CLINICAL GENETIC SERVICE

Laboratory User Guides

July 2018

DEPARTMENT OF HEALTH
GOVERNMENT OF HONG KONG SAR

Laboratory Address: 2/F., 2 Kwong Lee Road, Shum Shui Po, Kowloon, Hong Kong SAR, China
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Compared with the previous version of April 2018:

No amendment

Compared with the previous version of January 2018:

1. TAT for NGS-based test is re-defined;
2. Gene list for some larger NGS panels is attached as Appendix A for reference;
3. Additional 2 months for TAT of *DMPK* Southern blot testing.

Last updated on 16th June 2018

The Clinical Genetic Service (CGS) of the Department of Health in Hong Kong is a government-funded, tertiary referral center that provides clinical, laboratory, counseling services related to genetic disorders. The function of the Genetic Laboratory is to back up Genetic Counseling Clinic within this Service. Presently the Laboratory accepts specimens only via the Genetic Counseling Clinic of the CGS. For more information about the laboratory service, please call

Enquire Telephone : (852) 2725 4144
Fax : (852) 2729 1440
Email : so_cg@dh.gov.hk

Specimen Submission Information

1. Clinical Specimens

Cytogenetic testing: 3 ml **heparin** blood
1 to 2 ml more for additional FISH study

Molecular testing: 3 ml **EDTA** blood

Samples need to be identified accurately and patient should not be fasted for overnight. The container of specimen shall be labeled with patient's name (in capital letters) and the HKID/Travel document number (2 unique identifiers).

Each specimen must be accompanied with a requisition form. Please fill patient name in capital letters and the HKID/Travel document number on the requisition form, which **SHALL** also include the followings:

- Date of birth and sex
- Date of specimen taken
- Date of request
- Type of specimen
- Name and signature of physician requesting test
- Name and institution of referring doctor
- Type of test requested
- Pedigree (if needed)

Remarks:
Blood will be rejected if clotted, hemolyzed, quantity insufficient for Cytogenetic testing or delivery time was more than 7 days after collection.

2. Delivering Specimens

After blood taken, specimen should be delivered at the same day (before 5 PM) as soon as possible

- **Deliver at room temperature;**
- **Store at 4°C if unable to deliver at the same day. Never ice or freeze the blood;**
- **In a plastic bag separately with the Requisition Form.**

To: 2/F, Laboratory, Cheung Sha Wan Jockey Club Clinic
2 Kwong Lee Road, Sham Shui Po
Kowloon, Hong Kong
Tel: 2708 7112 (attention: Mr. Leung)

For operational need, it is advised to take blood and send on Monday or Tuesday or Friday for Cytogenetic testing.
For DEB fragility testing, it is advised to take blood and send on Monday or Tuesday with the control sample. Whenever possible, the control should be appropriately matched with the characteristics of the test sample, such as sex, age, cigarette smoking and undercurrent illness.

Further re-arrangement is necessary for the blood taken and delivery during or before public holidays.

3. Turnaround Time (TAT)

For routine service cases:	Cytogenetic testing:	28 calendar days
	FISH & array CGH:	2 months
	Molecular testing:	2-6 months (refer to the table below)
For urgent cases:	Cytogenetic testing:	8 calendar days
	Molecular testing:	14 calendar days
		For prenatal and urgent NGS-based testing, please consult our on call clinical geneticist.

Cytogenetics Service

Conventional Cytogenetic Investigations for Blood		
G banding		
DEB fragility study		
C staining		
Molecular Cytogenetics (FISH & Array CGH) Investigations		
Array CGH (Agilent CGX array 8x60K)		
Telomere probes and whole chromosome painting probes		
Microdeletion probes	**Investigation**	**Locus Involved**
	Cri du Chat syndrome	5p15.2-15.3
	DiGeorge syndrome	22q11.2
	Kallman syndrome	Xp22.3
	Miller Dieker syndrome	17p13.3
	Prader Willi / Angelman syndrome	15q11-13
	Retinoblatoma (Rb)	13q14
	Smith Magenis syndrome	17p11.2
	Steroid sulphatase deficiency	Xp22.3
	Williams syndrome	7q11.23
	Wolf-Hirschhorn syndrome	4p16.3
	SRY	Yp11.3

Molecular Service

Test	OMIM ID	Gene/Locus Involved	Investigation	TAT
Next Generation Sequencing and MLPA Targeted Panels				
Alport syndrome panel	-	***COL4A3, COL4A5***	**point mutation**	**4 months#**
Common lethal skeletal disease panel	-	***TP63, GLI3, COL1A2, COL1A1, TRIP11, COL2A1, FGFR3, ALPL***	**point mutation**	**4 months#**
Cardiomyopathy panel	-	**59 genes refer Appendix A**	**point mutation**	**4 months#**
Extended Hereditary cancer syndrome panel	-	**143 genes refer Appendix A**	**point mutation**	**4 months#**
Hereditary cancer syndrome panel (Breast and colorectal cancer)	-	***BRCA1, BRCA2, PTEN, TP53, MLH1,MSH2, MSH6, PMS2, EPCAM, APC, MUTYH***	**point mutation** **deletion / duplication for *BRCA1* and *BRCA2* only**	**4 months#**
DMD panel	-	***DMD***	**point mutation** **deletion / duplication**	**4 months#**
Hemophilia A panel	-	***F8***	**point mutation** **deletion / duplication**	**4 months#**
Kabuki syndrome panel	-	***KMT2D, KDM6A***	**point mutation** **deletion / duplication**	**4 months#**
Marfan and aortopathy panel	-	***FBN1, TGFBR1, TGFBR2, SMAD3, SLC2A10, MYLK, MYH11, COL3A1, ACTA2***	**point mutation** **deletion / duplication for *FBN1* and *TGFBR2* only**	**4 months#**
NF panel	-	***NF1, NF2***	**point mutation** **deletion / duplication**	**4 months#**

Test	OMIM ID	Gene/Locus Involved	Investigation	TAT
Polycystic kidney diseases panel	-	***PDK1, PKD2, PKHD1***	**point mutation deletion / duplication for *PKD1* and *PKD2* only**	**4 months#**
Rasopathy panel	-	***PTPN11, BRAF, CBL, HRAS, KRAS, NRAS, MAP2K1&2, RAF1, RIT1, SHOC2, SOS1, SPRED1, RASA1***	**point mutation**	**4 months#**
Tuberous sclerosis panel	-	***TSC1, TSC2***	**point mutation** **deletion / duplication**	**4 months#**
Chromatin related disease panel	-	**23 genes refer Appendix A**	**point mutation**	**4 months#**
X-linked intellectual disability		**91 genes refer Appendix A**	**point mutation**	**4 months#**
Skeletal dysplasia panel	-	**177 genes refer Appendix A**	**point mutation**	**4 months#**
Genetic myopathy panel	-	**83 genes refer Appendix A**	**point mutation**	**4 months#**
Genetic neuropathy panel		**50 genes refer Appendix A**	**point mutation**	**4 months#**
Hereditary Spastic Paraplegia panel	-	**40 genes refer Appendix A**	**point mutation**	**4 months#**
Ciliopathy panel		**74 genes refer Appendix A**	**point mutation**	**4 months#**
Inherited Arrhythmia disease panel	-	**30 genes refer Appendix A**	**point mutation**	**4 months#**
Genetic Endocrine disease panel		**167 genes refer Appendix A**	**point mutation**	**4 months#**
Eye disease panel	-	**73 genes refer Appendix A**	**point mutation**	**4 months#**

Test	OMIM ID	Gene/Locus Involved	Investigation	TAT
Hearing impairment panel		**94 genes refer Appendix A**	**point mutation**	**4 months#**
Epilepsy panel	-	**58 genes refer Appendix A**	**point mutation**	**4 months#**
Neurogenetic disease panel		**111 genes refer Appendix A**	**point mutation**	**4 months#**
# TAT for above NGS panel testing is around 4 months, but additional 1-2 months is needed if further confirmation testing is necessary. **# For those tests involving both sequencing and deletion/duplication, they are treated as 2 separate tests and are not tested in a reflex manner. Separate requests are required.**				
CHROMOSOMAL ABNORMALITIES				
Intellectual Disability (ID) related microdeletion syndromes	-	11 loci causing ID syndromes	deletion	**2 months**
Subtelomeric deletion/duplication	-	Telomeres	deletion / duplication	**2 months**
Aneuploidies	-	chromosomes 13, 18, 21, X, Y	trisomy 13, 18, 21	**2 months**
CRANIOSYNOSTOTIC SYNDROME				
Antley-Bixler syndrome	201750	*POR*	point mutation	4 months
Apert syndrome **Crouzon Syndrome** **Pfeiffer syndrome**	101200 123500 101600	*FGFR2*	point mutation	4 months
Saethre-Chotzen syndrome	101400	*TWIST*	deletion	4 months
EYE DISEASES				
Aniridia	106210	*PAX6*	point mutation / deletion	4 months#

Test	OMIM ID	Gene/Locus Involved	Investigation	TAT
Cone-Rod dystrophy, type 2	120970	*CRX*	point mutation	4 months
Corneal dystrophy, lattice type	122200	*TGFBI*	point mutation	4 months
Congenital fibrosis of the extraocular muscles, type 1	135700	*KIF21A*	point mutation	4 months
Norrie disease	310600	*NDP*	point mutation	4 months
Peters plus syndrome	261540	*B3GALTL*	point mutation	4 months
***SOX2*-related eye disorder**	184429	*SOX2*	point mutation	4 months
HEARING LOSS				
Branchio-oto-renal syndrome, type 1	113650	*EYA1*	Deletion / duplication	4 months
Deafness, congenital, with inner ear agenesis, microtia, and microdontia	610706	*FGF3*	point mutation	4 months
Non-syndromic deafness	220290	*GJB2 / GJB6*	point mutation / deletion	**2 months**
Non-syndromic deafness	500008	Mito - *RNR1*	m.1555A>G	**2 months**
Waardenburg syndrome, type 1	193500	*PAX3*	point mutation / deletion	4 months#
HEART DISEASE				
Non-syndromic congenital heart disease	108900	*NKX2-5*	point mutation	4 months
HEMATOLOGY				
α-Thalassemia	604131	α-globin region	Southeast Asia type deletion / rightward or leftward deletion	4 months
			point mutation	4 months

Test	OMIM ID	Gene/Locus Involved	Investigation	TAT
Hemophilia A	306700	*F8*	Introns 1 & 22 inversion	4 months
Hemophilia A	306700	*F8*	point mutation	4 months
Hemophilia B	306900	*F9*	point mutation / deletion	4 months#
INBORN ERRORS of METABOLISM				
Fabry disease	301500	*GLA*	point mutation	4 months
Hypothyroidism, athyroidal, with spiky hair and cleft palate	241850	*FOXE1*	point mutation / deletion	4 months#
Hypothyroidism, choreoathetosis, neonatal respiratory distress	610978	*NKX2-1*	point mutation / deletion	4 months#
Hypothyroidism, congenital, nongoitrous, type 1	275200	*TSHR*	point mutation / deletion	4 months#
Hypothyroidism, congenital, nongoitrous, type 2	218700	*PAX8*	point mutation / deletion	4 months#
Infantile systemic hyalinosis	228600	*ANTXR2*	point mutation	4 months
Leigh syndrome	256000	*SURF1*	point mutation	4 months
Mitochondrial disorder panel **MELAS (m.3243A>G)** **MERRF (m.8344A>G)** **NARP (m.8993T>G)** **LHON (m.3460G>A, m.11778G>A, m.14484T>C)**	 540000 545000 551500 535000	Mitochondrion	point mutation	**2 months**
Mowat-Wilson syndrome	235730	*ZEB2*	point mutation / deletion	4 months#
Mucolipidosis II α/β **Mucolipidosis III α/β**	252500 252600	*GNPTAB*	point mutation	4 months

Test	OMIM ID	Gene/Locus Involved	Investigation	TAT
Mucopolysaccharidosis type II	309900	*IDS*	point mutation	4 months
Pyruvate dehydrogenase E1-α deficiency	312170	*PDHA1*	point mutation / deletion	4 months#
Wilson disease	277900	*ATP7B*	point mutation / deletion	**2 months#**
MALIGNANCY				
Multiple endocrine neoplasia type 2A **Multiple endocrine neoplasia type 2B** **Familial medullary thyroid carcinoma**	171400	*RET*	point mutation / deletion	4 months#
Peutz-Jeghers syndrome	175200	*STK11*	point mutation	4 months
Retinoblastoma	180200	*RB1*	point mutation / deletion	4 months#
Schwannoma, vestibular nerve tumor	162091	*SMARCB1*	point mutation / deletion	4 months#
Von Hippel-Lindau syndrome	193300	*VHL*	point mutation / deletion	**2 months#**
NEUROLOGY				
Amyotrophic lateral sclerosis	205250	*SOD1*	point mutation	4 months
Charcot-Marrie-Tooth disease, 1A	118220	*PMP22*	Gene duplication	**2 months**
Hereditary Neuropathy with liability pressure palsies	162500	*PMP22*	Gene deletion	
Charcot-Marrie-Tooth disease, 1B	118200	*MPZ*	point mutation	4 months
Charcot-Marrie-Tooth disease, X-linked, 1	302800	*GJB1*	point mutation	4 months
Congenital central hypoventilation syndrome	209880	*PHOX2B*	point mutation / polyalanine expansion	4 months
Dystonia, AD	128100	*DYT1*	GAG deletion	4 months

Test	OMIM ID	Gene/Locus Involved	Investigation	TAT
Fragile X syndrome	300624	*FMR1*	CGG expansion	**2 months**
Fragile X syndrome, type E	309548	*AFF2*	GCC expansion	4 months
Friedreich ataxia	229300	*FXN*	GAA expansion	**2 months**
Huntington disease	143100	*HTT*	CAG expansion	**2 months**
Pelizaeus-Merzbacher disease	312080	*PLP1*	gene duplication	4 months
Spastic paraplegia 2, X-linked	312920		point mutation	4 months
Spastic paraplegia 44, AR	613206	*GJC2*	point mutation	4 months
Spinocerebellar ataxias panel:				
SCA1	164400	*ATXN1*	CAG expansion	**2 months**
SCA2	183090	*ATXN2*	CAG expansion	
SCA3	109150	*ATXN3*	CAG expansion	
SCA6	183086	*CACNA1A*	CAG expansion	
SCA7	164500	*ATXN7*	CAG expansion	
SCA8	608768	*ATXN80S*	CTG expansion	
SCA12	604326	*PPP2R2B*	CAG expansion	
SCA17	607136	*TBP*	CAG expansion	
Dentatorubral-pallidoluysian atrophy	125370	*ATN1*	CAG expansion	
Subcortical band heterotopias, X-linked	300067	*DCX*	point mutation / deletion	4 months
NEURO-MUSCULAR DISEASES				
Duchenne muscular dystrophy	310200	*DMD*	Exon(s) deletion / duplication	**2 months**
			point mutation	4 months
Kennedy's disease	313200	*AR*	CAG expansion	4 months

Test	OMIM ID	Gene/Locus Involved	Investigation	TAT
Myotonic dystrophy, type 1 (PCR)	160900	*DMPK*	CTG expansion	**2 months additional 2 months is need if further Southern blot testing is needed**
Myotonic dystrophy (Southern blot)				
Myotonic dystrophy, type 2	602668	*CNBP*	CCTG expansion	4 months
Oculopharyngeal muscular dystrophy	164300	*PABPN1*	GCG insertion	4 months
Spinal muscular atrophy	253300	*SMN1*	Exons 7&8 deletion	**2 months**
RENAL DISEASES				
Alport syndrome, AD	301050	*COL4A5*	point mutation	4 months
Alport syndrome, AR	203780	*COL4A3*	point mutation	4 months
Nephrogenic diabetes insipidus	304800	*AVPR2*	point mutation / deletion	4 months#
Polycystic kidney disease, AD	173900 613095	*PKD1 & PKD2*	point mutation / deletion	4 months#
Polycystic kidney disease, AR	263200	*ARPKD*	point mutation	4 months
DERMATOLOGIC DISORDER				
Ectodermal dysplasia 1, hypohidrotic, X-linked	305100	*EDA*	point mutation / deletion	4 months#
Epidermolysis bullosa dystrophica, AR	226600	*COL7A1*	point mutation	4 months
Incontinentia pigmenti	308300	*NEMO*	exons 6-12 deletion	4 months
			point mutation	4 months
SKELETAL DYSPLASIA				

Test	OMIM ID	Gene/Locus Involved	Investigation	TAT
Achondrogenesis, type Ib **Diatrophic dysplasia** **Epiphyseal dysplasia, multiple, 4**	600972 222600 226900	*SLC26A2*	point mutation	4 months
Achondroplasia	100800	*FGFR3*	c.1138G>A	4 months
Atelosteogenesis type 1 and type 3 **Larsen syndrome**	108720 108721	*FLNB*	point mutation	4 months
Brachydactyly type B1	113000	*ROR2*	point mutation	4 months
Campomelic dysplasia	114290	*SOX9*	point mutation	4 months
Chondrodysplasia with joint dislocations, GRAPP type	614078	*IMPAD1*	Point mutation	4 months
Cleidocranial dysplasia	119600	*RUNX2*	point mutation / deletion	4 months#
Desbuquois dysplasia	251450	*CANT1*	point mutation	4 months
Fibrodysplasia Ossificans Progressiva	135100	*ACVR1*	point mutation	4 months
Hypochondroplasia	146000	*FGFR3*	c.1620C>G or C>A	4 months
Hypophosphatatemic rickets, AD	193100	*FGF23*	point mutation	4 months
Hypophosphatatemic rickets, X-linked	307800	*PHEX*	point mutation	4 months
Leri-Weill dyschondrosteosis	127300	*SHOX*	gene deletion	4 months
Langer mesomelic dysplasia	249700		point mutation	4 months
Metaphyseal chondrodysplasia, McKusick type	157660	*RMRP*	point mutation	4 months
Multiple exotoses	133700	*EXT1*	point mutation / deletion	4 months#

Test	OMIM ID	Gene/Locus Involved	Investigation	TAT
Nail-patella syndrome	161200	*LMX1B*	point mutation / deletion	4 months#
Pseudoachondroplasia	177170	*COMP*	GAC expansion	4 months
Pseudohypoparathyroidism	174800 603233	*GNAS*	point mutation copy number & methylation	4 months 4 months
***TP63*-related disorder**	-	*TP63*	point mutation	4 months
Thanatophoric Dysplasia I	187600	*FGFR3*	point mutation	4 months
Schwartz-Jampel syndrome type 2	601559	*LIFR*	point mutation	4 months
SEX DISORDER				
46XY Sex reversal **46XX Sex reversal**	400044 400045	*SRY*	The presence of *SRY* point mutation	**2 months** 4 months
SYNDROMES / DYSMORPHOLOGY				
Angelman syndrome	105830	*UBE3A*	point mutation	4 months
Beckwith-Wiedemann syndrome **Russell-Silver syndrome**	130650	H19DMR & KvDMR domains *CDKN1C*	copy number & methylation point mutation	4 months 4 months
Blepharophimosis-Ptosis-Epicanhus- Inversus syndrome (BPES)	110100	*FOXL2*	point mutation / deletion	4 months#
Costello syndrome	218040	*HRAS*	point mutation	4 months
Craniofrontonasal syndrome	304110	*EFNB1*	point mutation / deletion	4 months#

Test	OMIM ID	Gene/Locus Involved	Investigation	TAT
Spondylocostal dysostosis 2, AR	608681	*MESP2*	point mutation	4 months
Spondyloepiphyseal dysplasia with congenital joint dislocation	143095	*CHST3*	point mutation	4 months
Li-Fraumeni syndrome	151623	*TP53*	point mutation / deletion	4 months#
LOWE syndrome	309000	*OCRL*	point mutation	4 months
Noonan syndrome	163950	*PTPN11*	point mutation	4 months
Prader Willi syndrome (PWS) / Angelman syndrome (AS)	176270 105830	PWS / AS critical region at 15q11-q13	Microdeletion / Uniparental disomy at 15q11-q13	**2 months**
Rett syndrome	312750	*MECP2*	point mutation / deletion	4 months#
Simpson-Golabi-Behmel syndrome, type 1	312870	*GPC3*	point mutation / deletion	4 months#
Sotos syndrome	117550	*NSD1*	point mutation / deletion	4 months#
Van der Woude syndrome **Popliteal pterygium syndrome**	119300	*IRF6*	point mutation	4 months
Variant Rett syndrome	613454	*FOXG1*	point mutation	4 months
Wolfram syndrome	222300	*WSF1*	point mutation	4 months
Miscellaneous				
Abnormal X-inactivation pattern	-	*AR*	Abnormal X-inactivation	4 months
Alveolar capillary dysplasia	265380	*FOXF1*	point mutation	4 months
Central diabetes insipidus	125700	*AVP*	point mutation	4 months

End of the Laboratory User Guides
Next version will be available in January 2019

Appendix A : Gene list of NGS panel

Updated on 15/6/2018

Cardio Myopathy 59 genes	Cancer Predisposition Syndrome 143 genes	Chromatin related disease 23 genes	X-Linked MR 91 genes	Skeletal dysplasia 177 genes	Genetic Myopathy 83 genes	Genetic Neuropathy 50 genes	Hereditary Spastic Paraplegia 40 genes	Ciliopathy 74 genes	Inherited Arrhythmia 30 genes	Genetic Endocrine disease 167 genes	Eye disease pnael 73 genes	Hearing impairment panel 94 genes	Epilepsy panel 58 genes	Neurogenetic disease panel 111 genes
ABCC9	AIP	CHD7	ACSL4,	ACAN	ACTA1	AARS	AFG3L2	ACVR2B	AKAP9	ABCC6	AGK	ACTG1	ADSL	ABCB7
ACTC1	AKT1	RSK2 (RPS6KA3)	AFF2,	ACP5	AGRN	ATL1	ALS2	AHI1	ANK2	ABCC8	B3GLCT(B3GALTL)	ATP2B2	ALDH7A1	ACVRL1/ALK1
ACTN2	ALK	ADNP	AP1S2,	AGPS	ANO5 (LGMD2L)	ATP7A	AP5Z1	ANKS6	CACNA1C	ABCC8	BCOR	BDP1	ALG13	AFG3L2
BAG3	ANTXR1	ESCO2	ARHGEF9,	ALPL	BIN1	BSCL2	ATL1	ARL13B	CACNB2	*AGPAT2*	BCOR	CABP2	ARFGEF2	AFG3L2
CALR3	ANTXR2	SMARCA2	ARX,	ANKH	CAPN3 (LGMD2A)	DNAJB2	B4GALNT1	ARL6	CASQ2	*AGPAT2*	BFSP1	CCDC50	ARGGEF15	ALS2
CAV3	APC	SOX11	ATP6AP2,	ANO5	CAV3 (LGMD1C)	DNM2	BSCL2	B9D1	CAV3	AGPS	BFSP2	CEACAM16	ARHGEF9	ANG
COX15	ASCC1	ANKRD11	ATP7A,	ARHGAP31	CCDC78	DNMT1	C12orf65	B9D2	DSC2	AKR1C2	BMP4	CLDN14	ARX	ANO3
CRYAB	ATM	HDAC8	ATRX,	ARSE	CFL2	DYNC1H1	CYP27A1	BBS1	DSG2	AP2S1	BMP4	COCH	ATP1A3	APP
CSRP3	ATR	SMARCA4	BCOR,	ATP6V0A2	CHAT	EGR2	CYP2U1	BBS10	DSP	AR	CHMP4B	COL11A2	ATRX	APTX
DES	AXIN2	TBC1D24	BRWD3,	B3GALT6	CHKB	FAM134B	CYP7B1	BBS12	GPD1L	ARSE	COL4A1	COL4A6	CDKL5	ATM
DMD	BAP1	ARID1A	CASK,	B4GALT7	CHRNA1	FGD4	DDHD1	BBS2	HCN4	ARX	COL4A1	CRYM	CHD2	ATP1A2
DSC2	BARD1	NIPBL	CCDC22,	BMP1	CHRNB1	FIG4	DDHD2	BBS4	JUP	ATRX	CRYAA	DFNA5	CHRNA2	ATP1A2
DSG2	BLM	SMARCB1	CDK16,	BMP2	CHRND	GAN	FA2H	BBS5	KCNE1	B3GALTL	CRYAB	DFNB31	CHRNA4	ATP1A3
DSP	BMPR1A	WNT5A	CDKL5,	BMPR1B	CHRNE	GARS	FIG4	BBS7	KCNE2	BCOR	CRYBA1	DFNB59	CHRNB2	ATP1A3

Cardio Myopathy 59 genes	Cancer Predisposition Syndrome 143 genes	Chromatin related disease 23 genes	X-Linked MR 91 genes	Skeletal dysplasia 177 genes	Genetic Myopathy 83 genes	Genetic Neuropathy 50 genes	Hereditary Spastic Paraplegia 40 genes	Ciliopathy 74 genes	Inherited Arrhythmia 30 genes	Genetic Endocrine disease 167 genes	Eye disease pnael 73 genes	Hearing impairment panel 94 genes	Epilepsy panel 58 genes	Neurogenetic disease panel 111 genes
DTNA	BRCA1	ARID1B	CLIC2,	CA2	CNTN1	GDAP1	GBA2	BBS9	KCNE3	BMP4	CRYBA2	DIABLO	CLCN4	ATP7B
EMD	BRCA2	PHF6	CNKSR2,	CANT1	COL6A1	GJB1	HSPD1	C5orf42	KCNH2 (HERG)	BSCL2	CRYBA4	DIAPH1	CNTNAP2	ATP7B
EYA4	BRIP1	SMARCE1	CUL4B,	CASR	COL6A2	GLA	KIAA0196	CC2D2A	KCNJ2	BSCL2	CRYBB1	DIAPH3	EEF1A2	BMP9/GDF2
FKTN	BUB1B	CREBBP	DCX,	CC2D2A	COL6A3	HSPB1	KIF1A	CCDC103	KCNJ5	*CASR*	CRYBB2	ESPN	EFHC1	BMPR2
GAA	CD96	RAD21	DKC1,	CDH3	COLQ	HSPB8	KIF5A	CCDC114	KCNJ8	CASR	CRYBB3	ESRRB	EHMT	C10orf2
GATAD1	CDC73	SMC1A	DLG3,	CDKN1C	DAG1 (LGMD2P)	IGHMBP2	L1CAM	CCDC39	KCNQ1	CASR	CRYGB	EYA4	FOXG1	CACNA1A
GLA	CDH1	EP300	DMD,	CEP290	DES (LGMD1D)	IKBKAP	MTPAP	CCDC40	NKX2.5	CAV1	CRYGC	GIPC3	GABRB3	CACNA1A
JUP	CDK4	ROR2	FANCB,	CHST14	DNAJB6 (LGMD1E)	KIF1A	NIPA1	CEP164	PKP2	CAV1	CRYGD	GJB2	GNAO1	CACNA1A
LAMA4	CDKN1B	SMC3	FGD1,	CHST3	DNM2	LITAF	PLP1	CEP290	RANGRF (MOG1)	CDC73	CRYGS	GJB3	GRIN2A	CACNB4
LAMP2	CDKN2A		FLNA,	CHSY1	DOK7	LMNA	PSEN1	CEP41	RYR2	CDKN1A	CTDP1	GJB6	GRIN2B	CACNB4
LDB3	CHEK1		FMR1,	CLCN5	DPAGT1	LRSAM1	REEP1	CFAP53	SCN1B	CDKN1B	CYP1B1	GRHL2	KCNH5	CAV1
LMNA	CHEK2		FRMPD4,	CLCN7	DPM2	MED25	RTN2	CFTR	SCN3B	CDKN1C	CYP1B1	GRXCR1	KCNQ2	CCM1/KRIT1
MYBPC3	CYLD		FTSJ1,	COL10A1	DPM3	MFN2	SACS	DNAAF1	SCN4B	CDKN2B	EPHA2	HGF	KCNT1	CCM2
MYH6	CYP21A2		GDI1,	COL11A1	DYSF (LGMD2B)	MPZ	SIGMAR1	DNAAF2	SCN5A	CDKN2C	EYA1	ILDR1	KIAA1279	CCM3/PDCD10
MYH7	DDB2		GK,	COL11A2	EMD	MTMR2	SLC16A2	DNAAF3	SNTA1	CEL	FOXC1	KARS	LGI1	CYP27A1
MYL2	DICER1		GPC3,	COL1A1	FHL1	NDRG1	SLC2A1	DNAAF5	TMEM43	CEP41	FOXC1	KCNQ4	MAGI2	CYP27A1

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MYL3	DIS3L2		GRIA3,	COL1A2	FKRP (LGMD2I, FRKP)	NEFL	SPAST	DNAH11		CHD7	FOXE3	LHFPL5	MBD5	CYP27A1
MYLK2	DKC1		HCCS,	COL2A1	FKTN (LGMD2M)	NGF	SPG11	DNAH5		CREBBP	FOXE3	LOXHD1	MECP2	EEF2
MYOZ2	EHBP1		HCFC1,	COL9A1	FKTN	NTRK1	SPG20	DNAI1		CUL3	FOXE3	LRTOMT	MEF2C	ENG
MYPN	EPCAM		HPRT1,	COL9A2	FLNC	PDK3	SPG21	DNAI2		CYB5A	FRAS1	MARVELD2	NRXN1	FA2H
NEXN	EPHB2		HSD17B10,	COL9A3	GAA	PLEKHG5	SPG7	DNAL1		CYP11A1	FRAS1	MIR96	PCDH19	FGF14
PKP2	ERCC2		HUWE1,	COMP	GFPT1	PMP22	VAMP1	FOXH1		CYP17A1	FREM1	MSRB3	PLCB1	FIG4
PLN	ERCC3		IDS,	CREB3L1	GMPPB	PRPS1	VPS37A	GDF1		CYP19A1	FREM1	MYH14	PNKP	FTL
PRKAG2	ERCC4		IGBP1,	CRTAP	GTDC2 (POMGNT2)	PRX	WDR45	GLIS2		CYP21A2	FREM2	MYH9	PNPO	FTL
PSEN1	ERCC5		IL1RAPL1,	CTSK	HNRNPDL	RAB7A				CYP11B1	FTL	MYO15A	POLG	FUS
PSEN2	ERCC6		IQSEC2,	CUL7	ISPD	REEP1 (C2ORF23)	ZFYVE26	INPP5E		DHCR24	FYCO1	MYO3A,	PRRT2	FXN
RBM20	ESCO2		KDM5C,	DDR2	ITGA7	SBF2	ZFYVE27	INVS		DHCR7	GALK1	MYO6	SCN1A	GBA
RYR2	EXT1		KIAA2022,	DHCR24	KBTB	SCN9A		IQCB1		DHH	GALT	OTOA	SCN2A	GBA
SCN5A	EXT2		KLF8,	DLL3	KBTBD13	SH3TC2		KIF7		DMP1	GCNT2	OTOF	SCN8A	GBA2
SCO2	FAH		L1CAM,	DLX3	LAMA2	SLC12A6		LEFTY2		DNMT3B	GJA3	OTOG	SIP1 [ZEB2]	GBA2
SDHA	FANCA		LAMP2,	DMP1	LARGE	SPTLC1		LRRC6		DUOX2	GJA8	PNPT1	SLC16A2	GCH1
SGCD	FANCB		MAOA,	DYM	LIMS2	SPTLC2		MKKS		DYNC2H1	GRIP1	POU3F4	SLC25A22	GLMN

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SLC25A4	FANCC		MBTPS2,	DYNC2H1	LMNA (LGMD1B)	TFG		MKS1		EBP	HCCS	POU4F3	SLC2A1 [GLUT1]	GNAL
TAZ	FANCD2		MECP2,	EBP	LMOD3	TRPV4		NEK8		ENNP1 (not found, discarded)	HSF4	PRPS1	SLC9A6	IFRD1
TBX20	FANCE		MED12,	EIF2AK3	MTM1	TTR		NKX2-5		*ENPP1*	LEPREL1 (P3H2)	PTPRQ	SPTAN1	ITPR1
TCAP	FANCF		MID1,	ENPP1	MUSK	WNK1		NME8		ESCO2	LIM2	P2RX2	ST3GAL3	KCNA1
TGFB3	FANCG		NAA10,	ESCO2	MYF6			NODAL		FAM58A	MAF	RDX	ST3GAL5	KCNA1
TMEM43	FANCI		NDP,	EVC	MYH7			NPHP1		FAT4	MFRP	RPGR	STXBP1	KCNC3
TMPO	FANCL		NDUFA1,	EVC2	MYOT			NPHP3		FEZF1	MIP	SERPINB6	SYNGAP1	KCND3
TNNC1	FANCM		NHS,	EXT1	NEB			NPHP4		FGF23	NDP	SLC17A8	SZT2	KCNK3
TNNI3	FH		NLGN3,	EXT2	PLEC (LGMD2Q)			OFD1		FGF23	NHS	SLC26A5	TBC1D24	LRRK2
TNNT2	FLCN		NLGN4X,	FAM20C	PLEC1			RPGR		FIG4	OTX2	SLC4A11	TCF4	MAPT
TPM1	GALNT12		NSDHL,	FBLN1	POMGNT1 (LGMD2O)			RPGRIP1L		FOXE1	PAX6	SMPX	UBE2A	MAPT
TTN	GATA2		OCRL,	FBN1	POMGnT1			RSPH4A		FRAS1	PAX6	STRC	UBE3A	MTPAP
VCL	GLI3		OFD1,	FBXW4	POMGNT2 (GTDC2)			RSPH9		FREM2	PITX2	TECTA		NOTCH3
	GLMN		OPHN1,	FERMT3,	POMK			SDCCAG8		GALNT3	PITX3	TJP2		OPTN
	GPC3		OTC,	FGF10,	POMT1 (LGMD2K)			TCTN1		GATA4	PITX3	TMC1		PANK2
	HFE		PAK3,	FGF23,	POMT2 (LGMD2N)			TCTN2		GATA6	PXDN	TMIE		PARK2

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	HRAS		PCDH19,	FGFR1,	PTPLA			TCTN3		GCK	SIL1	TMPRSS3		PARK7
	KDR		PDHA1,	FGFR2,	RAPSN			TMEM138		GCK	SLC16A12	TPRN		PDYN
	KIF1B		PGK1,	FGFR3,	RYR1			TMEM216		GCM2	SMOC1	TRIOBP		PFN1
	KIT		PHF6,	FKBP10,	SCN4A			TMEM231		GLUD1	SMOC1	EYA1		PINK1
	KLHDC8B		PHF8,	FLNA,	SEPN1			TMEM237		GNA11	SOX2	SIX1		PNKD
	LIG4		PLP1,	FLNB,	SGCA (LGMD2D)			TMEM67		GNA11	STRA6	SIX5		PRKCG
	LYST		PORCN,	FMN1,	SGCB (LGMD2E)			TRIM32		GNPAT	TDRD7	KCNJ10		PRKCG
	MAX		PQBP1,	GALNT3,	SGCD (LGMD2F)			TTC21B		GRIP1	VAX1	SLC26A4		PRRT2
	MC1R		PRPS1,	GDF5,	SGCG (LGMD2C)			TTC8		HADH	VIM	GPSM2		PRRT2
	MEN1		PTCHD1,	GLI3,	SYNE1			WDPCP		HCCS	VSX2	LARS2		PRRT2
	MET		RAB39B,	GNAS,	SYNE2			WDR19		HESX1	WFS1	HSD17B4		PRRT2
	MITF		RBM10,	GORAB,	TCAP (LGMD2G)			ZIC3		HNF1A		HARS2		PSEN1
	MLH1		RPL10,	GPC6,	TMEM43			ZNF423		HNF1B		CLPP		PSEN2
	MLH3		RPS6KA3,	GREM1,	TMEM5					HNF4A		PAX3		PTEN
	MRE11A		SHROOM4,	HDAC4,	TNNT1					HNF4A		SOX10		RASA1
	MSH2		SLC16A2,	HES7,	TNPO3 (LGMD1F)					HOXA13		MITF		SACS

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	MSH6		SLC9A6,	HOXD13,	TPM2					HSD17B3		SNAI2		SCN1A
	MSMB		SMC1A,	HPGD,	TPM3					HSD3B2		KIT		SCN1A
	MSR1		SMS,	HSPG2,	TRAPPC11					ICK		EDNRB		SETX
	MTAP		SOX3,	ICK,	TRIM32 (LGMD2H)					IL17RD		EDN3		SETX
	MUTYH		SYN1,	IFITM5,	TTN (LGMD2J)					INS		WFS1		SGCE
	NBN		SYP,	IFT122,						INSR		CDH23		SIGMAR1
	NDUFA13		TIMM8A,	IFT140,						INSR		CIB2		SIL1
	NF1		TSPAN7,	IFT80,						IRF6		CLRN1		SLC16A2
	NF2		UBE2A,	IHH,						KAL1		GPR98		SLC16A2
	NTRK1		UPF3B,	KIF22,						KCNJ11		HARS		SLC1A3
	PALB2		ZDHHC15,	KIF7,						KCNJ11		MYO7A		SLC1A3
	PALLD		ZDHHC9,	LEMD3,						KISS1R		PCDH15		SLC2A1
	PDE11A		ZNF711	LFNG,						KL		PDZD7		SLC2A1
	PDGFRA			LIFR,						KLHL3		USH1C		SNCA
	PIK3CA			LMBR1,						LHCGR		USH1G		SOD1
	PMS2			LMNA,						LHX3		USH2A		SPG11

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	POLD1			LRP4,						LHX4				SPG11
	POLE			LRP5,						LMNA				SPG20
	POLH			MAFB,						LMNA				SPG21
	POU6F2			MATN3,						MAP3K1				SPG7
	PRKAR1A			MESP2,						MC2R				SPR
	PTCH1			MGP,						MCM4				SPTBN2
	PTCH2			MKS1,						*MEN1*				TARDBP
	PTEN			MMP13,						MKKS				TGM6
	RAD50			MMP2,						MKS1				TH
	RAD51B			MMP9,						MRAP				THAP1
	RAD51C			MYCN,						NEK1				TTBK2
	RAD51D			NEK1,						NEUROD1				TTPA
	RB1			NIPBL,						NKX2-1				UBQLN2
	RECQL4			NKX3-2,						NNT				VAMP1
	RET			NOG,						NR0B1				VAPB
	RHBDF2			NOTCH2,						NR5A1				VCP

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	RNASEL RSPO1 RTEL1 RUNX1 SBDS SDHA SDHAF2 SDHB SDHC SDHD SLX4 SMAD4 SMARCA4 SMARCB1 STK11 SUFU			NPR2, OBSL1, OSTM1, P3H1, PAPSS2, PCNT, PHEX, PIGV, PITX1, PLOD2, PLS3 PPIB, PRKAR1A, PTH1R, PTHLH, PTPN11,						NSMF OPHN1 PAX6 *PAX8* PDX1 PEX7 PHEX POR POU1F1 PPARG PPARG PPARG PROP1 PTH PTPN11 PTRF				WDR45

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	TERT TGFBR1 TINF2 TMC6 TMC8 TMEM127 TP53 TSC1 TSC2 UROD VHL WAS WRN WT1 XPA XPC			PYCR1, RASGRP2, RECQL4, ROR2, RPGRIP1L, RUNX2, SALL1, SALL4, SERPINF1, SERPINH1, SH3PXD2B, SHH, SHOX, SLC26A2, SLC34A3, SLC35D1,						PTRF RET RIPK4 ROR2 RSPO1 SALL1 SAMD9 SCARF2 SEMA3A SETBP1 SLC16A1 SLC34A3 SOX9 SPECC1L SRD5A2 SRY				

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	XRCC3			SLC39A13, SMARCAL1, SOST, SOX9, SP7, SULF1, TBCE, TBX15, TBX3, TBX5, TBX6, TBXAS1, TCIRG1, TCTN3, TGFB1, THPO,						STAR STAR TBX15 TCTN3 TG THRA TPO TRMT10A TSHR TSPYL1 UBR1 WDR60 *WNK1* WNK4 WNT4 WNT7A				

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				TMEM216, TMEM38B TMEM67, TNFRSF11A, TNFRSF11B, TNFSF11, TP63, TREM2, TRIP11, TRPS1, TRPV4, TYROBP, WDR35, WISP3, WNT1 WNT3,						WT1 ZFP57 ZFPM2 GHR STAT5B IGF1 IGFALS IGF1R GATA3 HRPT2				

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				WNT5A, WNT7A, ZMPSTE24										