ 2287	☐ 2288
Popliteal pterygium syndrome, lethal type	*RIPK4*	☐ 2289	☐ 2290	☐ 2291
Porencephaly 1	*COL4A1*	☐ 2183	☐ 2184	☐ 2185
Porencephaly 2	*COL4A2*	☐ 2186	☐ 2187	☐ 2188
Postaxial acrofacial dysostosis	*DHODH*	☐ 2192	☐ 2193	☐ 2194
Progressive pseudorheumatoid arthropathy of childhood	*WISP3*	☐ 1276	☐ 1277	☐ 1631
Proliferative vasculopathy and hydranencephaly-hydrocephaly syndrome	*FLVCR2*	☐ 1278	☐ 1279	☐ 1632
Pseudoachondroplasia	*COMP*	☐ 1280	☐ 2076	☐ 2077
Pseudoxanthoma elasticum	*ABCC6*	☐ 1439	☐ 1440	☐ 1633
Pseudoxanthoma elasticum-like disorder with multiple coagulation factor deficiency	*GGCX*	☐ 2101	☐ 2102	☐ 2103
Pulmonary hypertension, primary, 1	*BMPR2*	☐ 1945	☐ 1946	☐ 1947
Pulmonary hypertension, primary, 2	*SMAD9*	☐ 2107	☐ 2108	☐ 2109
Pulmonary hypertension, primary, 3	*CAV1*	☐ 2110	☐ 2111	☐ 2112
Pulmonary hypertension, primary, 4	*KCNK3*	☐ 2113	☐ 2114	☐ 2115
Pulmonary venoocclusive disease 1, autosomal dominant	*BMPR2*	☐ 1948	☐ 1949	☐ 1950
Pulmonary venoocclusive disease 2, autosomal recessive	*EIF2AK4*	☐ 2051	☐ 2052	☐ 2053
Pycnodysostosis	*CTSK*	☐ 1881	☐ 1882	☐ 1883
Rhizomelic chondrodysplasia punctata type 1	*PEX7*	☐ 1997	☐ 1998	☐ 1999
Rhizomelic chondrodysplasia punctata, type 2	*GNPAT*	☐ 2057	☐ 2058	☐ 2059
Rhizomelic chondrodysplasia punctata, type 3	*AGPS*	☐ 2060	☐ 2061	☐ 2062
Rickets, hypophosphatemic, autosomal dominant	*FGF23*	☐ 1283	☐ 2355	☐ 2356
Rickets, hypophosphatemic, autosomal recessive, 1	*DMP1*	☐ 1284	☐ 1285	☐ 1634
Rickets, hypophosphatemic, autosomal recessive, 2	*ENPP1*	☐ 1286	☐ 1287	☐ 1635
Rickets, hypophosphatemic, X-linked dominant	*PHEX*	☐ 1288	☐ 1289	☐ 1636
Rickets, vitamin D-dependent type I	*CYP27B1*	☐ 1290	☐ 1291	☐ 1637
Roberts syndrome & SC phocomelia syndrome	*ESCO2*	☐ 1884	☐ 1885	☐ 1886
Robinow syndrome, autosomal dominant 1	*WNT5A*	☐ 1817	☐ 1818	☐ 1819
Robinow syndrome, autosomal dominant 2	*DVL1*	☐ 2119	☐ 2120	☐ 2121
Robinow syndrome, autosomal recessive	*ROR2*	☐ 1823	☐ 1824	☐ 1825
Rubinstein-Taybi syndrome 1	*CREBBP*	☐ 2292	☐ 2293	☐ 2294
Rubinstein-Taybi syndrome 2	*EP300*	☐ 2295	☐ 2296	☐ 2297
Schopf-Schulz-Passarge syndrome	*WNT10A*	☐ 2000	☐ 2001	☐ 2002

Disorder	Genes	Seq	Del/ Dup	Seq & Del/Dup
Scapuloperoneal spinal muscular atrophy	*TRPV4*	☐ 1292	☐ 1770	☐ 1771
Schneckenbecken dysplasia, INPPL1 related	*INPPL1*	☐ 2301	☐ 2302	☐ 2303
Schwartz-Jampel syndrome, type 1	*HSPG2*	☐ 1293	☐ 1294	☐ 1638
Sclerosteosis	*SOST*	☐ 1826	☐ 1827	☐ 1828
Serpentine fibula-polycystic kidney syndrome	*NOTCH2*	☐ 1419	☐ 1420	☐ 1639
Short-rib thoracic dysplasia 2 with or without polydactyly	*IFT80*	☐ 1040	☐ 1041	☐ 1481
Short-rib thoracic dysplasia 3 with or without polydactyly	*DYNC2H1*	☐ 1298	☐ 1299	☐ 1641
Short-rib thoracic dysplasia 4 with or without polydactyly	*TTC21B*	☐ 1044	☐ 1045	☐ 1483
Short-rib thoracic dysplasia 5 with or without polydactyly	*WDR19*	☐ 1046	☐ 1047	☐ 1484
Short-rib thoracic dysplasia 6 with or without polydactyly	*NEK1*	☐ 1296	☐ 1297	☐ 1640
Short-rib thoracic dysplasia 7 with or without polydactyly	*WDR35*	☐ 1300	☐ 1301	☐ 1642
Short-rib thoracic dysplasia 8 with or without polydactyly	*WDR60*	☐ 1715	☐ 1716	☐ 1717
Short stature, facial dysmorphism, and skeletal anomalies with or without cardiac anomalies	*BMP2*	☐ 2457	☐ 2458	☐ 2459
Short stature, idiopathic, X-linked	*SHOX*	☐ 2339	☐ 2340	☐ 2341
Shprintzen-Goldberg craniosynostosis syndrome	*SKI*	☐ 1302	☐ 1303	☐ 1643
Simpson-Golabi-Behmel syndrome, type 1	*GPC3*	☐ 1972	☐ 1973	☐ 1974
Simpson-Golabi-Behmel syndrome, type 2	*OFD1*	☐ 2304	☐ 2305	☐ 2306
Simpson-Golabi-Behmel syndrome, PIGA related	*PIGA*	☐ 2387	☐ 2388	☐ 2389
Skraban-Deardorff syndrome	*WDR26*	☐ 2460	☐ 2461	☐ 2462
Smith-Lemli-Opitz syndrome	*DHCR7*	☐ 2439	☐ 2443	☐ 2444
Smith-McCort dysplasia	*DYM*	☐ 1304	☐ 1305	☐ 1644
Snyder-Robinson mental retardation syndrome	*SMS*	☐ 2440	☐ 2441	☐ 2442
Sotos syndrome 1	*NSD1*	☐ 1975	☐ 1976	☐ 1977
Sotos syndrome 2	*NFIX*	☐ 1978	☐ 1979	☐ 1980
Spinal muscular atrophy, distal, congenital nonprogessive	*TRPV4*	☐ 1306	☐ 1772	☐ 1773
Spinal muscular atrophy, distal, X-linked 3	*ATP7A*	☐ 1307	☐ 1308	☐ 1645
Spondylocarpotarsal synostosis syndrome	*FLNB*	☐ 1309	☐ 1310	☐ 1646
Spondylocostal dysostosis 1, autosomal recessive	*DLL3*	☐ 1423	☐ 1424	☐ 1647
Spondylocostal dysostosis 2, autosomal recessive	*MESP2*	☐ 1425	☐ 1426	☐ 1648
Spondylocostal dysostosis 3, autosomal recessive	*LFNG*	☐ 1427	☐ 1428	☐ 1649
Spondylocostal dysostosis 4, autosomal recessive	*HES7*	☐ 1429	☐ 1430	☐ 1650
Spondylocostal dysostosis 5, autosomal dominant	*TBX6*	☐ 1778	☐ 1779	☐ 1780
Spondylocostal dysostosis 6, autosomal recessive	*RIPPLY2*	☐ 2342	☐ 2350	☐ 2351
Spondyloenchondrodysplasia with immune dysregulation	*ACP5*	☐ 1829	☐ 1830	☐ 1831
Spondyloepimetaphyseal dysplasia, Missouri type	*MMP13*	☐ 1311	☐ 1312	☐ 1653
Spondyloepimetaphyseal dysplasia, Strudwick type	*COL2A1*	☐ 1313	☐ 1314	☐ 1654
Spondylo-megaepiphyseal-metaphyseal dysplasia	*NKX3-2*	☐ 1890	☐ 1891	☐ 1892
Spondyloepimetaphyseal dysplasia with joint laxity type 2	*KIF22*	☐ 1432	☐ 1433	☐ 1655
Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures	*B3GALT6*	☐ 2066	☐ 5263	☐ 5264
Spondyloepiphyseal dysplasia congenita	*COL2A1*	☐ 1315	☐ 1316	☐ 1656
Spondyloepiphyseal dysplasia, Maroteaux type	*TRPV4*	☐ 1317	☐ 1774	☐ 1775
Spondyloepiphyseal dysplasia tarda, X-linked	*TRAPPC2*	☐ 1318	☐ 1319	☐ 1657
Spondyloepiphyseal dysplasia with congenital joint dislocations	*CHST3*	☐ 1320	☐ 1321	☐ 1658
Spondylometaphyseal dysplasia, Kozlowski type	*TRPV4*	☐ 1322	☐ 1776	☐ 1777
Spondylometaphyseal dysplasia with cone-rod dystrophy	*PCYT1A*	☐ 1832	☐ 1833	☐ 1834
Spondyloocular syndrome	*XYLT2*	☐ 2195	☐ 2196	☐ 2197
Spondyloperipheral dysplasia	*COL2A1*	☐ 1323	☐ 1324	☐ 1659
	COL2A1 Ex 51, 52, 53, 54	☐ 1325		
Steel syndrome	*COL27A1*	☐ 2430	☐ 2431	☐ 2432
Stickler syndrome, type I	*COL2A1*	☐ 1331	☐ 1332	☐ 1662
Stickler syndrome, type II	*COL11A1*	☐ 1333	☐ 1334	☐ 1663
Stickler syndrome, type III	*COL11A2*	☐ 1335	☐ 1336	☐ 1664
Stiff skin syndrome	*FBN1*	☐ 1442	☐ 2445	☐ 2446
Stuve-Wiedemann syndrome	*LIFR*	☐ 1338	☐ 1339	☐ 1667
Supravalvular aortic stenosis	*ELN*	☐ 1668	☐ 1669	☐ 1670
Terminal osseous dysplasia	*FLNA*	☐ 2207	☐ 5285	☐ 5286
Thanatophoric dysplasia, type I / II	*FGFR3*	☐ 1340	☐ 1758	☐ 1759
	FGFR3 Ex 7,10,15,19	☐ 1341		
Three M syndrome 1	*CUL7*	☐ 1343	☐ 1344	☐ 1671
Three M syndrome 2	*OBSL1*	☐ 1345	☐ 1346	☐ 1672
Three M syndrome 3	*CCDC8*	☐ 1347	☐ 1348	☐ 1673
Thrombocytopenia 1	*WAS*	☐ 2415	☐ 2416	☐ 2417
Thrombocytopenia-absent radius syndrome	*RBM8A*	☐ 1349	☐ 1350	☐ 1674
Tooth agenesis, selective, 1	*MSX1*	☐ 1351	☐ 1352	☐ 1675
Tooth agenesis, selective, 4	*WNT10A*	☐ 2003	☐ 2004	☐ 2005
Torg-Winchester syndrome	*MMP2*	☐ 1353	☐ 1354	☐ 1676
Treacher Collins syndrome 1	*TCOF1*	☐ 1355	☐ 1356	☐ 1677
Treacher Collins syndrome 2	*POLR1D*	☐ 1357	☐ 1358	☐ 1678
Treacher Collins syndrome 3	*POLR1C*	☐ 1359	☐ 1360	☐ 1679
Trichodentoosseus syndrome	*DLX3*	☐ 1915	☐ 1916	☐ 1917
Trichorhinophalangeal syndrome, type I / III	*TRPS1*	☐ 1361	☐ 1362	☐ 1680
Van Buchem disease, type 2	*LRP5*	☐ 1369	☐ 1370	☐ 1683
Van der Woude syndrome 1	*IRF6*	☐ 2307	☐ 2308	☐ 2309
Van der Woude syndrome 2	*GRHL3*	☐ 2310	☐ 2311	☐ 2312
Van Maldergem syndrome 1	*DCHS1*	☐ 2345	☐ 2346	☐ 2347
Wagner vitreoretinopathy	*VCAN*	☐ 1371	☐ 1372	☐ 1684
Weaver syndrome	*EZH2*	☐ 1981	☐ 1982	☐ 1983
Weill-Marchesani syndrome 1	*ADAMTS10*	☐ 1375	☐ 1376	☐ 1686
Weill-Marchesani syndrome 2	*FBN1*	☐ 1373	☐ 1374	☐ 1685
Weill-Marchesani syndrome 3	*LTBP2*	☐ 1724	☐ 1725	☐ 1726
Weill-Marchesani-like syndrome	*ADAMTS17*	☐ 1727	☐ 1728	☐ 1729
Weissenbacher-Zweymuller syndrome	*COL11A2*	☐ 1377	☐ 1378	☐ 1687
Wilson disease	*ATP7B*	☐ 1381	☐ 1382	☐ 1689
Wiskott-Aldrich syndrome	*WAS*	☐ 2418	☐ 2419	☐ 2420
Witkop syndrome	*MSX1*	☐ 1383	☐ 1384	☐ 1690
Wolcott-Rallison syndrome	*EIF2AK3*	☐ 1385	☐ 1386	☐ 1691
Wolman disease	*LIPA*	☐ 2421	☐ 2422	☐ 2423
Zimmermann-Laband syndrome 1	*KCNH1*	☐ 2198	☐ 2199	☐ 2200
Zimmermann-Laband syndrome 2	*ATP6V1B2*	☐ 2201	☐ 2202	☐ 2203

Patient Name:	Patient DOB:

Prenatal testing: ☐ Yes ☐ No Anticipated delivery date:_____ ☐ Female ☐ Male

☐ 1389: Maternal cell contamination testing *(recommended for all prenatal testing)*

Has karyotyping been performed? ☐ Yes ☐ No _____

TARGETED VARIANT TESTING

Individual to be tested:

☐ Affected/Symptomatic ☐ Unaffected/Asymptomatic

Proband Name:_____

Proband DOB: _____

Proband Case #: _____

Relationship to Proband: _____

☐ 1387 One Mutation ☐ 2027 Three Mutations

☐ 1388 Two Mutations ☐ 2044 CNV Mutation Deletion/Duplication Analysis

Gene/Mutation: _____

FAMILY HISTORY

Is there a family history of disease for which the patient is being tested? ☐ Yes ☐ No

Relationship to the individual being tested	Maternal	Paternal	DX	Age at DX
	☐	☐		
	☐	☐		
	☐	☐		
	☐	☐		

MEDICAL RECORDS

Clinical history for this individual is required for variant reclassification. Please include relevant clinical notes and other medical records or provide a brief description of the individual's clinical features in this box. If the patient has no relevant clinical features, please indicate in this box.

OTHER NOTES

TERMS OF SERVICES

The contracted price for any tests on the requisition form applies to that test only. Included in the contract price is the service of answering any question regarding test results. Additional charges apply to parental or familial testing. Additional charges may apply to any future requests relating to this test.

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