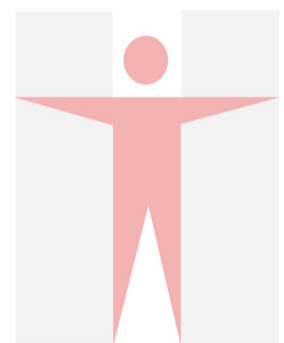


## **CLINICAL GENETIC SERVICE**

# **Laboratory User Guides**

**(Website version)**

**July 2011**



**DEPARTMENT OF HEALTH  
GOVERNMENT OF HONG KONG SAR**

**The Clinical Genetic Service of the Department of Health in Hong Kong is a government-funded, tertiary referral centre 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 Clinical Genetic Service. For more information about the laboratory service, please call**

**Enquire Telephone : (852) 2725 4144**

**Fax : (852) 2729 1440**

**Email : [so\\_cg@dh.gov.hk](mailto: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

Patients need be identified positively and not be fasted. 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 or quantity insufficient for Cytogenetic testing

### **2. Delivering Specimens**

After blood taken, specimen should be delivered at the same day (before 5 PM) as far 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. Lai)

For operational need, it is advice to take blood and send on Monday or Tuesday or Friday for Cytogenetic testing. For DEB fragility testing, it is advice to take blood and send on Monday or Tuesday with the control sample. Where possible, the control should be appropriately matched with 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: 30 calendar days  
FISH: 2 months  
Molecular testing: 3-4 months (refer to the table below)

**For urgent cases:** Cytogenetic testing: 8 calendar days  
Molecular testing: 7 calendar days for prenatal testing (mutation(s) must has been identified) and MLPA for trisomy  
14 calendar days for mutational screening testing (amplicons < 10)  
28 calendar days for Southern based testing

## Cytogenetics Service

Conventional Cytogenetic Investigations for Blood		
<b>G banding</b>		
<b>DEB fragility study</b>		
<b>C staining</b>		
Molecular Cytogenetics (FISH) Investigations		
<b>Telomere probes</b>		
<b>Whole chromosome painting probes</b>		
<b>Microdeletion probes</b>	<b>Investigation</b>	<b>Locus Involved</b>
	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
	Retinoblastoma (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

### ACCREDITED TEST

Test	Gene/Locus Involved	Investigation	TAT
Charcot-Marrie-Tooth, 1A	<i>PMP22</i>	Gene duplication	3 months
Duchenne muscular dystrophy	<i>DMD</i>	Exon(s) deletion/ duplication	3 months
Fragile X syndrome (PCR + Southern)	<i>FMR1</i>	CGG expansion	3 months
Friedreich ataxia	<i>FXN</i>	GAA expansion	3 months
Huntington's disease	<i>HTT</i>	CAG expansion	3 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)	<i>Mitochondrion</i>	Point mutation	3 months
Myotonic dystrophy (PCR + Southern)	<i>DMPK</i>	CTG expansion	3 months
Prader Willi syndrome (PWS) / Angelman syndrome (AS)	PWS / AS critical region at chromosome 15	Microdeletion / Uniparental disomy (UPD) at these regions	3 months
Spinal muscular atrophy	<i>SMN1</i>	Exon deletion	3 months
Spinocerebellar ataxias panel: SCA1 SCA2 SCA3 SCA6 SCA7 SCA8 SCA12 Dentatorubral-pallidoluysian atrophy	<i>ATXN1</i> <i>ATXN2</i> <i>ATXN3</i> <i>CACNA1A</i> <i>ATXN7</i> <i>ATXN80S</i> <i>PPP2R2B</i> <i>ATNI</i>	CAG expansion CAG expansion CAG expansion CAG expansion CAG expansion CTG expansion CAG expansion CAG expansion	3 months

## Molecular Service

### NON-ACCREDITED TEST

Test	Gene/Locus Involved	Investigation	TAT
<b>CHROMOSOMAL ABNORMALITIES</b>			
Mental Retard (MR) related microdeletion syndromes	multiple loci causing MR syndromes	deletion	4 months
Subtelomeric deletion/duplication	Telomeres	deletion / duplication	4 months
Trisomies	chromosomes 13, 18, 21	trisomy 13, 18, 21	4 months
<b>CRANIOSYNOSTOTIC SYNDROME</b>			
Antley-Bixler syndrome	<i>POR</i>	point mutation	4 months
Apert syndrome Crouzon Syndrome Pfeiffer syndrome	<i>FGFR2</i>	point mutation	4 months
Saethre-Chotzen syndrome	<i>Twist</i>	deletion	4 months
<b>EYE DISEASES</b>			
Cone-Rod dystrophy	<i>CRX</i>	point mutation	4 months
Corneal dystrophy (lattice type)	<i>BIGH3</i>	point mutation	4 months
Congenital fibrosis of the extraocular muscles type 1 <sup>New</sup>	<i>KIF21A</i>	point mutation	4 months
<b>HEARING LOSS</b>			
Non-syndromic deafness	<i>GJB2 / GJB6</i>	point mutation / deletion	4 months
	<i>Mitochondrion</i>	mtDNA 1555A>G point mutation	4 months
Waardenburg syndrome type 1	<i>PAX3</i>	point mutation / deletion	4 months
<b>HEMATOLOGY</b>			
$\alpha$ -Thalassemia	$\alpha$ -globin region	Southeast Asia type deletion/ rightward/ leftward deletion	4 months
		point mutation	4 months
Hemophilia A	<i>F8</i>	Introns 1 & 22 inversion	4 months
		linkage analysis*	4 months
<b>INBORN ERRORS of METABOLISM</b>			
Bartter syndrome	<i>BSND</i>	point mutation	4 months
Leigh disease	<i>SURF1</i>	point mutation	4 months
Mowat-Wilson syndrome	<i>ZFX1B</i>	point mutation	4 months

Test	Gene/Locus Involved	Investigation	TAT
Mucopolysaccharidosis <sup>New</sup>	<i>GNPTAB</i>	point mutation	4 months
Pyruvate dehydrogenase deficiency	<i>PDHA1</i>	point mutation	4 months
Wilson disease	<i>ATP7B</i>	point mutation	4 months
<b>NEUROLOGY</b>			
Charcot-Marrie-Tooth, 1B	<i>MPZ</i>	point mutation	4 months
Charcot-Marrie-Tooth, X-linked, 1	<i>GJB1</i>	point mutation	4 months
Congenital central hypoventilation syndrome	<i>CCHS</i>	point mutation / polyalanine expansion	4 months
Dystonia, type 1	<i>DYT1</i>	GAG deletion	4 months
Fragile X syndrome, type E	<i>FMR2</i>	GCC expansion	4 months
Pelizaeus Merzbacher disease Spastic paraplegia 2, X-linked	<i>PLP1</i>	gene duplication	4 months
Pelizaeus Merzbacher disease	<i>PLP1</i>	point mutation	4 months
Pelizaeus-Merzbacher-like disease	<i>GJA12</i>	point mutation	4 months
Subcortical band heterotopia	<i>DCX</i>	point mutation	4 months
<b>NEURO-MUSCULAR DISEASES</b>			
Duchenne muscular dystrophy	<i>DMD</i>	linkage analysis**	4 months
Kennedy's disease	<i>AR</i>	CAG expansion	4 months
Oculopharyngeal muscular dystrophy	<i>PABP2</i>	GCG insertion	4 months
<b>RENAL DISEASES</b>			
Alport syndrome, AD type	<i>COL4A5</i>	linkage analysis**	4 months
Alport syndrome, AR type <sup>New</sup>	<i>COL4A3</i>	point mutation	4 months
Polycystic kidney disease, adult	<i>PKD1 &amp; PKD2</i>	linkage analysis**	4 months
Polycystic kidney disease, AR type	<i>ARPKD</i>	linkage analysis**	4 months
<b>DERMATOLOGIC DISORDER</b>			
Ectodermal dysplasia	<i>EDA</i>	point mutation	4 months
Incontinentia pigmenti	<i>NEMO</i>	exons deletion point mutation	4 months 4 months
Epidermolysis bullosa dystrophica <sup>New</sup>	<i>COL7A1</i>	point mutation	4 months
<b>SKELETAL DYSPLASIA</b>			
Achondrogenesis, type Ib Diastrophic dysplasia Epiphyseal dysplasia, multiple, 4	<i>DTDST</i>	point mutation	4 months
Achondroplasia	<i>FGFR3</i>	c.1138G>A	4 months
Albright hereditary osteodystrophy / Pseudohypoparathyroidism	<i>GNAS1</i>	point mutation	4 months
Brachydactyly type B	<i>ROR2</i>	point mutation	4 months
Campomelic dysplasia	<i>SOX9</i>	point mutation	4 months
Fibrodysplasia Ossificans Progressiva	<i>ACVR1</i>	point mutation	4 months
Hypochondroplasia	<i>FGFR3</i>	c.1620C>G or C>A	4 months

Test	Gene/Locus Involved	Investigation	TAT
Hypophosphatemic ricket, AD type	<i>FGF23</i>	point mutation	4 months
Hypophosphatemic ricket, X-linked	<i>PHEX</i>	point mutation	4 months
Leri-Weill syndrome	<i>SHOX</i>	gene deletion	4 months
		point mutation	4 months
Metaphyseal chondrodysplasia (McKusick type)	<i>RMRP</i>	point mutation	4 months
Pseudoachondroplasia	<i>COMP</i>	GAC expansion	4 months
Thanatophoric Dysplasia	<i>FGFR3</i>	point mutation	4 months
<b>SEX DISORDER</b>			
Sex determination	<i>SRY</i>	deletion	4 months
		point mutation	4 months
<b>SYNDROMES / DYSMORPHOLOGY</b>			
Angelman syndrome	<i>UBE3A</i>	point mutation	4 months
Beckwith-Wiedemann / Russell-Silver syndrome	H19DMR & KvDMR domains	copy number & methylation	4 months
	<i>CDKN1C</i>	point mutation	4 months
Blepharophimosis-Ptosis-Epicanthus-Inversus syndrome (BPES)	<i>FOXL2</i>	point mutation / deletion	4 months
Costello syndrome	<i>HRAS</i>	point mutation	4 months
Craniofrontonasal syndrome	<i>EFNB1</i>	point mutation	4 months
Jarcho-Levin syndrome	<i>MESP2</i>	point mutation	4 months
Larsen syndrome	<i>CHST3</i>	point mutation	4 months
Li Fraumeni syndrome <sup>New</sup>	<i>TP53</i>	point mutation	4 months
LOWE syndrome	<i>OCRL1</i>	point mutation	4 months
Noonan syndrome	<i>PTPN11</i>	point mutation	4 months
Rett syndrome	<i>MECP2</i>	point mutation / deletion	4 months
Sotos syndrome	<i>NSD1</i>	point mutation / deletion	4 months
Van der Woude syndrome	<i>IRF6</i>	point mutation	4 months
Wolfram syndrome	<i>WSF1</i>	point mutation	4 months
<b>Miscellaneous</b>			
Tumor suppressor	<i>VHL</i>	point mutation	4 months
X-inactivation pattern	<i>AR</i>	Abnormal X-inactivation	4 months
<p>** Sufficient numbers of family member, especially the index patient must present before the start of a linkage analysis, judged by the Clinical Molecular Geneticist.</p>			

In addition, several mutational screenings are provided upon request. TAT is variable.  
Call **(852) 2725 4144** for details.

Test	Gene Involved	Investigation
Citrullinemia disease type 2	<i>SLC25A13</i>	point mutation
Conradi-Humermann syndrome	<i>EBP</i>	point mutation
Duchenne muscular dystrophy	<i>DMD</i>	point mutation
Gaucher disease	<i>GCB</i>	point mutation
Glutaric aciduria type 1	<i>GCDH</i>	point mutation
Greig syndrome	<i>GLI 3</i>	point mutation
Marfan syndrome	<i>Fibrillin</i>	point mutation
Neurofibromatosis type 1	<i>NF1</i>	point mutation / deletion
Senior-Loken syndrome 5	<i>IQCB1</i>	point mutation
Smith-Megenis syndrome	<i>RAI1</i>	point mutation
Spondylocostal dysostosis (Jarcho-Levin syndrome)	<i>DLL3</i>	point mutation
Triple A syndrome	<i>GL003</i>	point mutation

\* NF1: Next batch results are available at the end of June 2012. Last day to receive specimen for this batch reporting is the end of March, 2012.

\* DMD: Next batch results are available at the end of June 2013. Last day to receive specimen for this batch reporting is the end of March, 2013.

\* Marfan syndrome: Next batch results are available at the end of June 2014. Last day to receive specimen for this batch reporting is the end of March, 2014.

## ***End of the Laboratory User Guide***

**Next version will be available in January 2012**