

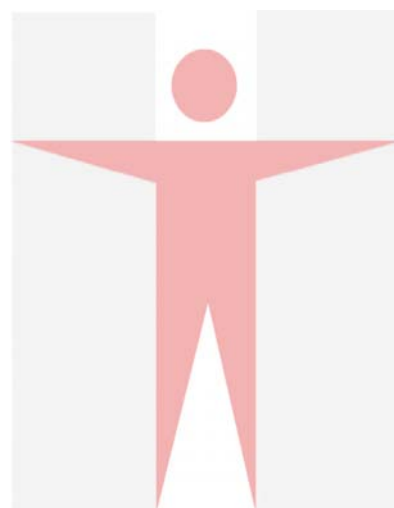


**CLINICAL GENETIC SERVICE**

# **LABORATORY GENETIC INVESTIGATIONS**

**December 2006**

DEPARTMENT OF HEALTH  
HONG KONG SAR GOVERNMENT



## **Specimen Submission Information**

### **1. Clinical Specimens**

Cytogenetic testing: 3 ml of heparin blood

Molecular testing: 3 ml of EDTA blood

Each specimen must be accompanied by a requisition form (attached), which should include the followings:

Name and ID number unique on specimen container

Date of birth or age and sex

Date of collection

Type of specimen

Name of physician requesting test

Type of test requested

Date for next follow up

Indication for testing

Pedigree (if needed)

### **2. Send Specimen/Requisition at Room Temperature to**

2/F., Cheung Sha Wan Jockey Club Clinic

2 Kwong Lee Road, Sham Shui Po

Kowloon, Hong Kong

Tel: 2725 4144 (attention: Mr. Tong)

### **3. Turnaround Time (Urgent cases)**

Cytogenetic testing: 5 working days

Molecular testing: 5 working days for PCR based testing

4 weeks for Southern based testing

For multi-exons (>15) gene testing: enquire to medical staff

### **4. Trace of Results**

Contact medical officer on call at 9726 1332.

## Cytogenetic Service

### A. Conventional Cytogenetic Investigations for Blood, Bone Marrow, Amniotic Cells, Chorionic Villi, Skin, Tumours and Tissues

1. G banding and metaphase studies
2. Prometaphase study
3. DEB fragility study
4. Sister chromatid exchange study
5. C staining
6. G11 staining
7. Late replicating X study
8. Other special staining
9. Tissue cultures and karyotyping
10. Leukemia studies

### B. Molecular Cytogenetics (FISH) Investigations

#### Microdeletion Diseases Detected by FISH Probes for Specific Chromosome Locations

- |                                   |             |
|-----------------------------------|-------------|
| 11. Wolf-Hirschhorn syndrome      | 4p16.3      |
| 12. Cri du Chat syndrome          | 5p15.2-15.3 |
| 13. Williams syndrome             | 7q11.23     |
| 14. Prader Willi syndrome         | 15q11-13    |
| 15. Angelman syndrome             | 15q11-13    |
| 16. DiGeorge syndrome             | 22q11.2     |
| 17. Miller Dieker syndrome        | 17p13.3     |
| 18. Smith Magenis syndrome        | 17p11.2     |
| 19. Steroid sulphatase deficiency | Xp22.3      |
| 20. Kallmann syndrome             | Xp22.3      |

#### Multiple Congenital Anomalies / Mental Retardation and Marker Chromosomes

- |  |         |
|--|---------|
| 21. $\alpha$ Satellite probes for autosomes              | 1 to 22 |
| 22. $\alpha$ Satellite probes for sex chromosomes        | X and Y |
| 23. Whole chromosome painting probes for autosomes       | 1 to 22 |
| 24. Whole chromosome painting probes for sex chromosomes | X and Y |

### **Telomere Studies**

- |   |         |
|---|---------|
| 25. Telomeric region probes for 22 autosomes    | 1 to 22 |
| 26. Telomeric region probes for sex chromosomes | X and Y |

### **Prenatal Diagnosis for Suspected Trisomy Syndrome or Mosaicism Studies**

- |  |           |
|--|-----------|
| 27. Numeric screening probes for trisomies       | 13,18, 21 |
| 28. Numeric screening probes for sex chromosomes | X and Y   |

### **Oncology**

- |  |                   |
|--|-------------------|
| 29. Bcr/abl probe for Philadelphia chromosome  | t(9;22)(q34;q1.2) |
| 30. FISH probes for retinoblastoma (Rb) gene   | 13q14.3           |
| 31. FISH probes for tumour suppressor gene p53 | 17p13.1           |

## Molecular Diagnostic Service

Diseases/Syndromes	Genes Involved	Investigation
<b>1. Chromosomal Abnormalities</b>		
Subtelomeric deletion/duplication	Telomeres	deletion/duplication
Trisomies	chr 13, 18, 21	trisomy 13, 18, 21
<b>2. Connective Tissue Disorder</b>		
Marfan syndrome	<i>Fibrillin</i>	point mutation
<b>3. Craniosynostotic Syndromes</b>		
Antley-Bixler syndrome	<i>POR</i>	point mutation
Crouzon Syndrome	<i>FGFR2</i>	point mutation
Apert syndrome	<i>FGFR2</i>	point mutation
Pfeiffer syndrome	<i>FGFR2</i>	point mutation
Saethre-Chotzen syndrome	<i>Twist</i>	deletion
<b>4. Eye Diseases</b>		
Cone -Rod dystrophy	<i>CRX</i>	point mutation
Corneal dystrophy (lattice type)	<i>BIGH3</i>	point mutation
<b>5. Hearing Loss</b>		
Non-syndromic deafness	<i>GJB2</i>	point mutation
Non-syndromic deafness	mtDNA 1555A/G	point mutation
Waardenburg syndrome type 1	<i>PAX3</i>	point mutation
<b>6. Hematology</b>		
$\alpha$ -Thalassemia	<i>HBA</i>	SEA/R/L deletion point mutation
Hemophilia A	<i>Factor VIII</i>	linkage analysis inversion in intron 22
<b>7. Inborn Errors of Metabolism</b>		
Fabry disease	<i>GLE</i>	point mutation
Gaucher disease	<i>GCB</i>	point mutation
Glutaric aciduria type 1	<i>GCDH</i>	point mutation
Pyruvate dehydrogenase deficiency	<i>PDHA1</i>	point mutation
Ornithine carbamoyltransferase deficiency	<i>OTC</i>	point mutation

## 8. Mitochondrial Disorders

MELAS, MERRF, NARP

*Mitochondrion*

point mutation

### Diseases/Syndromes

### Genes Involved

### Investigation

## 9. Neurology

Charcot-Marrie-Tooth, 1A

*PMP22*

gene duplication

Charcot-Marrie-Tooth, 1B

*MPZ*

point mutation

Charcot-Marrie-Tooth, X-linked, 1

*GJB1*

point mutation

Congenital central hypoventilation syndrome

*CCHS*

point mutation

polyalanine expansion

Dentatorubral-pallidoluysian atrophy

*DRPLA*

CAG expansion

Fragile X syndrome, type A

*FMR1*

CGG expansion

Fragile X syndrome, type E

*FMR2*

GCC expansion

Friedreich ataxia

*X25*

GAA expansion

Huntington's disease

*IT15*

CAG expansion

Pelizaeus Merzbacher disease

*PLP*

gene duplication

Pelizaeus-Merzbacher-like disease

*GJA12*

point mutation

Spastic paraplegia 2, X-linked

*PLP*

point mutation

Spinocerebellar ataxias:

SCA1

*ATXN1*

CAG expansion

SCA2

*ATXN2*

CAG expansion

SCA3

*ATXN3*

CAG expansion

SCA6

*CACNA1A*

CAG expansion

SCA7

*ATXN7*

CAG expansion

SCA8

*SCA8*

CTG expansion

SCA12

*PPP2R2B*

CAG expansion

## 10. Neuro-cutaneous Syndrome

Neurofibromatosis type 1

*NF1*

point mutation deletion

## 11. Neuro-muscular Diseases

Duchenne muscular dystrophy

*DMD*

exon(s) deletion/ duplication

linkage analysis

point mutation

Myotonic dystrophy

*DMPK*

CTG expansion

Kennedy's disease

*Androgen receptor*

CAG expansion

Spinal muscular atrophy

*SMN1*

exon deletion

**12. Renal Diseases**

Alport syndrome	<i>COL4A5</i>	linkage analysis
Polycystic kidney disease, adult	<i>PKD</i> <i>PKD2</i>	linkage analysis
Polycystic kidney disease, AR type	<i>ARPKD</i>	
Senior-Loken syndrome 5	<i>IQCB1</i>	point mutation

**13. Dermatologic Disorder**

Incontinentia pigmenti	<i>NEMO</i>	exons deletion point mutation
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**Diseases/Syndromes**

**Genes Involved**

**Investigation**

**14. Skeletal Dysplasia**

Achondroplasia	<i>FGFR3</i>	point mutation
Hypochondroplasia	<i>FGFR3</i>	
Thanatophoric Dysplasia	<i>FGFR3</i>	
Metaphyseal chondrodysplasia (McKusick type)	<i>RMRP</i>	point mutation
Conradi-Humermann syndrome	<i>EBP</i>	point mutation
Epiphyseal dysplasia, multiple, 4	<i>DTDST</i>	point mutation
Achondrogenesis, type Ib	<i>DTDST</i>	point mutation
Diatrophic dysplasia	<i>DTDST</i>	point mutation
Hypophosphatemic ricket, X-linked type	<i>PHEX</i>	point mutation
Hypophosphatemic ricket, AD type	<i>FGF23</i>	point mutation
Leri-Weill syndrome	<i>SHOX</i>	deletion point mutation
Pseudoachondroplasia	<i>COMP</i>	GAC expansion
Schwartz-Jampel syndrome, type 2	<i>LIFR</i>	point mutation
Spondylocostal dysostosis (Jarcho-Levin syndrome)	<i>DLL3</i>	point mutation
Pseudohypoparathyroidism / Albright hereditary osteodystrophy	<i>GNAS1</i>	point mutation

**15. Sex Disorder**

Sex determination	<i>SRY</i>	deletion point mutation
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**16. Syndromes / Dysmorphology**

Angelman syndrome	Chromosome 15	microdeletion / UPD
Blepharophimosis-Ptosis-Epicanthus-Inversus syndrome (BPES)	<i>FOXL2</i>	point mutation deletion
Costello syndrome	<i>HRAS</i>	point mutation
Greig syndrome	<i>GLI 3</i>	point mutation
Noonan syndrome	<i>PTPN11</i>	point mutation
Prader Willi syndrome	Chromosome 15	microdeletion / UPD
Rett syndrome	<i>MECP2</i>	point mutation deletion
Smith-Megenis syndrome	<i>RAI1</i>	point mutation
Sotos syndrome	<i>NSD1</i>	point mutation deletion
Triple A syndrome	<i>GL003</i>	point mutation

**17. Miscellaneous**

X-inactivation pattern	<i>AR</i>	Abnormal X-inactivation
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**Contact Information:**

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Department of Health, Hong Kong SAR Government

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Website: [http://www.dh.gov.hk/english/main/main\\_cgs/main\\_cgs.html](http://www.dh.gov.hk/english/main/main_cgs/main_cgs.html)