



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			
<input type="checkbox"/> INSTITUTIONAL BILLING			
FACILITY NAME	CONTACT PERSON		
ADDRESS	PHONE	FAX	
CITY, STATE, ZIP	E-MAIL		
<input type="checkbox"/> SELF PAY (ALSO REQUIRED FOR ALL INSURANCE CASES – PLEASE SEE “INSURANCE” BELOW.)			
<input type="checkbox"/> CHECK <input type="checkbox"/> M.O.	Please make check or money order payable to Health Network Laboratories.	CARD HOLDER NAME	BILLING ZIP CODE
<input type="checkbox"/> MC <input type="checkbox"/> 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)	
<input type="checkbox"/> 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 CTGT via fax 484-244-2904 or emailed to HNLPriorAuth@hnl.com . This form can be found on our website at www.ctgt.net/billing-payments			
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.			

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

<input type="checkbox"/> 5184 NGS	<input type="checkbox"/> 2044 Del / Dup	<input type="checkbox"/> 5287 NGS & Del / Dup	Gene:
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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	<input type="checkbox"/> 5082	<input type="checkbox"/> 5083	<input type="checkbox"/> 5084
Achondrogenesis	COL2A1, SLC26A2, TRIP11	<input type="checkbox"/> 5139	<input type="checkbox"/> 5140	<input type="checkbox"/> 5141
Adams-Oliver syndrome	ARHGAP31, DLL4, DOCK6, EOGT, NOTCH1, RBPJ	<input type="checkbox"/> 1933	<input type="checkbox"/> 1934	<input type="checkbox"/> 1935
Alagille syndrome	ATP8B1, JAG1, NOTCH2	<input type="checkbox"/> 5157	<input type="checkbox"/> 5158	<input type="checkbox"/> 5159
Alport syndrome	COL4A3, COL4A4, COL4A5, COL4A6	<input type="checkbox"/> 5142	<input type="checkbox"/> 5143	<input type="checkbox"/> 5144
Amelogenesis imperfecta and related disorders	AMELX, C4ORF26, DLX3, DSPP, ENAM, FAM20A, FAM83H, GPR68, ITGB6, KLK4, LAMA3, LAMB3, MMP20, SLC24A4, SMOCC2, WDR72	<input type="checkbox"/> 5197	<input type="checkbox"/> 5198	<input type="checkbox"/> 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	<input type="checkbox"/> 5235	<input type="checkbox"/> 5236	<input type="checkbox"/> 5237
Arterial calcification, generalized, of infancy	ABCC6, ENPP1	<input type="checkbox"/> 1438	<input type="checkbox"/> 1478	<input type="checkbox"/> 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	<input type="checkbox"/> 5312	<input type="checkbox"/> 5313	<input type="checkbox"/> 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	<input type="checkbox"/> 5352	<input type="checkbox"/> 5353	<input type="checkbox"/> 5354
Axial spondylometaphyseal dysplasia	C21orf2, NEK1	<input type="checkbox"/> 5254	<input type="checkbox"/> 5255	<input type="checkbox"/> 5256
Bartter syndrome	BSND, CASR, CLCNKA, CLCNKB, GNA11, KCNJ1, MAGED2, SLC12A1, SLC12A3	<input type="checkbox"/> 5391	<input type="checkbox"/> 5392	<input type="checkbox"/> 5393
Basal cell nevus syndrome	PTCH1, PTCH2, SUFU	<input type="checkbox"/> 2220	<input type="checkbox"/> 2221	<input type="checkbox"/> 2222
Bethlem myopathy & Ullrich congenital muscular dystrophy	COL6A1, COL6A2, COL6A3, COL12A1	1059	1486	1487
Blepharocheilodontic syndrome	CDH1, CTNND1	<input type="checkbox"/> 5346	<input type="checkbox"/> 5347	<input type="checkbox"/> 5348
Brittle cornea syndrome	PRDM5, ZNF469	<input type="checkbox"/> 5247	<input type="checkbox"/> 5248	<input type="checkbox"/> 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	<input type="checkbox"/> 5415	<input type="checkbox"/> 5416	<input type="checkbox"/> 5417
Cantu syndrome	ABCC9, KCNJ8	<input type="checkbox"/> 5160	<input type="checkbox"/> 5161	<input type="checkbox"/> 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	<input type="checkbox"/> 5321	<input type="checkbox"/> 5322	<input type="checkbox"/> 5323
Cerebral cavernous malformations	KRIT1, CCM2, PDCD10	<input type="checkbox"/> 1502	<input type="checkbox"/> 1503	<input type="checkbox"/> 1504
Cerebral small vessel disease	COL4A1, COL4A2, CTC1, GLA, HTRA1, NOTCH3, TREX1	<input type="checkbox"/> 5418	<input type="checkbox"/> 5419	<input type="checkbox"/> 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	<input type="checkbox"/> 2262	<input type="checkbox"/> 2263	<input type="checkbox"/> 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	<input type="checkbox"/> 5291	<input type="checkbox"/> 5292	<input type="checkbox"/> 5293
Cockayne syndrome	ERCC6, ERCC8	<input type="checkbox"/> 5367	<input type="checkbox"/> 5368	<input type="checkbox"/> 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, RAPSIN, TNNI2, TNNT3, TPM2, VIPAS39, VPS33B, ZBTB42, ZMPSTE24	<input type="checkbox"/> 5294	<input type="checkbox"/> 5295	<input type="checkbox"/> 5296
Congenital heart disease	CHD7, ELN, GATA4, GATA6, GDF1, JAG1, NKX2-5, NKX2-6, NOTCH1, NOTCH2, NR2F2, TAB2, TBX1, TBX5, TBX20, ZIC3	<input type="checkbox"/> 5148	<input type="checkbox"/> 5149	<input type="checkbox"/> 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, TGFB3R1, TGFB3R2, ZNF469	5433	5434	5435
Cornelia de Lange syndrome and related disorders	AFF4, ANKRD11, HDAC8, KMT2A, NIPBL, RAD21, SMC1A, SMC3	<input type="checkbox"/> 5181	<input type="checkbox"/> 5182	<input type="checkbox"/> 5183
Craniosynostosis core	FGFR1, FGFR2, FGFR3, TCF12, TWIST1	<input type="checkbox"/> 5194	<input type="checkbox"/> 5195	<input type="checkbox"/> 5196
Craniosynostosis	CDC45, CYP26B1, EFN1, ERF, FGFR1, FGFR2, FGFR3, FREM1, GLI3, IFT43, IFT122, IL11RA, MEGF8, MSX2, POR, RAB23, RECQL4, SKI, SLC25A24, TCF12, TGFB3R1, TGFB3R2, 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, PTSS1, SLC02A1, SOST, TBXAS1, TGFB1, TNFRSF11B, TYROBP	<input type="checkbox"/> 5088	<input type="checkbox"/> 5089	<input type="checkbox"/> 5090
Desbuquois dysplasia core	CANT1, CSGALNACT1, IMPAD1, XYLT1	<input type="checkbox"/> 1854	<input type="checkbox"/> 1855	<input type="checkbox"/> 1856
Desbuquois dysplasia and related disorders	B3GALT6, B3GAT3, CANT1, CHST3, CSGALNACT1, FLNB, GZF1, IMPAD1, KIF22, SLC26A2, XYLT1	<input type="checkbox"/> 5124	<input type="checkbox"/> 5125	<input type="checkbox"/> 5126
Diamond-Blackfan anemia	GATA1, RPL11, RPL15, RPL26, RPL27, RPL31, RPL35A, RPL5, RPS10, RPS17, RPS19, RPS24, RPS26, RPS27, RPS28, RPS29, RPS7, TSR2	<input type="checkbox"/> 5288	<input type="checkbox"/> 5289	<input type="checkbox"/> 5290
Distal arthrogyposes	ECEL1, FBN2, MYBPC1, MYH3, MYH8, NALCN, PIEZO2, TNNI2, TNNT3, TPM2	<input type="checkbox"/> 5133	<input type="checkbox"/> 5134	<input type="checkbox"/> 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	<input type="checkbox"/> 5200	<input type="checkbox"/> 5201	<input type="checkbox"/> 5202
Distal myopathy	ANO5, BAG3, CAV3, CRYAB, DES, DNAJB6, DNM2, DYSF, FHL1, FLNC, GNE, LDB3, MATR3, MYH7, MYOT, SQSTM1, TCAP, TIA1, TTN, VCP	<input type="checkbox"/> 5238	<input type="checkbox"/> 5239	<input type="checkbox"/> 5240
Dyggve-Melchior-Clausen disease	DYM, RAB33B	<input type="checkbox"/> 5297	<input type="checkbox"/> 5298	<input type="checkbox"/> 5299
Dyskeratosis congenita	ACD, CTC1, DKC1, NHP2, NOP10, PARN, RTEL1, TERC, TERT, TINF2, USB1, WRAP53	<input type="checkbox"/> 5203	<input type="checkbox"/> 5204	<input type="checkbox"/> 5205
Ectodermal dysplasia	EDA, EDAR, EDARADD, GJB6, HOXC13, KDF1, KREMEN1, KRT74, KRT85, MSX1	<input type="checkbox"/> 5206	<input type="checkbox"/> 5207	<input type="checkbox"/> 5208
Ectopia lentis	ADAMTSL4, FBN1	<input type="checkbox"/> 5163	<input type="checkbox"/> 5164	<input type="checkbox"/> 5165
Ehlers-Danlos syndrome - Dominant	C1R, C1S, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, COL12A1, FLNA	<input type="checkbox"/> 5064	<input type="checkbox"/> 5065	<input type="checkbox"/> 5066
Ehlers-Danlos syndrome - Dominant & Recessive	ADAMTSL2, AEBP1, ATP7A, B3GALT6, B4GALT7, C1R, C1S, CHST14, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, COL12A1, DSE, FKBP14, FLNA, PLOD1, PRDM5, SLC39A13, ZNF469	<input type="checkbox"/> 5067	<input type="checkbox"/> 5068	<input type="checkbox"/> 5069
Ehlers-Danlos syndrome - Recessive	ADAMTSL2, AEBP1, ATP7A, B3GALT6, B4GALT7, CHST14, COL12A1, DSE, FKBP14, PLOD1, PRDM5, SLC39A13, ZNF469	<input type="checkbox"/> 5070	<input type="checkbox"/> 5071	<input type="checkbox"/> 5072
Ehlers-Danlos syndrome, arthrochalasia type NGS panel	COL1A1, COL1A2	<input type="checkbox"/> 1138	<input type="checkbox"/> 1139	<input type="checkbox"/> 1538
Ehlers-Danlos syndrome, classic type	COL5A1, COL5A2	<input type="checkbox"/> 1134	<input type="checkbox"/> 1535	<input type="checkbox"/> 1536
Ehlers-Danlos syndrome core	COL3A1, COL5A1, COL5A2	<input type="checkbox"/> 5209	<input type="checkbox"/> 5210	<input type="checkbox"/> 5211
Ehlers-Danlos syndrome, periodontal type	C1R, C1S	<input type="checkbox"/> 5250	<input type="checkbox"/> 5251	<input type="checkbox"/> 5252
Ehlers-Danlos syndrome, spondylodysplastic type	B3GALT6, B4GALT7, SLC39A13	<input type="checkbox"/> 2035	<input type="checkbox"/> 2036	<input type="checkbox"/> 2037
Ellis-van Creveld syndrome & Weyers acrofacial dysostosis	EVC, EVC2	<input type="checkbox"/> 1140	<input type="checkbox"/> 1141	<input type="checkbox"/> 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	<input type="checkbox"/> 5073	<input type="checkbox"/> 5074	<input type="checkbox"/> 5075
Erythrokeratodermias and related disorders	DSP, GJA1, GJB3, GJB4, KDSR, KRT83, LOR	<input type="checkbox"/> 5373	<input type="checkbox"/> 5374	<input type="checkbox"/> 5375
Exudative vitreoretinopathy	CAPN5, FZD4, KIF11, LRP5, NDP, TSPAN12, ZNF408	<input type="checkbox"/> 1399	<input type="checkbox"/> 1548	<input type="checkbox"/> 1549
Fibrillinopathy	CBS, FBN1, FBN2	<input type="checkbox"/> 5003	<input type="checkbox"/> 5012	<input type="checkbox"/> 5013
Fibrochondrogenesis	COL11A1, COL11A2	<input type="checkbox"/> 1694	<input type="checkbox"/> 1695	<input type="checkbox"/> 1696
Frontometaphyseal dysplasia	FLNA, MAP3K7, TAB2	<input type="checkbox"/> 5212	<input type="checkbox"/> 5213	<input type="checkbox"/> 5214
Frontonasal dysplasia and craniofrontonasal syndrome	ALX3, ALX4, ALX1, EFNB1, SIX2	<input type="checkbox"/> 1918	<input type="checkbox"/> 1919	<input type="checkbox"/> 1920
Glomuvenous malformations / Cutaneomucosal venous malformations	GLMN, TEK	<input type="checkbox"/> 1736	<input type="checkbox"/> 1737	<input type="checkbox"/> 1738
Hereditary hemorrhagic telangiectasia	ACVRL1, ENG, GDF2, RASA1, SMAD4	<input type="checkbox"/> 1721	<input type="checkbox"/> 1722	<input type="checkbox"/> 1723
Hyperphosphatemic familial tumoral calcinosis	FGF23, GALNT3, KL	<input type="checkbox"/> 2369	<input type="checkbox"/> 2370	<input type="checkbox"/> 2371
Hyperuricemic nephropathy, familial juvenile	REN, SEC61A1, UMOD	<input type="checkbox"/> 5400	<input type="checkbox"/> 5401	<input type="checkbox"/> 5402
Hypotrichosis	APCDD1, CDSN, DSG4, HR, KRT25, KRT71, KRT74, LIPH, LPAR6, RPL21, SNRPE	<input type="checkbox"/> 5306	<input type="checkbox"/> 5307	<input type="checkbox"/> 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, SUL2B1, TGM1, TGM5	<input type="checkbox"/> 5091	<input type="checkbox"/> 5092	<input type="checkbox"/> 5093
Insulin-like growth factor I deficiency and Insulin-like growth factor I, resistance to	IGF1, IGF1R	<input type="checkbox"/> 2093	<input type="checkbox"/> 2094	<input type="checkbox"/> 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	<input type="checkbox"/> 5136	<input type="checkbox"/> 5137	<input type="checkbox"/> 5138
Kabuki syndrome	HNRNPK, KDM6A, KMT2D, RAP1A, RAP1B	<input type="checkbox"/> 5427	<input type="checkbox"/> 5428	<input type="checkbox"/> 5429
Kenny-Caffey syndrome	FAM111A, TBCE	<input type="checkbox"/> 1906	<input type="checkbox"/> 1907	<input type="checkbox"/> 1908
Keratoconus and related disorders	MIR184, PRDM5, VSX1, ZNF469	<input type="checkbox"/> 5421	<input type="checkbox"/> 5422	<input type="checkbox"/> 5423
Klippel-Feil syndrome	GDF3, GDF6, MEOX1, MYO18B, RIPPLY2	<input type="checkbox"/> 1963	<input type="checkbox"/> 1964	<input type="checkbox"/> 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, MERK1, MYO7A, NMNAT1, OTX2, PRPH2, RD3, RDH5, RDH12, ROM1, RPE65, RPGRIP1, SNRNP200, SPATA7, TULP1	<input type="checkbox"/> 5268	<input type="checkbox"/> 5269	<input type="checkbox"/> 5270
Lethal congenital contractural syndrome and related disorders	ADCY6, ADGRG6, CHRNA1, CHRND, CHRNG, CNTNAP1, DNM2, DOK7, ERBB3, GLDN, GLE1, LGI4, LMNA, MUSK, MYBPC1, NEK9, PIP5K1C, RAPSIN, VIPAS39, VPS33B, ZBTB42, ZMPSTE24	<input type="checkbox"/> 5300	<input type="checkbox"/> 5301	<input type="checkbox"/> 5302
Limb girdle muscular dystrophy	ANO5, CAPN3, CAV3, DAG1, DES, DNAJB6, DYSF, EMD, FHL1, FKRP, FKTN, FLNC, GAA, GMPBB, HNRNPD, ISPD, LARGE1, LIMS2, LMNA, MYOT, PLEC, POMGNT1, POMK, POMT1, POMT2, SGCA, SGCB, SGCD, SGCG, TCAP, TNPO3, TOR1AIP1, TRAPPC11, TRIM32, TTN	<input type="checkbox"/> 5215	<input type="checkbox"/> 5216	<input type="checkbox"/> 5217
Lissencephaly core	ARX, CDK5, DCX, KATNB1, LAMB1, NDE1, PAFAH1B1, RELN, TMTC3, TUBA1A	<input type="checkbox"/> 5403	<input type="checkbox"/> 5404	<input type="checkbox"/> 5405
Lissencephaly and related disorders	ACTB, ACTG1, ADGRG1, ARX, ATP6V0A2, B3GALNT2, B4GAT1, CDK5, DAG1, DCX, DYNC1H1, FKRP, FKTN, GMPBB, 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	<input type="checkbox"/> 5406	<input type="checkbox"/> 5407	<input type="checkbox"/> 5408
Loeys-Dietz syndrome core	TGFB1, TGFB2	<input type="checkbox"/> 1181	<input type="checkbox"/> 1571	<input type="checkbox"/> 1572

NGS panel	Genes	Seq	Del/Dup	Seq & Del/Dup
Loeys-Dietz syndrome	SMAD3, TGFB1, TGFB2, TGFB3	<input type="checkbox"/> 2208	<input type="checkbox"/> 2209	<input type="checkbox"/> 2210
Long QT syndrome	AKAP9, ANK2, CACNA1C, CALM1, CALM2, CALM3, CAV3, KCNE1, KCNE2, KCNH2, KCNJ2, KCNJ5, KCNQ1, NOS1AP, RYR2, SCN4B, SCN5A, SNTA1, TRDN	<input type="checkbox"/> 5324	<input type="checkbox"/> 5325	<input type="checkbox"/> 5326
Marfan syndrome, type I / II	FBN1, TGFB2	<input type="checkbox"/> 1191	<input type="checkbox"/> 1579	<input type="checkbox"/> 1580
Marfan syndrome and Loeys-Dietz syndrome core	FBN1, TGFB1, TGFB2	<input type="checkbox"/> 5260	<input type="checkbox"/> 5261	<input type="checkbox"/> 5262
Marfan syndrome and Loeys-Dietz syndrome	FBN1, SMAD3, TGFB1, TGFB2, TGFB3	<input type="checkbox"/> 1192	<input type="checkbox"/> 1581	<input type="checkbox"/> 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, TGFB1, TGFB2	<input type="checkbox"/> 5076	<input type="checkbox"/> 5077	<input type="checkbox"/> 5078
Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome	AKT3, CCND2, PIK3R2	<input type="checkbox"/> 5409	<input type="checkbox"/> 5410	<input type="checkbox"/> 5411
Meier-Gorlin syndrome	CDC45, CDC6, CDT1, GMNN, MCM5, ORC1, ORC4, ORC6	<input type="checkbox"/> 1875	<input type="checkbox"/> 1876	<input type="checkbox"/> 1877
Metaphyseal anadysplasia	MMP13, MMP9	<input type="checkbox"/> 1202	<input type="checkbox"/> 1587	<input type="checkbox"/> 1588
Microcephalic primordial dwarfism	ATR, ATRIP, CDC45, CDC6, CDT1, CENPJ, CEP63, CEP152, DNA2, DONSON, GMNN, LIG4, NIN, ORC1, ORC4, ORC6, PCNT, RBBP8, RNU4ATAC, TRAP1, XRCC4	<input type="checkbox"/> 5166	<input type="checkbox"/> 5167	<input type="checkbox"/> 5168
Mucopolysaccharidosis	ARSB, GALNS, GLB1, GNPTAB, GNPTG, GNS, GUSB, HGSNAT, HYAL1, IDUA, IDS, MCOLN1, NAGLU, SGSH	<input type="checkbox"/> 5241	<input type="checkbox"/> 5242	<input type="checkbox"/> 5243
Multiple epiphyseal dysplasia (MED)	CANT1, COL2A1, COL9A1, COL9A2, COL9A3, COMP, MATN3, SLC26A2	<input type="checkbox"/> 5094	<input type="checkbox"/> 5095	<input type="checkbox"/> 5096
Multiple exostoses	EXT1, EXT2	<input type="checkbox"/> 1146	<input type="checkbox"/> 1542	<input type="checkbox"/> 1543
Multiple pterygium syndrome, lethal type	CHRNA1, CHRND, CHRNG	<input type="checkbox"/> 2274	<input type="checkbox"/> 2275	<input type="checkbox"/> 2276
Myofibrillar myopathy and related disorders	ACTA1, BAG3, CRYAB, DES, DNAJB6, FHL1, FLNC, KY, LDB3, MYOT, PYROXD1	<input type="checkbox"/> 5218	<input type="checkbox"/> 5219	<input type="checkbox"/> 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	<input type="checkbox"/> 5333	<input type="checkbox"/> 5334	<input type="checkbox"/> 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	<input type="checkbox"/> 5394	<input type="checkbox"/> 5395	<input type="checkbox"/> 5396
Nephrotic syndrome and related disorders	ACTN4, ANLN, ARHGAP24, ARHGDI1, ARHGDI2, 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	<input type="checkbox"/> 5221	<input type="checkbox"/> 5222	<input type="checkbox"/> 5223
Neu-Laxova syndrome	PHGDH, PSAT1	<input type="checkbox"/> 2171	<input type="checkbox"/> 2172	<input type="checkbox"/> 2173
Neurofibromatosis and related disorders	MLH1, MSH2, MSH6, NF1, NF2, SPRED1	<input type="checkbox"/> 5191	<input type="checkbox"/> 5192	<input type="checkbox"/> 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	<input type="checkbox"/> 5079	<input type="checkbox"/> 5080	<input type="checkbox"/> 5081
Noonan syndrome core	BRAF, CBL, HRAS, KRAS, LZTR1, MAP2K1, MAP2K2, NRAS, PTPN11, RAF1, RIT1, SHOC2, SOS1, SOS2	<input type="checkbox"/> 5349	<input type="checkbox"/> 5350	<input type="checkbox"/> 5351
Oligodontia - Selective tooth agenesis	AXIN2, EDA, LRP6, LTBP3, MSX1, PAX9, PTH1R, WNT10A, WNT10B	<input type="checkbox"/> 5169	<input type="checkbox"/> 5170	<input type="checkbox"/> 5171
Omodysplasia	FZD2, GPC6	<input type="checkbox"/> 1213	<input type="checkbox"/> 1214	<input type="checkbox"/> 1595
Opitz GBBB syndrome	MID1, SPECC1L	<input type="checkbox"/> 5355	<input type="checkbox"/> 5356	<input type="checkbox"/> 5357
Osteogenesis imperfecta COL1A1 & COL1A2	COL1A1, COL1A2	<input type="checkbox"/> 1216	<input type="checkbox"/> 1599	<input type="checkbox"/> 1600
Osteogenesis imperfecta core	COL1A1, COL1A2, IFITM5	<input type="checkbox"/> 5232	<input type="checkbox"/> 5233	<input type="checkbox"/> 5234
Osteogenesis imperfecta - Dominant	ALPL, ANO5, COL1A1, COL1A2, IFITM5, P4HB, PLS3	<input type="checkbox"/> 5097	<input type="checkbox"/> 5098	<input type="checkbox"/> 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	<input type="checkbox"/> 5100	<input type="checkbox"/> 5101	<input type="checkbox"/> 5102
Osteogenesis imperfecta - Recessive	ALPL, BMP1, CREB3L1, CRTAP, FKBP10, LRP5, MBTPS2, P3H1, PLOD2, PLS3, PPIB, SEC24D, SERPINF1, SERPINH1, SP7, SPARC, TAPT1, TMEM38B, WNT1, XYLT2	<input type="checkbox"/> 5103	<input type="checkbox"/> 5104	<input type="checkbox"/> 5105
Osteopetrosis core	TCIRG1, CLCN7, OSTM1	<input type="checkbox"/> 1253	<input type="checkbox"/> 1621	<input type="checkbox"/> 1622
Osteopetrosis	AMER1, CA2, CLCN7, CTSK, FAM20C, FERMT3, LEMD3, LRP5, OSTM1, PLEKHM1, SNX10, TCIRG1, TNFRSF11A, TNFSF11	<input type="checkbox"/> 5106	<input type="checkbox"/> 5107	<input type="checkbox"/> 5108
Osteopetrosis & Dense bone dysplasia	AMER1, ANKH, CA2, CLCN7, COL1A1, CTSK, DLX3, FAM20C, FERMT3, GJA1, HPGD, LEMD3, LRP4, LRP5, MTAP, OSTM1, PLEKHM1, PTSS1, SLC02A1, SNX10, SOST, TBXAS1, TCIRG1, TGFB1, TNFRSF11A, TNFRSF11B, TNFSF11, TYROBP	<input type="checkbox"/> 5109	<input type="checkbox"/> 5110	<input type="checkbox"/> 5111
Overgrowth syndrome	EED, EZH2, GPC3, NFIX, NSD1, OFD1, PIGA	<input type="checkbox"/> 1984	<input type="checkbox"/> 1985	<input type="checkbox"/> 1986
Paget disease of bone and related disorders	HNRNPA1, HNRNPA2B1, SQSTM1, TNFRSF11A, TNFRSF11B, VCP, ZNF687	<input type="checkbox"/> 2384	<input type="checkbox"/> 2385	<input type="checkbox"/> 2386
Peeling skin syndrome	CAST, CDSN, CHST8, CSTA, FLG2, SERPINB8, TGM5	<input type="checkbox"/> 5376	<input type="checkbox"/> 5377	<input type="checkbox"/> 5378
Platelet bleeding disorders	ACTN1, ANO6, AP3B1, BLOC1S3, BLOC1S6, CD36, DTNBP1, GBA, GF1B, GP1BA, GP1BB, GP6, GP9, HPS1, HPS3, HPS4, HPS5, HPS6, ITGA2B, ITGB3, LYST, MYH9, NBEAL2, P2RY12, PLAU, PRKACG, RASGRP2, SLC11A4, SMPD1, TBXA2R, TBXAS1, VWF, WAS, WIPF1	<input type="checkbox"/> 5154	<input type="checkbox"/> 5155	<input type="checkbox"/> 5156
Polycystic kidney disease and related disorders	ALG9, ANKS6, ATP6V0A4, BICC1, GANAB, GLIS3, HNF1B, INVS, LRP5, MUC1, NOTCH2, NPHP3, OFD1, PKD1, PKD2, PKHD1, SEC61A1, TMEM231, TSC1, TSC2, UMOD, ZNF423	<input type="checkbox"/> 5336	<input type="checkbox"/> 5337	<input type="checkbox"/> 5338
Polycystic liver disease	GANAB, LRP5, PKD1, PKD2, PKHD1, PRKCSH, SEC63	<input type="checkbox"/> 5339	<input type="checkbox"/> 5340	<input type="checkbox"/> 5341
Popliteal pterygium syndrome	IRF6, RIPK4	<input type="checkbox"/> 5412	<input type="checkbox"/> 5413	<input type="checkbox"/> 5414
Porencephaly	COL4A1, COL4A2	<input type="checkbox"/> 2189	<input type="checkbox"/> 2190	<input type="checkbox"/> 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	<input type="checkbox"/> 5379	<input type="checkbox"/> 5380	<input type="checkbox"/> 5381
Progressive familial heart block type I	SCN5A, TRPM4	<input type="checkbox"/> 5327	<input type="checkbox"/> 5328	<input type="checkbox"/> 5329
Pseudoxanthoma elasticum	ABCC6, GGCX	<input type="checkbox"/> 2104	<input type="checkbox"/> 2105	<input type="checkbox"/> 2106
Pulmonary hypertension	ACVRL1, BMPR1B, BMPR2, CAV1, EIF2AK4, ENG, FOXF1, GDF2, KCNA5, KCNK3, SMAD4, SMAD9	<input type="checkbox"/> 2116	<input type="checkbox"/> 2117	<input type="checkbox"/> 2118
Restrictive dermopathy, lethal	LMNA, ZMPSTE24	<input type="checkbox"/> 5382	<input type="checkbox"/> 5383	<input type="checkbox"/> 5384
Renal tubular dysgenesis	ACE, AGT, AGTR1, REN	<input type="checkbox"/> 5397	<input type="checkbox"/> 5398	<input type="checkbox"/> 5399
Robinow syndrome	DVL1, DVL3, FZD2, NXN, ROR2, WNT5A	<input type="checkbox"/> 2127	<input type="checkbox"/> 2128	<input type="checkbox"/> 2129
Rubinstein-Taybi syndrome	CREBBP, EP300	<input type="checkbox"/> 2298	<input type="checkbox"/> 2299	<input type="checkbox"/> 2300
Short QT syndrome	CACNA1C, CACNA2D1, CACNB2, KCNH2, KCNJ2, KCNQ1	<input type="checkbox"/> 5330	<input type="checkbox"/> 5331	<input type="checkbox"/> 5332
Short stature with endocrinopathy	BTK, GH1, GHR, GHRHR, GHSR, HESX1, IGF1, IGF1R, LHX3, LHX4, OTX2, POU1F1, PROP1, SOX3	<input type="checkbox"/> 5358	<input type="checkbox"/> 5359	<input type="checkbox"/> 5360
Skeletal dysplasia ciliopathy	C21orf2, C2CD3, CEP120, DYNC2H1, DYNCL1, EVC, EVC2, ICK, IFT43, IFT52, IFT80, IFT81, IFT122, IFT140, IFT172, KIAA0586, KIAA0753, TCTEX1D2, NEK1, TTC21B, WDR19, WDR34, WDR35, WDR60	<input type="checkbox"/> 5112	<input type="checkbox"/> 5113	<input type="checkbox"/> 5114
Skeletal dysplasia core	ALPL, COL1A1, COL1A2, COL2A1, FGF3, INPPL1, NKX3-2, SLC26A2, SOX9, TRIP11	<input type="checkbox"/> 5115	<input type="checkbox"/> 5116	<input type="checkbox"/> 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	<input type="checkbox"/> 5118	<input type="checkbox"/> 5119	<input type="checkbox"/> 5120
Skeletal dysplasia extended	ARSL, COL10A1, COL11A1, COL11A2, DDR2, EBP, FLNB, HSPG2, LBR, LIFR, MMP9, MMP13, NSDHL, PEX7, PTH1R, RMRP, SBDS, SLC35D1, TRPV4	<input type="checkbox"/> 5121	<input type="checkbox"/> 5122	<input type="checkbox"/> 5123
Skeletal dysplasia and skeletal ciliopathy NGS panel	ALPL, ARSL, C21orf2, C2CD3, CEP120, COL1A1, COL1A2, COL2A1, COL10A1, COL11A1, COL11A2, DDR2, DYNC2H1, DYNC2L1, 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	<input type="checkbox"/> 5430	<input type="checkbox"/> 5431	<input type="checkbox"/> 5432
Smith-McCort dysplasia	DYM, RAB33B	<input type="checkbox"/> 5257	<input type="checkbox"/> 5258	<input type="checkbox"/> 5259
SOTOS syndrome and related disorders	APC2, EZH2, NFIX, NSD1	<input type="checkbox"/> 5424	<input type="checkbox"/> 5425	<input type="checkbox"/> 5426
Spondylocostal dysostosis	DLL3, HES7, LFNG, MESP2, RIPPLY2, TBX6	<input type="checkbox"/> 1781	<input type="checkbox"/> 1782	<input type="checkbox"/> 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, PAPS2, PCYT1A, PTH1R, RAB33B, RMRP, RNU4ATAC, RSPRY1, RUNX2, SBDS, SLC26A2, SMARCAL1, TRAPPC2, TRPV4, WISP3, XYLT1	<input type="checkbox"/> 5185	<input type="checkbox"/> 5186	<input type="checkbox"/> 5187
Spondyloepimetaphyseal dysplasia with joint laxity	B3GALT6, KIF22	<input type="checkbox"/> 2069	<input type="checkbox"/> 2070	<input type="checkbox"/> 2071
Stickler syndrome	COL2A1, COL9A1, COL9A2, COL9A3, COL11A1, COL11A2, LOXL3, VCAN	<input type="checkbox"/> 5127	<input type="checkbox"/> 5128	<input type="checkbox"/> 5129
Stickler syndrome - Recessive	COL9A1, COL9A2, COL9A3	<input type="checkbox"/> 1330	<input type="checkbox"/> 1660	<input type="checkbox"/> 1661
Stickler syndrome core	COL2A1, COL11A1, COL11A2	<input type="checkbox"/> 1337	<input type="checkbox"/> 1665	<input type="checkbox"/> 1666
Three M syndrome	CCDC8, CUL7, OBSL1	<input type="checkbox"/> 5172	<input type="checkbox"/> 5173	<input type="checkbox"/> 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	<input type="checkbox"/> 5224	<input type="checkbox"/> 5225	<input type="checkbox"/> 5226
Treacher Collins syndrome core	POLR1C, POLR1D, TCOF1	<input type="checkbox"/> 5303	<input type="checkbox"/> 5304	<input type="checkbox"/> 5305
Treacher Collins syndrome and related disorders	DHODH, EDNRA, EFTUD2, POLR1A, POLR1C, POLR1D, SF3B4, TCOF1, TXNL4A	<input type="checkbox"/> 5175	<input type="checkbox"/> 5176	<input type="checkbox"/> 5177
Trichothiodystrophy	ERCC2, ERCC3, GTF2E2, GTF2H5, MPLKIP, RNF113A	<input type="checkbox"/> 5385	<input type="checkbox"/> 5386	<input type="checkbox"/> 5387
Tuberous sclerosis	TSC1, TSC2	<input type="checkbox"/> 5342	<input type="checkbox"/> 5343	<input type="checkbox"/> 5344
Van der Woude syndrome	GRHL3, IRF6	<input type="checkbox"/> 2313	<input type="checkbox"/> 2314	<input type="checkbox"/> 2315
Vascular malformations	ACVRL1, BMPR2, CAV1, CCM2, EIF2AK4, ELMO2, ENG, FOXF1, GATA2, GDF2, GLMN, KCNK3, KRIT1, PDCD10, PTEN, RASA1, SMAD4, SMAD9, TEK	<input type="checkbox"/> 5178	<input type="checkbox"/> 5179	<input type="checkbox"/> 5180
Vitreoretinopathy	BEST1, CAPN5, FZD4, COL2A1, COL9A1, COL9A2, COL9A3, COL11A1, COL11A2, COL18A1, KCNJ13, KIF11, LRP5, NDP, NR2E3, RS1, TSPAN12, VCAN, ZNF408	<input type="checkbox"/> 5227	<input type="checkbox"/> 5228	<input type="checkbox"/> 5229
Waardenburg syndrome	EDN3, EDNRB, MITF, PAX3, SNAI2, SOX10, TYR	<input type="checkbox"/> 5244	<input type="checkbox"/> 5245	<input type="checkbox"/> 5246
Weaver and Cohen-Gibson syndrome	EED, EZH2	<input type="checkbox"/> 5265	<input type="checkbox"/> 5266	<input type="checkbox"/> 5267
Weill-Marchesani syndrome	ADAMTS10, ADAMTS17, FBN1, LTBP2	<input type="checkbox"/> 1730	<input type="checkbox"/> 1731	<input type="checkbox"/> 1732
Xeroderma pigmentosum	DDB2, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, POLH, XPA, XPC	<input type="checkbox"/> 5388	<input type="checkbox"/> 5389	<input type="checkbox"/> 5390
Zimmermann-Laband syndrome	KCNH1, ATP6V1B2	<input type="checkbox"/> 2204	<input type="checkbox"/> 2205	<input type="checkbox"/> 2206

SINGLE GENE AND SANGER PANEL TESTS

Disorder	Genes	Seq	Del/Dup	Seq & Del/Dup
Ablepharon-macrostomia syndrome	TWIST2	<input type="checkbox"/> 2138	<input type="checkbox"/> 2139	<input type="checkbox"/> 2140
Achondrogenesis, type IA	TRIP11	<input type="checkbox"/> 1001	<input type="checkbox"/> 1002	<input type="checkbox"/> 1444
Achondrogenesis, type IB	SLC26A2	<input type="checkbox"/> 1003	<input type="checkbox"/> 1004	<input type="checkbox"/> 1445
Achondrogenesis, type II / Hypochondrogenesis	COL2A1	<input type="checkbox"/> 1005	<input type="checkbox"/> 1006	<input type="checkbox"/> 1446
Achondroplasia / Hypochondroplasia	FGFR3	<input type="checkbox"/> 1007	<input type="checkbox"/> 1756	<input type="checkbox"/> 1757
	FGFR3 Ex 3,5, 7,9,10,13,15	<input type="checkbox"/> 1008		
Acrofacial dysostosis 1, Nager type	SF3B4	<input type="checkbox"/> 1988	<input type="checkbox"/> 1989	<input type="checkbox"/> 1990
Acromelic frontonasal dysostosis	ZSWIM6	<input type="checkbox"/> 2006	<input type="checkbox"/> 2007	<input type="checkbox"/> 2008
Acromesomelic dysplasia, Hunter-Thompson type	GDF5	<input type="checkbox"/> 1842	<input type="checkbox"/> 1843	<input type="checkbox"/> 1844
Acromesomelic dysplasia, Maroteaux type	NPR2	<input type="checkbox"/> 1010	<input type="checkbox"/> 1011	<input type="checkbox"/> 1447
Acromicric dysplasia	FBN1 Ex 41,42	<input type="checkbox"/> 1012		
Adams-Oliver syndrome 1	ARHGAP31	<input type="checkbox"/> 1013	<input type="checkbox"/> 1014	<input type="checkbox"/> 1448
Adams-Oliver syndrome 2	DOCK6	<input type="checkbox"/> 1449	<input type="checkbox"/> 1450	<input type="checkbox"/> 1451
Adams-Oliver syndrome 3	RBPJ	<input type="checkbox"/> 1452	<input type="checkbox"/> 1453	<input type="checkbox"/> 1454
Adams-Oliver syndrome 4	EOGT	<input type="checkbox"/> 1455	<input type="checkbox"/> 1456	<input type="checkbox"/> 1457
Adams-Oliver syndrome 5	NOTCH1	<input type="checkbox"/> 1936	<input type="checkbox"/> 1937	<input type="checkbox"/> 1938
Adams-Oliver syndrome 6	DLL4	<input type="checkbox"/> 2352	<input type="checkbox"/> 2353	<input type="checkbox"/> 2354
Alagille syndrome 1	JAG1	<input type="checkbox"/> 1015	<input type="checkbox"/> 1016	<input type="checkbox"/> 1461
Alagille syndrome 2	NOTCH2	<input type="checkbox"/> 1390	<input type="checkbox"/> 1391	<input type="checkbox"/> 1462
Alagille syndrome, ATP8B1 related	ATP8B1	<input type="checkbox"/> 2141	<input type="checkbox"/> 2142	<input type="checkbox"/> 2143
Alopecia universalis congenita & Atrichia with papular lesions	HR	<input type="checkbox"/> 2451	<input type="checkbox"/> 2452	<input type="checkbox"/> 2453

Alport syndrome, X-linked	COL4A5	<input type="checkbox"/> 1017	<input type="checkbox"/> 1018	<input type="checkbox"/> 1463
Amelogenesis imperfecta, type IV	DLX3	<input type="checkbox"/> 1912	<input type="checkbox"/> 1913	<input type="checkbox"/> 1914
Anauxetic dysplasia	RMRP	<input type="checkbox"/> 1019	<input type="checkbox"/> 1893	<input type="checkbox"/> 1894
Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps	COL4A1	<input type="checkbox"/> 2144	<input type="checkbox"/> 2145	<input type="checkbox"/> 2146
Aortic aneurysm, familial thoracic 3	TGFBR2	<input type="checkbox"/> 1020	<input type="checkbox"/> 1021	<input type="checkbox"/> 1464
Aortic aneurysm, familial thoracic 4	MYH11	<input type="checkbox"/> 1023	<input type="checkbox"/> 1024	<input type="checkbox"/> 1467
Aortic aneurysm, familial thoracic 5	TGFBR1	<input type="checkbox"/> 1025	<input type="checkbox"/> 1026	<input type="checkbox"/> 1468
Aortic aneurysm, familial thoracic 6	ACTA2	<input type="checkbox"/> 1029	<input type="checkbox"/> 1030	<input type="checkbox"/> 1473
Aortic aneurysm, familial thoracic 7	MYLK	<input type="checkbox"/> 1031	<input type="checkbox"/> 5273	<input type="checkbox"/> 5274
Aortic aneurysm, familial thoracic 8	PRKG1	<input type="checkbox"/> 1739	<input type="checkbox"/> 1740	<input type="checkbox"/> 1741
Aortic aneurysm, familial thoracic 9	MFAP5	<input type="checkbox"/> 2009	<input type="checkbox"/> 2010	<input type="checkbox"/> 2011
Aortic valve disease 1	NOTCH1	<input type="checkbox"/> 1939	<input type="checkbox"/> 1940	<input type="checkbox"/> 1941
Arterial calcification, generalized, of infancy, 1	ENPP1	<input type="checkbox"/> 1036	<input type="checkbox"/> 1037	<input type="checkbox"/> 1476
Arterial calcification, generalized, of infancy, 2	ABCC6	<input type="checkbox"/> 1436	<input type="checkbox"/> 1437	<input type="checkbox"/> 1477
Arterial tortuosity syndrome	SLC2A10	<input type="checkbox"/> 1038	<input type="checkbox"/> 1039	<input type="checkbox"/> 1480
Atelosteogenesis, type I / III	FLNB	<input type="checkbox"/> 1048	<input type="checkbox"/> 2390	<input type="checkbox"/> 2391
	FLNB Ex 2, 3, 4, 27, 28, 29	<input type="checkbox"/> 1049		
Atelosteogenesis, type II	SLC26A2	<input type="checkbox"/> 1051	<input type="checkbox"/> 1052	<input type="checkbox"/> 1485
Avascular necrosis of femoral head, primary	COL2A1	<input type="checkbox"/> 1053	<input type="checkbox"/> 1835	<input type="checkbox"/> 1836
Barber-Say syndrome	TWIST2	<input type="checkbox"/> 2147	<input type="checkbox"/> 2148	<input type="checkbox"/> 2149
Basal cell nevus syndrome	PTCH1	<input type="checkbox"/> 2211	<input type="checkbox"/> 2212	<input type="checkbox"/> 2213
Basal cell nevus syndrome	PTCH2	<input type="checkbox"/> 2214	<input type="checkbox"/> 2215	<input type="checkbox"/> 2216
Basal cell nevus syndrome	SUFU	<input type="checkbox"/> 2217	<input type="checkbox"/> 2218	<input type="checkbox"/> 2219
Benign chronic pemphigus	ATP2C1	<input type="checkbox"/> 5361	<input type="checkbox"/> 5362	<input type="checkbox"/> 5363

LABORATORY TEST REQUISITION, PAGE 6

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Disorder	Genes	Seq	Del/ Dup	Seq & Del/Dup
Bent bone dysplasia syndrome	FGFR2	<input type="checkbox"/> 1909	<input type="checkbox"/> 1910	<input type="checkbox"/> 1911
Birt-Hogg-Dube syndrome	FLCN	<input type="checkbox"/> 2223	<input type="checkbox"/> 2224	<input type="checkbox"/> 2225
Boomerang dysplasia	FLNB	<input type="checkbox"/> 1060	<input type="checkbox"/> 2392	<input type="checkbox"/> 2393
	FLNB Ex 2, 3, 4, 27, 28, 29	<input type="checkbox"/> 1061		
Brachyolmia type 3	TRPV4	<input type="checkbox"/> 1063	<input type="checkbox"/> 1760	<input type="checkbox"/> 1761
Brain small vessel disease with or without ocular anomalies	COL4A1	<input type="checkbox"/> 2150	<input type="checkbox"/> 2151	<input type="checkbox"/> 2152
Branchiooculofacial syndrome	TFAP2A	<input type="checkbox"/> 2078	<input type="checkbox"/> 2079	<input type="checkbox"/> 2080
Brittle cornea syndrome 1	ZNF469	<input type="checkbox"/> 1064	<input type="checkbox"/> 1065	<input type="checkbox"/> 1488
Brittle cornea syndrome 2	PRDM5	<input type="checkbox"/> 1066	<input type="checkbox"/> 1067	<input type="checkbox"/> 1489
Bruck syndrome 2	PLOD2	<input type="checkbox"/> 1068	<input type="checkbox"/> 1692	<input type="checkbox"/> 1693
Buschke-Ollendorff syndrome	LEMD3	<input type="checkbox"/> 1069	<input type="checkbox"/> 1070	<input type="checkbox"/> 1490
Caffey disease	COL1A1	<input type="checkbox"/> 1071	<input type="checkbox"/> 2449	<input type="checkbox"/> 2450
Campomelic dysplasia	SOX9	<input type="checkbox"/> 1072	<input type="checkbox"/> 1073	<input type="checkbox"/> 1491
Camptodactyly, tall stature, and hearing loss syndrome	FGFR3	<input type="checkbox"/> 2316	<input type="checkbox"/> 2317	<input type="checkbox"/> 2318
Camurati-Engelmann disease	TGFB1	<input type="checkbox"/> 1074	<input type="checkbox"/> 2437	<input type="checkbox"/> 2438
Cantu syndrome	ABCC9	<input type="checkbox"/> 1784	<input type="checkbox"/> 1785	<input type="checkbox"/> 1786
Cantu syndrome, KCNJ8 related	KCNJ8	<input type="checkbox"/> 2226	<input type="checkbox"/> 2227	<input type="checkbox"/> 2228
Capillary malformation arteriovenous malformation	RASA1	<input type="checkbox"/> 1075	<input type="checkbox"/> 1076	<input type="checkbox"/> 1492
Cardiac valvular dysplasia, X-linked	FLNA	<input type="checkbox"/> 1077	<input type="checkbox"/> 5275	<input type="checkbox"/> 5276
Cartilage-hair hypoplasia	RMRP	<input type="checkbox"/> 1078	<input type="checkbox"/> 1895	<input type="checkbox"/> 1896
Catel-Manzke syndrome	TGDS	<input type="checkbox"/> 2012	<input type="checkbox"/> 2013	<input type="checkbox"/> 2014
Cerebral arteriopathy, with subcortical infarcts and leukoencephalopathy 1	NOTCH3	<input type="checkbox"/> 1991	<input type="checkbox"/> 2136	<input type="checkbox"/> 2137
Cerebral cavernous malformations 1	KRIT1	<input type="checkbox"/> 1493	<input type="checkbox"/> 1494	<input type="checkbox"/> 1495
Cerebral cavernous malformations 2	CCM2	<input type="checkbox"/> 1496	<input type="checkbox"/> 1497	<input type="checkbox"/> 1498
Cerebral cavernous malformations 3	PDCD10	<input type="checkbox"/> 1499	<input type="checkbox"/> 1500	<input type="checkbox"/> 1501
CHARGE syndrome	CHD7	<input type="checkbox"/> 2229	<input type="checkbox"/> 2230	<input type="checkbox"/> 2231
Cholestasis, benign recurrent intrahepatic, 1	ATP8B1	<input type="checkbox"/> 2232	<input type="checkbox"/> 2233	<input type="checkbox"/> 2234
Cholestasis, benign recurrent intrahepatic, 2	ABCB11	<input type="checkbox"/> 2235	<input type="checkbox"/> 2236	<input type="checkbox"/> 2237
Cholestasis, intrahepatic, of pregnancy, 1	ATP8B1	<input type="checkbox"/> 2241	<input type="checkbox"/> 2242	<input type="checkbox"/> 2243
Cholestasis, intrahepatic, of pregnancy, 3	ABCB4	<input type="checkbox"/> 2244	<input type="checkbox"/> 2245	<input type="checkbox"/> 2246
Cholestasis, progressive familial intrahepatic, 1	ATP8B1	<input type="checkbox"/> 2250	<input type="checkbox"/> 2251	<input type="checkbox"/> 2252
Cholestasis, progressive familial intrahepatic, 2	ABCB11	<input type="checkbox"/> 2253	<input type="checkbox"/> 2254	<input type="checkbox"/> 2255
Cholestasis, progressive familial intrahepatic, 3	ABCB4	<input type="checkbox"/> 2256	<input type="checkbox"/> 2257	<input type="checkbox"/> 2258
Cholestasis, progressive familial intrahepatic, 4	TJP2	<input type="checkbox"/> 2259	<input type="checkbox"/> 2260	<input type="checkbox"/> 2261
Chondrocalcinosis 2	ANKH	<input type="checkbox"/> 1079	<input type="checkbox"/> 1505	<input type="checkbox"/> 1506
Chondrodysplasia, Blomstrand type	PTH1R	<input type="checkbox"/> 2015	<input type="checkbox"/> 2016	<input type="checkbox"/> 2017
Chondrodysplasia, Grebe type	GDF5	<input type="checkbox"/> 1845	<input type="checkbox"/> 1846	<input type="checkbox"/> 1847
Chondrodysplasia punctata 1, X-linked recessive	ARSL	<input type="checkbox"/> 2018	<input type="checkbox"/> 2019	<input type="checkbox"/> 2020
Chondrodysplasia punctata 2, X-linked dominant	EBP	<input type="checkbox"/> 2021	<input type="checkbox"/> 2022	<input type="checkbox"/> 2023
Chondrodysplasia with joint dislocations, GPAPP type	IMPAD1	<input type="checkbox"/> 1848	<input type="checkbox"/> 1849	<input type="checkbox"/> 1850
Cleidocranial dysplasia	RUNX2	<input type="checkbox"/> 1080	<input type="checkbox"/> 1392	<input type="checkbox"/> 1507
Cohen syndrome	VPS13B	<input type="checkbox"/> 2454	<input type="checkbox"/> 2455	<input type="checkbox"/> 2456
Cole-Carpenter syndrome 1	P4HB	<input type="checkbox"/> 2081	<input type="checkbox"/> 2082	<input type="checkbox"/> 2083
Cole-Carpenter syndrome 2	SEC24D	<input type="checkbox"/> 2084	<input type="checkbox"/> 2085	<input type="checkbox"/> 2086
Congenital contractural arachnodactyly	FBN2	<input type="checkbox"/> 1081	<input type="checkbox"/> 1082	<input type="checkbox"/> 1508
Cornelia de Lange syndrome 1	NIPBL	<input type="checkbox"/> 2424	<input type="checkbox"/> 2425	<input type="checkbox"/> 2426

Craniodiphyseal dysplasia, autosomal dominant	SOST	<input type="checkbox"/> 1787	<input type="checkbox"/> 1788	<input type="checkbox"/> 1789
Craniocotodermal dysplasia 2	WDR35	<input type="checkbox"/> 1083	<input type="checkbox"/> 1084	<input type="checkbox"/> 1509
Craniocotodermal dysplasia 4	WDR19	<input type="checkbox"/> 1085	<input type="checkbox"/> 1086	<input type="checkbox"/> 1510
Craniofrontonasal syndrome	EFNB1	<input type="checkbox"/> 1930	<input type="checkbox"/> 1931	<input type="checkbox"/> 1932
Craniometaphyseal dysplasia, autosomal dominant	ANKH	<input type="checkbox"/> 1087	<input type="checkbox"/> 1511	<input type="checkbox"/> 1512
CRANIOSYNOSTOSIS				
Apert syndrome	FGFR2 Ex 8	<input type="checkbox"/> 1088		
Beare-Stevenson cutis gyrata syndrome	FGFR2 Ex 8, 11b	<input type="checkbox"/> 1091		
Craniostenosis, type 1	Twist1	<input type="checkbox"/> 1092	<input type="checkbox"/> 1093	<input type="checkbox"/> 1513
Crouzon Syndrome	FGFR2 Ex 8, 10	<input type="checkbox"/> 1094		
Crouzon syndrome with acanthosis nigricans	FGFR3 Ex 10	<input type="checkbox"/> 1096		
Jackson-Weiss syndrome	FGFR2 Ex 8, 10	<input type="checkbox"/> 1097		
Muenke syndrome	FGFR3 Ex 7	<input type="checkbox"/> 1098		
Pfeiffer syndrome	FGFR1 Ex 10 & FGFR2 Ex 8,10	<input type="checkbox"/> 1099		
Saethre-Chatzen syndrome	Twist1 & FGFR3 Ex 7	<input type="checkbox"/> 1101	<input type="checkbox"/> 1102	<input type="checkbox"/> 5345
FGFR1, FGFR2 & FGFR3 related craniostenosis panel	FGFR1 Ex 10, FGFR2 Ex 8,10 FGFR3 Ex 7,10	<input type="checkbox"/> 1103		
FGFR1, FGFR2, FGFR3 & Twist1 related craniostenosis panel	FGFR1 Ex 10 FGFR2 Ex 8,10 FGFR3 Ex 7,10 Twist1	<input type="checkbox"/> 1104		
FGFR2 related craniostenosis	FGFR2	<input type="checkbox"/> 1105	<input type="checkbox"/> 1106	<input type="checkbox"/> 1514
Cutaneous mucosal venous malformations	TEK	<input type="checkbox"/> 1733	<input type="checkbox"/> 1734	<input type="checkbox"/> 1735
Cutis laxa, autosomal dominant 1	ELN	<input type="checkbox"/> 1515	<input type="checkbox"/> 1516	<input type="checkbox"/> 1517
Cutis laxa, autosomal dominant 2 & autosomal recessive, type IA	FBLN5	<input type="checkbox"/> 1107	<input type="checkbox"/> 1108	<input type="checkbox"/> 1518
Cutis laxa, autosomal recessive, type IB	EFEMP2	<input type="checkbox"/> 1109	<input type="checkbox"/> 1110	<input type="checkbox"/> 1523
Cutis laxa, autosomal recessive, type IC	LTBP4	<input type="checkbox"/> 1697	<input type="checkbox"/> 1698	<input type="checkbox"/> 1699
Cutis laxa, autosomal recessive, type IIA	ATP6V0A2	<input type="checkbox"/> 1700	<input type="checkbox"/> 1701	<input type="checkbox"/> 1702
Cutis laxa, autosomal recessive, type IIB & type IIIB	PYCR1	<input type="checkbox"/> 1703	<input type="checkbox"/> 1704	<input type="checkbox"/> 1705
Cutis laxa, autosomal recessive, type IIIA	ALDH18A1	<input type="checkbox"/> 1706	<input type="checkbox"/> 1707	<input type="checkbox"/> 1708
Czech dysplasia	COL2A1	<input type="checkbox"/> 2319	<input type="checkbox"/> 2320	<input type="checkbox"/> 2321
Darier-White disease	ATP2A2	<input type="checkbox"/> 5370	<input type="checkbox"/> 5371	<input type="checkbox"/> 5372
Desbuquois dysplasia 1	CANT1	<input type="checkbox"/> 1113	<input type="checkbox"/> 1114	<input type="checkbox"/> 1524
Desbuquois dysplasia 2	XYLT1	<input type="checkbox"/> 1851	<input type="checkbox"/> 1852	<input type="checkbox"/> 1853
Diaphanospondylodysostosis	BMPER	<input type="checkbox"/> 1115	<input type="checkbox"/> 1525	<input type="checkbox"/> 1526
Diastrophic dysplasia	SLC26A2	<input type="checkbox"/> 1116	<input type="checkbox"/> 1117	<input type="checkbox"/> 1527
Digital arthropathy-brachydactyly, familial	TRPV4	<input type="checkbox"/> 1118	<input type="checkbox"/> 1762	<input type="checkbox"/> 1763
Dyggve-Melchior-Clausen disease	DYM	<input type="checkbox"/> 1119	<input type="checkbox"/> 1120	<input type="checkbox"/> 1528
Dyssegmental dysplasia, Silverman-Handmaker type	HSPG2	<input type="checkbox"/> 1121	<input type="checkbox"/> 1122	<input type="checkbox"/> 1529
Ectodermal dysplasia 1, hypohidrotic, X-linked	EDA	<input type="checkbox"/> 2029	<input type="checkbox"/> 2030	<input type="checkbox"/> 2031
Ectopia lentis, isolated, autosomal dominant	FBN1	<input type="checkbox"/> 1123	<input type="checkbox"/> 1124	<input type="checkbox"/> 1530
Ectopia lentis, isolated, autosomal recessive	ADAMTSL4	<input type="checkbox"/> 1125	<input type="checkbox"/> 1126	<input type="checkbox"/> 1531
Ehlers-Danlos syndrome, classic type, 1	COL5A1	<input type="checkbox"/> 1130	<input type="checkbox"/> 1131	<input type="checkbox"/> 1533
Ehlers-Danlos syndrome, classic type 2	COL5A2	<input type="checkbox"/> 1132	<input type="checkbox"/> 1133	<input type="checkbox"/> 1534
Ehlers-Danlos syndrome, kyphoscoliotic type, 1	PLOD1	<input type="checkbox"/> 1137	<input type="checkbox"/> 2433	<input type="checkbox"/> 2434
Ehlers-Danlos syndrome, musculocontractural type, 1	CHST14	<input type="checkbox"/> 1127	<input type="checkbox"/> 1128	<input type="checkbox"/> 1532

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Disorder	Genes	Seq	Del/ Dup	Seq & Del/Dup
Ehlers-Danlos syndrome, spondylodysplastic type, 1	B4GALT7	<input type="checkbox"/> 1951	<input type="checkbox"/> 1952	<input type="checkbox"/> 1953
Ehlers-Danlos syndrome, spondylodysplastic type, 2	B3GALT6	<input type="checkbox"/> 2032	<input type="checkbox"/> 5271	<input type="checkbox"/> 5272
Ehlers-Danlos syndrome, spondylodysplastic type, 3	SLC39A13	<input type="checkbox"/> 1129	<input type="checkbox"/> 1754	<input type="checkbox"/> 1755
Ehlers-Danlos syndrome, vascular type	COL3A1	<input type="checkbox"/> 1135	<input type="checkbox"/> 1136	<input type="checkbox"/> 1537
Eiken syndrome	PTH1R	<input type="checkbox"/> 2038	<input type="checkbox"/> 2039	<input type="checkbox"/> 2040
Epidermolysis bullosa dystrophica, autosomal dominant & recessive	COL7A1	<input type="checkbox"/> 1790	<input type="checkbox"/> 1791	<input type="checkbox"/> 1792
Exostoses, multiple, types I	EXT1	<input type="checkbox"/> 1142	<input type="checkbox"/> 1143	<input type="checkbox"/> 1540
Exostoses, multiple, types II	EXT2	<input type="checkbox"/> 1144	<input type="checkbox"/> 1145	<input type="checkbox"/> 1541
Exudative vitreoretinopathy 1	FZD4	<input type="checkbox"/> 1393	<input type="checkbox"/> 1394	<input type="checkbox"/> 1544
Exudative vitreoretinopathy 2, X-linked	NDP	<input type="checkbox"/> 1395	<input type="checkbox"/> 1396	<input type="checkbox"/> 1545
Exudative vitreoretinopathy 4	LRP5	<input type="checkbox"/> 1147	<input type="checkbox"/> 1148	<input type="checkbox"/> 1546
Exudative vitreoretinopathy 5	TSPAN12	<input type="checkbox"/> 1397	<input type="checkbox"/> 1398	<input type="checkbox"/> 1547
Fabry disease	GLA	<input type="checkbox"/> 2463	<input type="checkbox"/> 2464	<input type="checkbox"/> 2465
Failure of tooth eruption, primary	PTH1R	<input type="checkbox"/> 2041	<input type="checkbox"/> 2042	<input type="checkbox"/> 2043
Fibrochondrogenesis 1	COL11A1	<input type="checkbox"/> 1149	<input type="checkbox"/> 1150	<input type="checkbox"/> 1550
Fibrochondrogenesis 2	COL11A2	<input type="checkbox"/> 1151	<input type="checkbox"/> 1152	<input type="checkbox"/> 1551
Fibrodysplasia ossificans progressiva	ACVR1	<input type="checkbox"/> 1796	<input type="checkbox"/> 1797	<input type="checkbox"/> 1798
Fibular hypoplasia and complex brachydactyly	GDF5	<input type="checkbox"/> 1857	<input type="checkbox"/> 1858	<input type="checkbox"/> 1859
Focal dermal hypoplasia	PORCN	<input type="checkbox"/> 1153	<input type="checkbox"/> 1154	<input type="checkbox"/> 1552
Frontometaphyseal dysplasia	FLNA	<input type="checkbox"/> 1155	<input type="checkbox"/> 5277	<input type="checkbox"/> 5278
Frontonasal dysplasia 1	ALX3	<input type="checkbox"/> 1921	<input type="checkbox"/> 1922	<input type="checkbox"/> 1923
Frontonasal dysplasia 2	ALX4	<input type="checkbox"/> 1924	<input type="checkbox"/> 1925	<input type="checkbox"/> 1926
Frontonasal dysplasia 3	ALX1	<input type="checkbox"/> 1927	<input type="checkbox"/> 1928	<input type="checkbox"/> 1929
Frontonasal dysplasia, SIX2 related	SIX2	<input type="checkbox"/> 2357	<input type="checkbox"/> 2358	<input type="checkbox"/> 2359
Geleophysic dysplasia 1	ADAMTSL2	<input type="checkbox"/> 1156	<input type="checkbox"/> 1157	<input type="checkbox"/> 1553
Geleophysic dysplasia 2	FBN1 Ex 41, 42	<input type="checkbox"/> 1158		
Genitopatellar syndrome	KAT6B	<input type="checkbox"/> 1799	<input type="checkbox"/> 1800	<input type="checkbox"/> 1801
Glass syndrome	SATB2	<input type="checkbox"/> 2153	<input type="checkbox"/> 2154	<input type="checkbox"/> 2155
Glomuvenous malformations	GLMN	<input type="checkbox"/> 1159	<input type="checkbox"/> 1160	<input type="checkbox"/> 1554
Gnathodiaphyseal dysplasia	ANO5	<input type="checkbox"/> 2156	<input type="checkbox"/> 2157	<input type="checkbox"/> 2158
Gracile bone dysplasia	FAM111A	<input type="checkbox"/> 1802	<input type="checkbox"/> 1803	<input type="checkbox"/> 1804
Greig cephalopolysyndactyly syndrome	GLI3	<input type="checkbox"/> 1161	<input type="checkbox"/> 1162	<input type="checkbox"/> 1555
Hajdu-Cheney syndrome	NOTCH2	<input type="checkbox"/> 1400	<input type="checkbox"/> 1401	<input type="checkbox"/> 1556
Hereditary hemorrhagic telangiectasia type 1	ENG	<input type="checkbox"/> 1404	<input type="checkbox"/> 1405	<input type="checkbox"/> 1557
Hereditary hemorrhagic telangiectasia type 2	ACVRL1	<input type="checkbox"/> 1406	<input type="checkbox"/> 1407	<input type="checkbox"/> 1558
Hereditary hemorrhagic telangiectasia type 5	GDF2	<input type="checkbox"/> 1718	<input type="checkbox"/> 1719	<input type="checkbox"/> 1720
Hereditary motor and sensory neuropathy, type IIC	TRPV4	<input type="checkbox"/> 1163	<input type="checkbox"/> 1764	<input type="checkbox"/> 1765
Homocystinuria	CBS	<input type="checkbox"/> 1164	<input type="checkbox"/> 1165	<input type="checkbox"/> 1563
Hyperostosis corticalis generalisata	SOST	<input type="checkbox"/> 1805	<input type="checkbox"/> 1806	<input type="checkbox"/> 1807
Hyperostosis corticalis generalisata, benign form of worth, with torus palatinus	LRP5	<input type="checkbox"/> 1166	<input type="checkbox"/> 1167	<input type="checkbox"/> 1564
Hyperparathyroidism, neonatal severe	CASR	<input type="checkbox"/> 1742	<input type="checkbox"/> 1743	<input type="checkbox"/> 1744
Hyperphosphatemic familial tumoral calcinosis	FGF23	<input type="checkbox"/> 2363	<input type="checkbox"/> 2364	<input type="checkbox"/> 2365
Hyperphosphatemic familial tumoral calcinosis	GALNT3	<input type="checkbox"/> 2366	<input type="checkbox"/> 2435	<input type="checkbox"/> 2436
Hypocalcemia, autosomal dominant 1	CASR	<input type="checkbox"/> 1745	<input type="checkbox"/> 1746	<input type="checkbox"/> 1747
Hypocalciuric hypercalcemia, familial, type 1	CASR	<input type="checkbox"/> 1751	<input type="checkbox"/> 1752	<input type="checkbox"/> 1753

Hypophosphatasia, infantile, childhood, or adult type	ALPL	<input type="checkbox"/> 1168	<input type="checkbox"/> 1169	<input type="checkbox"/> 1565
Inclusion body myopathy with early-onset Paget disease with or without frontotemporal dementia 1	VCP	<input type="checkbox"/> 2400	<input type="checkbox"/> 2401	<input type="checkbox"/> 2402
Insulin-like growth factor I deficiency	IGF1	<input type="checkbox"/> 2087	<input type="checkbox"/> 2088	<input type="checkbox"/> 2089
Insulin-like growth factor I, resistance to	IGF1R	<input type="checkbox"/> 2090	<input type="checkbox"/> 2091	<input type="checkbox"/> 2092
Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome	SMAD4	<input type="checkbox"/> 1410	<input type="checkbox"/> 1411	<input type="checkbox"/> 1566
KBG syndrome	ANKRD11	<input type="checkbox"/> 1966	<input type="checkbox"/> 1967	<input type="checkbox"/> 1968
Kenny-Caffey syndrome, type 1	TBCE	<input type="checkbox"/> 1903	<input type="checkbox"/> 1904	<input type="checkbox"/> 1905
Kenny-Caffey syndrome, type 2	FAM111A	<input type="checkbox"/> 1808	<input type="checkbox"/> 1809	<input type="checkbox"/> 1810
Klippel-Feil syndrome 1	GDF6	<input type="checkbox"/> 1954	<input type="checkbox"/> 1955	<input type="checkbox"/> 1956
Klippel-Feil syndrome 2	MEOX1	<input type="checkbox"/> 1957	<input type="checkbox"/> 1958	<input type="checkbox"/> 1959
Klippel-Feil syndrome 3	GDF3	<input type="checkbox"/> 1960	<input type="checkbox"/> 1961	<input type="checkbox"/> 1962
Klippel-Feil syndrome, RIPPLY2 related	RIPPLY2	<input type="checkbox"/> 2324	<input type="checkbox"/> 2348	<input type="checkbox"/> 2349
Kniest dysplasia	COL2A1	<input type="checkbox"/> 1170	<input type="checkbox"/> 1171	<input type="checkbox"/> 1567
Knobloch syndrome 1	COL18A1	<input type="checkbox"/> 1942	<input type="checkbox"/> 1943	<input type="checkbox"/> 1944
Langer mesomelic dysplasia	SHOX	<input type="checkbox"/> 2327	<input type="checkbox"/> 2328	<input type="checkbox"/> 2329
Larsen syndrome, autosomal dominant	FLNB	<input type="checkbox"/> 1172	<input type="checkbox"/> 2394	<input type="checkbox"/> 2395
Larsen syndrome, autosomal recessive	CHST3	<input type="checkbox"/> 1175	<input type="checkbox"/> 1176	<input type="checkbox"/> 1568
Lateral meningocele syndrome	NOTCH3	<input type="checkbox"/> 2096	<input type="checkbox"/> 2097	<input type="checkbox"/> 2098
Leri-Weill dyschondrosteosis	SHOX	<input type="checkbox"/> 2330	<input type="checkbox"/> 2331	<input type="checkbox"/> 2332
Loeys-Dietz syndrome 1	TGFBR1	<input type="checkbox"/> 1177	<input type="checkbox"/> 1178	<input type="checkbox"/> 1569
Loeys-Dietz syndrome 2	TGFBR2	<input type="checkbox"/> 1179	<input type="checkbox"/> 1180	<input type="checkbox"/> 1570
Loeys-Dietz syndrome 3	SMAD3	<input type="checkbox"/> 1182	<input type="checkbox"/> 1183	<input type="checkbox"/> 1573
Loeys-Dietz syndrome 4	TGFBR2	<input type="checkbox"/> 1184	<input type="checkbox"/> 1185	<input type="checkbox"/> 1574
Loeys-Dietz syndrome 5	TGFBR3	<input type="checkbox"/> 2133	<input type="checkbox"/> 2134	<input type="checkbox"/> 2135
Lysosomal acid lipase deficiency	LIPA	<input type="checkbox"/> 2403	<input type="checkbox"/> 2404	<input type="checkbox"/> 2405
Mandibulofacial dysostosis, Guion-Almeida type	EFTUD2	<input type="checkbox"/> 2159	<input type="checkbox"/> 2160	<input type="checkbox"/> 2161
Marfan syndrome, type I	FBN1	<input type="checkbox"/> 1187	<input type="checkbox"/> 1188	<input type="checkbox"/> 1577
Marfan syndrome, type II	TGFBR2	<input type="checkbox"/> 1189	<input type="checkbox"/> 1190	<input type="checkbox"/> 1578
Marshall-Smith syndrome	NFIX	<input type="checkbox"/> 1969	<input type="checkbox"/> 1970	<input type="checkbox"/> 1971
Marshall syndrome	COL11A1	<input type="checkbox"/> 1193	<input type="checkbox"/> 1194	<input type="checkbox"/> 1583
Meier-Gorlin syndrome 1	ORC1	<input type="checkbox"/> 1860	<input type="checkbox"/> 1861	<input type="checkbox"/> 1862
Meier-Gorlin syndrome 2	ORC4	<input type="checkbox"/> 1863	<input type="checkbox"/> 1864	<input type="checkbox"/> 1865
Meier-Gorlin syndrome 3	ORC6	<input type="checkbox"/> 1866	<input type="checkbox"/> 1867	<input type="checkbox"/> 1868
Meier-Gorlin syndrome 4	CDT1	<input type="checkbox"/> 1869	<input type="checkbox"/> 1870	<input type="checkbox"/> 1871
Meier-Gorlin syndrome 5	CDC6	<input type="checkbox"/> 1872	<input type="checkbox"/> 1873	<input type="checkbox"/> 1874
Melnick-Needles syndrome	FLNA	<input type="checkbox"/> 1195	<input type="checkbox"/> 5279	<input type="checkbox"/> 5280
Menkes disease	ATP7A	<input type="checkbox"/> 1196	<input type="checkbox"/> 1197	<input type="checkbox"/> 1584
Metaphyseal anadysplasia 1	MMP13	<input type="checkbox"/> 1198	<input type="checkbox"/> 1199	<input type="checkbox"/> 1585
Metaphyseal anadysplasia 2	MMP9	<input type="checkbox"/> 1200	<input type="checkbox"/> 1201	<input type="checkbox"/> 1586
Metaphyseal chondrodysplasia, Jansen type	PTH1R	<input type="checkbox"/> 2045	<input type="checkbox"/> 2046	<input type="checkbox"/> 2047
Metaphyseal chondrodysplasia, Schmid type	COL10A1	<input type="checkbox"/> 1203	<input type="checkbox"/> 2072	<input type="checkbox"/> 2073
Metaphyseal dysplasia, Spahr type	MMP13	<input type="checkbox"/> 1839	<input type="checkbox"/> 1840	<input type="checkbox"/> 1841
Metaphyseal dysplasia with maxillary hypoplasia with or without brachydactyly	RUNX2	<input type="checkbox"/> 1412	<input type="checkbox"/> 1413	<input type="checkbox"/> 1589
Metaphyseal dysplasia without hypotrichosis	RMRP	<input type="checkbox"/> 1204	<input type="checkbox"/> 1897	<input type="checkbox"/> 1898
Metatropic dysplasia	TRPV4	<input type="checkbox"/> 1205	<input type="checkbox"/> 1766	<input type="checkbox"/> 1767
Microcephaly-capillary malformation syndrome	STAMBP	<input type="checkbox"/> 2162	<input type="checkbox"/> 2163	<input type="checkbox"/> 2164
Mitral valve prolapse 2	DCHS1	<input type="checkbox"/> 2333	<input type="checkbox"/> 2334	<input type="checkbox"/> 2335
Mowat-Wilson syndrome	ZEB2	<input type="checkbox"/> 2336	<input type="checkbox"/> 2337	<input type="checkbox"/> 2338
Multiple epiphyseal dysplasia	COMP	<input type="checkbox"/> 1206	<input type="checkbox"/> 2074	<input type="checkbox"/> 2075

Disorder	Genes	Seq	Del/ Dup	Seq & Del/Dup
Multiple joint dislocations, short stature, craniofacial dysmorphism, and congenital heart defects	<i>B3GAT3</i>	<input type="checkbox"/> 2048	<input type="checkbox"/> 2049	<input type="checkbox"/> 2050
Multiple pterygium syndrome, lethal type	<i>CHRNA1</i>	<input type="checkbox"/> 2265	<input type="checkbox"/> 2266	<input type="checkbox"/> 2267
Multiple pterygium syndrome, lethal type	<i>CHRND</i>	<input type="checkbox"/> 2268	<input type="checkbox"/> 2269	<input type="checkbox"/> 2270
Multiple pterygium syndrome, lethal type & Escobar variant	<i>CHRNG</i>	<input type="checkbox"/> 2271	<input type="checkbox"/> 2272	<input type="checkbox"/> 2273
Multiple self-healing squamous epithelioma, susceptibility to	<i>TGFBR1</i>	<input type="checkbox"/> 1590	<input type="checkbox"/> 1591	<input type="checkbox"/> 1592
Myhre syndrome	<i>SMAD4</i>	<input type="checkbox"/> 1414	<input type="checkbox"/> 1415	<input type="checkbox"/> 1416
Nail-Patella syndrome	<i>LMX1B</i>	<input type="checkbox"/> 1210	<input type="checkbox"/> 5230	<input type="checkbox"/> 5231
Neu-Laxova syndrome 1	<i>PHGDH</i>	<input type="checkbox"/> 2165	<input type="checkbox"/> 2166	<input type="checkbox"/> 2167
Neu-Laxova syndrome 2	<i>PSAT1</i>	<input type="checkbox"/> 2168	<input type="checkbox"/> 2169	<input type="checkbox"/> 2170
Neurofibromatosis type I	<i>NF1</i>	<input type="checkbox"/> 2427	<input type="checkbox"/> 2428	<input type="checkbox"/> 2429
Neutropenia, severe congenital, X-linked	<i>WAS</i>	<input type="checkbox"/> 2406	<input type="checkbox"/> 2407	<input type="checkbox"/> 2408
NKX2-5 related heart malformations	<i>NKX2-5</i>	<input type="checkbox"/> 2409	<input type="checkbox"/> 2410	<input type="checkbox"/> 2411
Norrie disease	<i>NDP</i>	<input type="checkbox"/> 1417	<input type="checkbox"/> 1418	<input type="checkbox"/> 1593
Occipital horn syndrome	<i>ATP7A</i>	<input type="checkbox"/> 1211	<input type="checkbox"/> 1212	<input type="checkbox"/> 1594
Oculodentodigital dysplasia	<i>GJA1</i>	<input type="checkbox"/> 2412	<input type="checkbox"/> 2413	<input type="checkbox"/> 2414
Odontonchodermal dysplasia	<i>WNT10A</i>	<input type="checkbox"/> 1994	<input type="checkbox"/> 1995	<input type="checkbox"/> 1996
Ohdo syndrome, SBBYS variant	<i>KAT6B</i>	<input type="checkbox"/> 1811	<input type="checkbox"/> 1812	<input type="checkbox"/> 1813
Opsismodysplasia	<i>INPPL1</i>	<input type="checkbox"/> 1596	<input type="checkbox"/> 1597	<input type="checkbox"/> 1598
Orofaciodigital syndrome I	<i>OFD1</i>	<input type="checkbox"/> 2280	<input type="checkbox"/> 2281	<input type="checkbox"/> 2282
Osteoarthritis with mild chondrodysplasia	<i>COL2A1</i>	<input type="checkbox"/> 1215	<input type="checkbox"/> 1837	<input type="checkbox"/> 1838
Osteogenesis imperfecta, types I / II / III / IV	<i>COL1A1</i>	<input type="checkbox"/> 1601	<input type="checkbox"/> 1217	<input type="checkbox"/> 1602
Osteogenesis imperfecta, types I / II / III / IV	<i>COL1A2</i>	<input type="checkbox"/> 1603	<input type="checkbox"/> 1218	<input type="checkbox"/> 1604
Osteogenesis imperfecta, type V	<i>IFITM5</i>	<input type="checkbox"/> 1219	<input type="checkbox"/> 1220	<input type="checkbox"/> 1605
Osteogenesis imperfecta, type VI	<i>SERPINF1</i>	<input type="checkbox"/> 1221	<input type="checkbox"/> 1222	<input type="checkbox"/> 1606
Osteogenesis imperfecta, type VII	<i>CRTAP</i>	<input type="checkbox"/> 1223	<input type="checkbox"/> 1224	<input type="checkbox"/> 1607
Osteogenesis imperfecta, type VIII	<i>P3H1</i>	<input type="checkbox"/> 1225	<input type="checkbox"/> 1226	<input type="checkbox"/> 1608
Osteogenesis imperfecta, type IX	<i>PP1B</i>	<input type="checkbox"/> 1227	<input type="checkbox"/> 1228	<input type="checkbox"/> 1609
Osteogenesis imperfecta, type X	<i>SERPINH1</i>	<input type="checkbox"/> 1229	<input type="checkbox"/> 1230	<input type="checkbox"/> 1610
Osteogenesis imperfecta, type XI	<i>FKBP10</i>	<input type="checkbox"/> 1231	<input type="checkbox"/> 1232	<input type="checkbox"/> 1611
Osteogenesis imperfecta, type XII	<i>SP7</i>	<input type="checkbox"/> 1233	<input type="checkbox"/> 1234	<input type="checkbox"/> 1612
Osteogenesis imperfecta, type XIII	<i>BMP1</i>	<input type="checkbox"/> 1235	<input type="checkbox"/> 1236	<input type="checkbox"/> 1613
Osteogenesis imperfecta, type XVI	<i>CREB3L1</i>	<input type="checkbox"/> 1878	<input type="checkbox"/> 1879	<input type="checkbox"/> 1880
Osteogenesis imperfecta, type XVII	<i>SPARC</i>	<input type="checkbox"/> 2174	<input type="checkbox"/> 2175	<input type="checkbox"/> 2176
Osteogenesis imperfecta, PLS3 related	<i>PLS3</i>	<input type="checkbox"/> 1899	<input type="checkbox"/> 1900	<input type="checkbox"/> 1901
Osteopathia striata with cranial sclerosis	<i>AMER1</i>	<input type="checkbox"/> 1814	<input type="checkbox"/> 1815	<input type="checkbox"/> 1816
Osteopetrosis, autosomal dominant 1	<i>LRP5</i>	<input type="checkbox"/> 1238	<input type="checkbox"/> 1239	<input type="checkbox"/> 1614
Osteopetrosis, autosomal dominant 2 & autosomal recessive 4	<i>CLCN7</i>	<input type="checkbox"/> 1240	<input type="checkbox"/> 1241	<input type="checkbox"/> 1615
Osteopetrosis, autosomal recessive 1	<i>TCIRG1</i>	<input type="checkbox"/> 1242	<input type="checkbox"/> 1243	<input type="checkbox"/> 1616
Osteopetrosis, autosomal recessive 2	<i>TNFRSF11</i>	<input type="checkbox"/> 1244	<input type="checkbox"/> 1245	<input type="checkbox"/> 1617
Osteopetrosis, autosomal recessive 5	<i>OSTM1</i>	<input type="checkbox"/> 1248	<input type="checkbox"/> 1249	<input type="checkbox"/> 1619
Osteopetrosis, autosomal recessive 6	<i>PLEKHM1</i>	<input type="checkbox"/> 1250	<input type="checkbox"/> 2447	<input type="checkbox"/> 2448
Osteopetrosis, autosomal recessive 7	<i>TNFRSF11A</i>	<input type="checkbox"/> 1251	<input type="checkbox"/> 1252	<input type="checkbox"/> 1620
Osteopetrosis with renal tubular acidosis	<i>CA2</i>	<input type="checkbox"/> 1254	<input type="checkbox"/> 1255	<input type="checkbox"/> 1623
Osteoporosis-pseudoglioma syndrome	<i>LRP5</i>	<input type="checkbox"/> 1256	<input type="checkbox"/> 1257	<input type="checkbox"/> 1624
Otopalatodigital syndrome, type I/II	<i>FLNA</i>	<input type="checkbox"/> 1258	<input type="checkbox"/> 5281	<input type="checkbox"/> 5282
Otospondylomegapiphysal dysplasia	<i>COL11A2</i>	<input type="checkbox"/> 1259	<input type="checkbox"/> 1260	<input type="checkbox"/> 1625

Paget disease of bone 2	<i>TNFRSF11A</i>	<input type="checkbox"/> 2372	<input type="checkbox"/> 2373	<input type="checkbox"/> 2374
Paget disease of bone 3	<i>SQSTM1</i>	<input type="checkbox"/> 2375	<input type="checkbox"/> 2376	<input type="checkbox"/> 2377
Paget disease of bone 5	<i>TNFRSF11B</i>	<input type="checkbox"/> 2378	<input type="checkbox"/> 2379	<input type="checkbox"/> 2380
Paget disease of bone 6	<i>ZNF687</i>	<input type="checkbox"/> 2381	<input type="checkbox"/> 2382	<input type="checkbox"/> 2383
Pallister-Hall syndrome	<i>GLI3</i>	<input type="checkbox"/> 1261	<input type="checkbox"/> 1262	<input type="checkbox"/> 1626
Parastremmatic dwarfism	<i>TRPV4</i>	<input type="checkbox"/> 1263	<input type="checkbox"/> 1768	<input type="checkbox"/> 1769
Parkes Weber syndrome	<i>RASA1</i>	<input type="checkbox"/> 1264	<input type="checkbox"/> 1265	<input type="checkbox"/> 1627
Periventricular nodular heterotopia 1	<i>FLNA</i>	<input type="checkbox"/> 1267	<input type="checkbox"/> 5283	<input type="checkbox"/> 5284
Phosphoglycerate dehydrogenase deficiency	<i>PHGDH</i>	<input type="checkbox"/> 2177	<input type="checkbox"/> 2178	<input type="checkbox"/> 2179
Phosphoserine aminotransferase deficiency	<i>PSAT1</i>	<input type="checkbox"/> 2180	<input type="checkbox"/> 2181	<input type="checkbox"/> 2182
Platyspondylic lethal skeletal dysplasia, Torrance type	<i>COL2A1</i>	<input type="checkbox"/> 1268	<input type="checkbox"/> 1269	<input type="checkbox"/> 1628
	<i>COL2A1 Ex 51, 52, 53, 54</i>	<input type="checkbox"/> 1270		
Pneumothorax, primary spontaneous	<i>FLCN</i>	<input type="checkbox"/> 2283	<input type="checkbox"/> 2284	<input type="checkbox"/> 2285
Polydactyly, preaxial IV & postaxial, type A1	<i>GLI3</i>	<input type="checkbox"/> 1272	<input type="checkbox"/> 1273	<input type="checkbox"/> 1629
Popliteal pterygium syndrome	<i>IRF6</i>	<input type="checkbox"/> 2286	<input type="checkbox"/> 2287	<input type="checkbox"/> 2288
Popliteal pterygium syndrome, lethal type	<i>RIPK4</i>	<input type="checkbox"/> 2289	<input type="checkbox"/> 2290	<input type="checkbox"/> 2291
Porencephaly 1	<i>COL4A1</i>	<input type="checkbox"/> 2183	<input type="checkbox"/> 2184	<input type="checkbox"/> 2185
Porencephaly 2	<i>COL4A2</i>	<input type="checkbox"/> 2186	<input type="checkbox"/> 2187	<input type="checkbox"/> 2188
Postaxial acrofacial dysostosis	<i>DHODH</i>	<input type="checkbox"/> 2192	<input type="checkbox"/> 2193	<input type="checkbox"/> 2194
Progressive pseudorheumatoid arthropathy of childhood	<i>WISP3</i>	<input type="checkbox"/> 1276	<input type="checkbox"/> 1277	<input type="checkbox"/> 1631
Proliferative vasculopathy and hydranencephaly-hydrocephaly syndrome	<i>FLVCR2</i>	<input type="checkbox"/> 1278	<input type="checkbox"/> 1279	<input type="checkbox"/> 1632
Pseudoachondroplasia	<i>COMP</i>	<input type="checkbox"/> 1280	<input type="checkbox"/> 2076	<input type="checkbox"/> 2077
Pseudoachondroplasia-like disorder with multiple coagulation factor deficiency	<i>GGCX</i>	<input type="checkbox"/> 2101	<input type="checkbox"/> 2102	<input type="checkbox"/> 2103
Pulmonary hypertension, primary, 1	<i>BMPR2</i>	<input type="checkbox"/> 1945	<input type="checkbox"/> 1946	<input type="checkbox"/> 1947
Pulmonary hypertension, primary, 2	<i>SMAD9</i>	<input type="checkbox"/> 2107	<input type="checkbox"/> 2108	<input type="checkbox"/> 2109
Pulmonary hypertension, primary, 3	<i>CAV1</i>	<input type="checkbox"/> 2110	<input type="checkbox"/> 2111	<input type="checkbox"/> 2112
Pulmonary hypertension, primary, 4	<i>KCNK3</i>	<input type="checkbox"/> 2113	<input type="checkbox"/> 2114	<input type="checkbox"/> 2115
Pulmonary venoocclusive disease 1, autosomal dominant	<i>BMPR2</i>	<input type="checkbox"/> 1948	<input type="checkbox"/> 1949	<input type="checkbox"/> 1950
Pulmonary venoocclusive disease 2, autosomal recessive	<i>EIF2AK4</i>	<input type="checkbox"/> 2051	<input type="checkbox"/> 2052	<input type="checkbox"/> 2053
Pycnodysostosis	<i>CTSK</i>	<input type="checkbox"/> 1881	<input type="checkbox"/> 1882	<input type="checkbox"/> 1883
Rhizomelic chondrodysplasia punctata type 1	<i>PEX7</i>	<input type="checkbox"/> 1997	<input type="checkbox"/> 1998	<input type="checkbox"/> 1999
Rhizomelic chondrodysplasia punctata, type 2	<i>GNPAT</i>	<input type="checkbox"/> 2057	<input type="checkbox"/> 2058	<input type="checkbox"/> 2059
Rhizomelic chondrodysplasia punctata, type 3	<i>AGPS</i>	<input type="checkbox"/> 2060	<input type="checkbox"/> 2061	<input type="checkbox"/> 2062
Rickets, hypophosphatemic, autosomal dominant	<i>FGF23</i>	<input type="checkbox"/> 1283	<input type="checkbox"/> 2355	<input type="checkbox"/> 2356
Rickets, hypophosphatemic, autosomal recessive, 1	<i>DMP1</i>	<input type="checkbox"/> 1284	<input type="checkbox"/> 1285	<input type="checkbox"/> 1634
Rickets, hypophosphatemic, autosomal recessive, 2	<i>ENPP1</i>	<input type="checkbox"/> 1286	<input type="checkbox"/> 1287	<input type="checkbox"/> 1635
Rickets, hypophosphatemic, X-linked dominant	<i>PHEX</i>	<input type="checkbox"/> 1288	<input type="checkbox"/> 1289	<input type="checkbox"/> 1636
Rickets, vitamin D-dependent type I	<i>CYP27B1</i>	<input type="checkbox"/> 1290	<input type="checkbox"/> 1291	<input type="checkbox"/> 1637
Roberts syndrome & SC phocomelia syndrome	<i>ESCO2</i>	<input type="checkbox"/> 1884	<input type="checkbox"/> 1885	<input type="checkbox"/> 1886
Robinow syndrome, autosomal dominant 1	<i>WNT5A</i>	<input type="checkbox"/> 1817	<input type="checkbox"/> 1818	<input type="checkbox"/> 1819
Robinow syndrome, autosomal dominant 2	<i>DVL1</i>	<input type="checkbox"/> 2119	<input type="checkbox"/> 2120	<input type="checkbox"/> 2121
Robinow syndrome, autosomal recessive	<i>ROR2</i>	<input type="checkbox"/> 1823	<input type="checkbox"/> 1824	<input type="checkbox"/> 1825
Rubinstein-Taybi syndrome 1	<i>CREBBP</i>	<input type="checkbox"/> 2292	<input type="checkbox"/> 2293	<input type="checkbox"/> 2294
Rubinstein-Taybi syndrome 2	<i>EP300</i>	<input type="checkbox"/> 2295	<input type="checkbox"/> 2296	<input type="checkbox"/> 2297
Schopf-Schulz-Passarge syndrome	<i>WNT10A</i>	<input type="checkbox"/> 2000	<input type="checkbox"/> 2001	<input type="checkbox"/> 2002

LABORATORY TEST REQUISITION, PAGE 9

CTGT

Disorder	Genes	Seq	Del/ Dup	Seq & Del/Dup
Scapulo-peroneal spinal muscular atrophy	TRPV4	<input type="checkbox"/> 1292	<input type="checkbox"/> 1770	<input type="checkbox"/> 1771
Schneckenbecken dysplasia, INPPL1 related	INPPL1	<input type="checkbox"/> 2301	<input type="checkbox"/> 2302	<input type="checkbox"/> 2303
Schwartz-Jampel syndrome, type 1	HSPG2	<input type="checkbox"/> 1293	<input type="checkbox"/> 1294	<input type="checkbox"/> 1638
Sclerosteosis	SOST	<input type="checkbox"/> 1826	<input type="checkbox"/> 1827	<input type="checkbox"/> 1828
Serpentine fibula-polycystic kidney syndrome	NOTCH2	<input type="checkbox"/> 1419	<input type="checkbox"/> 1420	<input type="checkbox"/> 1639
Short-rib thoracic dysplasia 2 with or without polydactyly	IFT80	<input type="checkbox"/> 1040	<input type="checkbox"/> 1041	<input type="checkbox"/> 1481
Short-rib thoracic dysplasia 3 with or without polydactyly	DYNC2H1	<input type="checkbox"/> 1298	<input type="checkbox"/> 1299	<input type="checkbox"/> 1641
Short-rib thoracic dysplasia 4 with or without polydactyly	TTC21B	<input type="checkbox"/> 1044	<input type="checkbox"/> 1045	<input type="checkbox"/> 1483
Short-rib thoracic dysplasia 5 with or without polydactyly	WDR19	<input type="checkbox"/> 1046	<input type="checkbox"/> 1047	<input type="checkbox"/> 1484
Short-rib thoracic dysplasia 6 with or without polydactyly	NEK1	<input type="checkbox"/> 1296	<input type="checkbox"/> 1297	<input type="checkbox"/> 1640
Short-rib thoracic dysplasia 7 with or without polydactyly	WDR35	<input type="checkbox"/> 1300	<input type="checkbox"/> 1301	<input type="checkbox"/> 1642
Short-rib thoracic dysplasia 8 with or without polydactyly	WDR60	<input type="checkbox"/> 1715	<input type="checkbox"/> 1716	<input type="checkbox"/> 1717
Short stature, facial dysmorphism, and skeletal anomalies with or without cardiac anomalies	BMP2	<input type="checkbox"/> 2457	<input type="checkbox"/> 2458	<input type="checkbox"/> 2459
Short stature, idiopathic, X-linked	SHOX	<input type="checkbox"/> 2339	<input type="checkbox"/> 2340	<input type="checkbox"/> 2341
Shprintzen-Goldberg craniosynostosis syndrome	SKI	<input type="checkbox"/> 1302	<input type="checkbox"/> 1303	<input type="checkbox"/> 1643
Simpson-Golabi-Behmel syndrome, type 1	GPC3	<input type="checkbox"/> 1972	<input type="checkbox"/> 1973	<input type="checkbox"/> 1974
Simpson-Golabi-Behmel syndrome, type 2	OFD1	<input type="checkbox"/> 2304	<input type="checkbox"/> 2305	<input type="checkbox"/> 2306
Simpson-Golabi-Behmel syndrome, PIGA related	PIGA	<input type="checkbox"/> 2387	<input type="checkbox"/> 2388	<input type="checkbox"/> 2389
Skraban-Deardorff syndrome	WDR26	<input type="checkbox"/> 2460	<input type="checkbox"/> 2461	<input type="checkbox"/> 2462
Smith-Lemli-Opitz syndrome	DHCR7	<input type="checkbox"/> 2439	<input type="checkbox"/> 2443	<input type="checkbox"/> 2444
Smith-McCort dysplasia	DYM	<input type="checkbox"/> 1304	<input type="checkbox"/> 1305	<input type="checkbox"/> 1644
Snyder-Robinson mental retardation syndrome	SMS	<input type="checkbox"/> 2440	<input type="checkbox"/> 2441	<input type="checkbox"/> 2442
Sotos syndrome 1	NSD1	<input type="checkbox"/> 1975	<input type="checkbox"/> 1976	<input type="checkbox"/> 1977
Sotos syndrome 2	NFIX	<input type="checkbox"/> 1978	<input type="checkbox"/> 1979	<input type="checkbox"/> 1980
Spinal muscular atrophy, distal, congenital nonprogressive	TRPV4	<input type="checkbox"/> 1306	<input type="checkbox"/> 1772	<input type="checkbox"/> 1773
Spinal muscular atrophy, distal, X-linked 3	ATP7A	<input type="checkbox"/> 1307	<input type="checkbox"/> 1308	<input type="checkbox"/> 1645
Spondylocarpotarsal synostosis syndrome	FLNB	<input type="checkbox"/> 1309	<input type="checkbox"/> 1310	<input type="checkbox"/> 1646
Spondylocostal dysostosis 1, autosomal recessive	DLL3	<input type="checkbox"/> 1423	<input type="checkbox"/> 1424	<input type="checkbox"/> 1647
Spondylocostal dysostosis 2, autosomal recessive	MESP2	<input type="checkbox"/> 1425	<input type="checkbox"/> 1426	<input type="checkbox"/> 1648
Spondylocostal dysostosis 3, autosomal recessive	LFNG	<input type="checkbox"/> 1427	<input type="checkbox"/> 1428	<input type="checkbox"/> 1649
Spondylocostal dysostosis 4, autosomal recessive	HES7	<input type="checkbox"/> 1429	<input type="checkbox"/> 1430	<input type="checkbox"/> 1650
Spondylocostal dysostosis 5, autosomal dominant	TBX6	<input type="checkbox"/> 1778	<input type="checkbox"/> 1779	<input type="checkbox"/> 1780
Spondylocostal dysostosis 6, autosomal recessive	RIPPLY2	<input type="checkbox"/> 2342	<input type="checkbox"/> 2350	<input type="checkbox"/> 2351
Spondyloenchondrodysplasia with immune dysregulation	ACP5	<input type="checkbox"/> 1829	<input type="checkbox"/> 1830	<input type="checkbox"/> 1831
Spondyloepimetaphyseal dysplasia, Missouri type	MMP13	<input type="checkbox"/> 1311	<input type="checkbox"/> 1312	<input type="checkbox"/> 1653
Spondyloepimetaphyseal dysplasia, Strudwick type	COL2A1	<input type="checkbox"/> 1313	<input type="checkbox"/> 1314	<input type="checkbox"/> 1654
Spondylo-megaepiphyseal-metaphyseal dysplasia	NKX3-2	<input type="checkbox"/> 1890	<input type="checkbox"/> 1891	<input type="checkbox"/> 1892
Spondyloepimetaphyseal dysplasia with joint laxity type 2	KIF22	<input type="checkbox"/> 1432	<input type="checkbox"/> 1433	<input type="checkbox"/> 1655

Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures	B3GAL76	<input type="checkbox"/> 2066	<input type="checkbox"/> 5263	<input type="checkbox"/> 5264
Spondyloepiphyseal dysplasia congenita	COL2A1	<input type="checkbox"/> 1315	<input type="checkbox"/> 1316	<input type="checkbox"/> 1656
Spondyloepiphyseal dysplasia, Maroteaux type	TRPV4	<input type="checkbox"/> 1317	<input type="checkbox"/> 1774	<input type="checkbox"/> 1775
Spondyloepiphyseal dysplasia tarda, X-linked	TRAPPC2	<input type="checkbox"/> 1318	<input type="checkbox"/> 1319	<input type="checkbox"/> 1657
Spondyloepiphyseal dysplasia with congenital joint dislocations	CHST3	<input type="checkbox"/> 1320	<input type="checkbox"/> 1321	<input type="checkbox"/> 1658
Spondylometaphyseal dysplasia, Kozlowski type	TRPV4	<input type="checkbox"/> 1322	<input type="checkbox"/> 1776	<input type="checkbox"/> 1777
Spondylometaphyseal dysplasia with cone-rod dystrophy	PCYT1A	<input type="checkbox"/> 1832	<input type="checkbox"/> 1833	<input type="checkbox"/> 1834
Spondyloocular syndrome	XYLT2	<input type="checkbox"/> 2195	<input type="checkbox"/> 2196	<input type="checkbox"/> 2197
Spondyloperipheral dysplasia	COL2A1	<input type="checkbox"/> 1323	<input type="checkbox"/> 1324	<input type="checkbox"/> 1659
	COL2A1 Ex 51, 52, 53, 54	<input type="checkbox"/> 1325		
Steel syndrome	COL27A1	<input type="checkbox"/> 2430	<input type="checkbox"/> 2431	<input type="checkbox"/> 2432
Stickler syndrome, type I	COL2A1	<input type="checkbox"/> 1331	<input type="checkbox"/> 1332	<input type="checkbox"/> 1662
Stickler syndrome, type II	COL11A1	<input type="checkbox"/> 1333	<input type="checkbox"/> 1334	<input type="checkbox"/> 1663
Stickler syndrome, type III	COL11A2	<input type="checkbox"/> 1335	<input type="checkbox"/> 1336	<input type="checkbox"/> 1664
Stiff skin syndrome	FBN1	<input type="checkbox"/> 1442	<input type="checkbox"/> 2445	<input type="checkbox"/> 2446
Stuve-Wiedemann syndrome	LIFR	<input type="checkbox"/> 1338	<input type="checkbox"/> 1339	<input type="checkbox"/> 1667
Supravalvular aortic stenosis	ELN	<input type="checkbox"/> 1668	<input type="checkbox"/> 1669	<input type="checkbox"/> 1670
Terminal osseous dysplasia	FLNA	<input type="checkbox"/> 2207	<input type="checkbox"/> 5285	<input type="checkbox"/> 5286
	FGFR3	<input type="checkbox"/> 1340	<input type="checkbox"/> 1758	<input type="checkbox"/> 1759
Thanatophoric dysplasia, type I / II	FGFR3	<input type="checkbox"/> 1341		
	FGFR3 Ex 7,10,15,19			
Three M syndrome 1	CUL7	<input type="checkbox"/> 1343	<input type="checkbox"/> 1344	<input type="checkbox"/> 1671
Three M syndrome 2	OBSL1	<input type="checkbox"/> 1345	<input type="checkbox"/> 1346	<input type="checkbox"/> 1672
Three M syndrome 3	CCDC8	<input type="checkbox"/> 1347	<input type="checkbox"/> 1348	<input type="checkbox"/> 1673
Thrombocytopenia 1	WAS	<input type="checkbox"/> 2415	<input type="checkbox"/> 2416	<input type="checkbox"/> 2417
Thrombocytopenia-absent radius syndrome	RBM8A	<input type="checkbox"/> 1349	<input type="checkbox"/> 1350	<input type="checkbox"/> 1674
Tooth agenesis, selective, 1	MSX1	<input type="checkbox"/> 1351	<input type="checkbox"/> 1352	<input type="checkbox"/> 1675
Tooth agenesis, selective, 4	WNT10A	<input type="checkbox"/> 2003	<input type="checkbox"/> 2004	<input type="checkbox"/> 2005
Torg-Winchester syndrome	MMP2	<input type="checkbox"/> 1353	<input type="checkbox"/> 1354	<input type="checkbox"/> 1676
Treacher Collins syndrome 1	TCOF1	<input type="checkbox"/> 1355	<input type="checkbox"/> 1356	<input type="checkbox"/> 1677
Treacher Collins syndrome 2	POLR1D	<input type="checkbox"/> 1357	<input type="checkbox"/> 1358	<input type="checkbox"/> 1678
Treacher Collins syndrome 3	POLR1C	<input type="checkbox"/> 1359	<input type="checkbox"/> 1360	<input type="checkbox"/> 1679
Trichodontoosseus syndrome	DLX3	<input type="checkbox"/> 1915	<input type="checkbox"/> 1916	<input type="checkbox"/> 1917
Trichorhinophalangeal syndrome, type I / III	TRPS1	<input type="checkbox"/> 1361	<input type="checkbox"/> 1362	<input type="checkbox"/> 1680
Van Buchem disease, type 2	LRP5	<input type="checkbox"/> 1369	<input type="checkbox"/> 1370	<input type="checkbox"/> 1683
Van der Woude syndrome 1	IRF6	<input type="checkbox"/> 2307	<input type="checkbox"/> 2308	<input type="checkbox"/> 2309
Van der Woude syndrome 2	GRHL3	<input type="checkbox"/> 2310	<input type="checkbox"/> 2311	<input type="checkbox"/> 2312
Van Maldergem syndrome 1	DCHS1	<input type="checkbox"/> 2345	<input type="checkbox"/> 2346	<input type="checkbox"/> 2347
Wagner vitreoretinopathy	VCAN	<input type="checkbox"/> 1371	<input type="checkbox"/> 1372	<input type="checkbox"/> 1684
Weaver syndrome	EZH2	<input type="checkbox"/> 1981	<input type="checkbox"/> 1982	<input type="checkbox"/> 1983
Weill-Marchesani syndrome 1	ADAMTS10	<input type="checkbox"/> 1375	<input type="checkbox"/> 1376	<input type="checkbox"/> 1686
Weill-Marchesani syndrome 2	FBN1	<input type="checkbox"/> 1373	<input type="checkbox"/> 1374	<input type="checkbox"/> 1685
Weill-Marchesani syndrome 3	LTBP2	<input type="checkbox"/> 1724	<input type="checkbox"/> 1725	<input type="checkbox"/> 1726
Weill-Marchesani-like syndrome	ADAMTS17	<input type="checkbox"/> 1727	<input type="checkbox"/> 1728	<input type="checkbox"/> 1729
Weissenbacher-Zweymuller syndrome	COL11A2	<input type="checkbox"/> 1377	<input type="checkbox"/> 1378	<input type="checkbox"/> 1687
Wilson disease	ATP7B	<input type="checkbox"/> 1381	<input type="checkbox"/> 1382	<input type="checkbox"/> 1689
Wiskott-Aldrich syndrome	WAS	<input type="checkbox"/> 2418	<input type="checkbox"/> 2419	<input type="checkbox"/> 2420
Witkop syndrome	MSX1	<input type="checkbox"/> 1383	<input type="checkbox"/> 1384	<input type="checkbox"/> 1690
Wolcott-Rallison syndrome	EIF2AK3	<input type="checkbox"/> 1385	<input type="checkbox"/> 1386	<input type="checkbox"/> 1691
Wolman disease	LIPA	<input type="checkbox"/> 2421	<input type="checkbox"/> 2422	<input type="checkbox"/> 2423
Zimmermann-Laband syndrome 1	KCNH1	<input type="checkbox"/> 2198	<input type="checkbox"/> 2199	<input type="checkbox"/> 2200
Zimmermann-Laband syndrome 2	ATP6V1B2	<input type="checkbox"/> 2201	<input type="checkbox"/> 2202	<input type="checkbox"/> 2203

TARGETED VARIANT TESTING

Patient Name: _____	Patient DOB: _____
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Prenatal testing: <input type="checkbox"/> Yes <input type="checkbox"/> No Anticipated delivery date: _____ <input type="checkbox"/> Female <input type="checkbox"/> Male
<input type="checkbox"/> 1389: Maternal cell contamination testing (<i>recommended for all prenatal testing</i>)
Has karyotyping been performed? <input type="checkbox"/> Yes <input type="checkbox"/> No _____

TARGETED VARIANT TESTING	FAMILY HISTORY																									
Individual to be tested: <input type="checkbox"/> Affected/Symptomatic <input type="checkbox"/> Unaffected/Asymptomatic	Is there a family history of disease for which the patient is being tested? <input type="checkbox"/> Yes <input type="checkbox"/> No																									
Proband Name: _____ Proband DOB: _____ Proband Case #: _____ Relationship to Proband: _____ <input type="checkbox"/> 1387 One Mutation <input type="checkbox"/> 2027 Three Mutations <input type="checkbox"/> 1388 Two Mutations <input type="checkbox"/> 2044 CNV Mutation Deletion/Duplication Analysis Gene/Mutation: _____	<table border="1" style="width:100%; border-collapse: collapse; text-align: center;"> <thead> <tr> <th style="width:20%;">Relationship to the individual being tested</th> <th style="width:10%;">Maternal</th> <th style="width:10%;">Paternal</th> <th style="width:10%;">DX</th> <th style="width:10%;">Age at DX</th> </tr> </thead> <tbody> <tr><td> </td><td><input type="checkbox"/></td><td><input type="checkbox"/></td><td> </td><td> </td></tr> <tr><td> </td><td><input type="checkbox"/></td><td><input type="checkbox"/></td><td> </td><td> </td></tr> <tr><td> </td><td><input type="checkbox"/></td><td><input type="checkbox"/></td><td> </td><td> </td></tr> <tr><td> </td><td><input type="checkbox"/></td><td><input type="checkbox"/></td><td> </td><td> </td></tr> </tbody> </table>	Relationship to the individual being tested	Maternal	Paternal	DX	Age at DX		<input type="checkbox"/>	<input type="checkbox"/>				<input type="checkbox"/>	<input type="checkbox"/>				<input type="checkbox"/>	<input type="checkbox"/>				<input type="checkbox"/>	<input type="checkbox"/>		
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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.
CTGT is not responsible for obtaining or submitting blood, tissue or other samples from patients for testing, nor is it responsible for communicating test results to patients. CTGT, therefore, assumes no liability for: Any injury or illness incurred from obtaining a blood sample, biopsy or other specimen. Mislabeling or misidentification of submitted samples. Incorrect or incomplete information provided in test requisition forms. Information provided to CTGT outside of its requisition forms or order process. Any loss incurred as a result of communicating the outcome of any genetic tests to patients or their representatives. CTGT further disclaims any liability for incorrect or incomplete information in any written or verbal communication other than test results reports prepared by CTGT, including, but not limited to, its web site, brochures, technical information, emails, letters or telephone conversations. IN NO EVENT SHALL CTGT BE LIABLE FOR ANY INCIDENTAL, INDIRECT, CONSEQUENTIAL OR PUNITIVE DAMAGES ARISING OUT OF ANY USE OF OR INABILITY TO USE CTGT'S SERVICES, WHETHER BASED ON WARRANTY, CONTRACT, TORT OR ANY OTHER LEGAL THEORY, REGARDLESS OF WHETHER CTGT IS ADVISED OF THE POSSIBILITY OF SUCH DAMAGES.