CLINICAL GENETIC SERVICE

Laboratory User Guides

Jan 2019

DEPARTMENT OF HEALTH
GOVERNMENT OF HONG KONG SAR

Laboratory Address: 2/F., 2 Kwong Lee Road, Shum Shui Po, Kowloon, Hong Kong SAR, China
Tel: (852) 2725-4144 Fax: (852) 2729-1440 Email: so_cg@dh.gov.hk

We comply with the Personal Data (Privacy) Ordinance of Hong Kong and applicable laws and regulations to protect personal data privacy.

To complaint our service, please write or contact to Laboratory Director, Genetic Laboratory with the information at the footer.

Compared with the previous version of July 2018:

A new test for *C9ORF72* gene

Last updated on 27 December 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 4ºC 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	-	**58 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
Kennedy's disease	313200	*AR*	CAG expansion	4 months
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

Test	OMIM ID	Gene/Locus Involved	Investigation	TAT
SKELETAL DYSPLASIA				
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

Test	OMIM ID	Gene/Locus Involved	Investigation	TAT
Multiple exotoses	133700	*EXT1*	point mutation / deletion	4 months#
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

Test	OMIM ID	Gene/Locus Involved	Investigation	TAT
Craniofrontonasal syndrome	304110	*EFNB1*	point mutation / deletion	4 months#
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 July 2019

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