### Instructions for Prenatal Diagnosis Testing

#### Specimens for Testing

<table>
<thead>
<tr>
<th>Specimen</th>
<th>Quantity</th>
<th>Container</th>
<th>Specimen delivery and storage</th>
</tr>
</thead>
<tbody>
<tr>
<td>Amniotic fluid</td>
<td>15 – 30 ml+</td>
<td>Sterile bottle</td>
<td>Specimen should arrive in the laboratory: Mon-Fri: 9 a.m. to 3:30 p.m. Excluding Saturdays and public holidays (working day: call before 12 noon, sample will be collected on same day) Overnight specimens may be stored at 4°C in the refrigerator but must <strong>NOT</strong> be frozen.</td>
</tr>
<tr>
<td>Chorionic villi</td>
<td>Villi tissue (about 25 mg)</td>
<td>Sterile bottle with medium*</td>
<td></td>
</tr>
<tr>
<td>Blood for karyotyping</td>
<td>3 ml</td>
<td>Heparin tube</td>
<td></td>
</tr>
<tr>
<td>Fetal blood / cord blood / baby blood (&lt;1yr) for karyotyping</td>
<td>Prefer at least 2 ml (2 tubes × 1 ml)</td>
<td>Special small 1.3 ml heparin tube (for Paed use)*</td>
<td></td>
</tr>
<tr>
<td>Blood for molecular genetic test</td>
<td>3 ml (1 ml for fetal / cord blood and Paed case)</td>
<td>EDTA bottle</td>
<td></td>
</tr>
<tr>
<td>Other tissues</td>
<td>Tissues pieces (about 25 mg)</td>
<td>Sterile bottle with medium</td>
<td></td>
</tr>
</tbody>
</table>

* Containers with medium and the special small heparin tube can be obtained from the Prenatal Genetic Diagnosis Laboratories (PGDL)

+ For earlier gestational age or with additional examination request (e.g. Fetal DNA Chips), higher volume (e.g. 30-40 ml fluid) is recommended

#### Referral Instructions

Referral clinic is recommended to contact PGDL first to confirm the availability of test requested.

The molecular genetic and cytogenetic request form [PWH/ACPPD-L0701-A-V6-F1] and consent form must be completed together with the specimen and send to PGDL as soon as possible:

**Prenatal Genetic Diagnosis Laboratories**
4/F, Block K (Day Treatment Block)
Prince of Wales Hospital
Shatin, N.T., Hong Kong

**Referral, Specimen and Results Enquiry:** 3505 1557
Specimen taking and sending

1 Laboratory Operation Procedures

1.1 Prenatal diagnosis is offered to patients at risk of chromosome anomalies/genetic diseases.

Reasons for referral will include the following:
- Abnormal ultrasound scan
- Carrier of a chromosomal structural rearrangement/genetic disease
- Elevated risk indicated by biochemical and/or ultrasound screening
- High risk indicated by non-invasive prenatal screening
- Advanced maternal age
- Previous chromosome anomaly

Specimens for Prenatal diagnosis are usually one of the following sample types:
- Amniotic fluid
- Chorionic villi / placental tissues
- Fetal blood / peripheral blood

2 Specimens

2.1 All prenatal diagnostic laboratory tests should be prearranged with the laboratory for test availability and specimen bottles to be supplied with special instructions for specimen taking, storing and transport.

2.2 Collection of amniotic fluid

2.2.1 Amniotic fluid about 15 to 30 ml should be collected in sterile universal.
2.3 Collection of chorionic villi / placental tissues

2.3.1 Chorionic villi fragments about 25 mg should be collected in sterile universal bottles with transport medium.

2.3.2 Transport medium bottles can be obtained from the Prenatal Genetic Diagnosis Laboratories. If it is not used immediately, it should be kept at 4°C in the refrigerator for maximum three months (expiry date is stated on the tube).

2.4 Collection of fetal blood and peripheral blood

2.4.1 At least 3 ml peripheral blood (1.5 ml for fetal/cord blood) should be put in heparin bottles for chromosome analysis.

2.4.2 At least 3 ml peripheral blood (1.5 ml for fetal/cord blood) should be put in EDTA bottles for molecular genetic analysis.

2.4.3 Fetal blood should be verified of the fetal origin by the sender or referral unit

2.4.4 Heparin and EDTA bottles are obtainable from Prenatal Genetic Diagnosis Laboratories or the general Pathology Laboratory.

2.5 Specimen delivery

2.5.1 Specimen should be sent to the laboratory at cool room temperature on the sampling day.

2.5.2 If specimen cannot be sent on the sampling day, it can be kept at 4°C in the refrigerator but **must not be frozen**. Specimen should arrive in the laboratory within 48 hrs of sampling.

2.5.3 For overseas, specimen should be shipped at 4°C to the laboratory by overnight express. Specimen should arrive in the laboratory within 48 hrs of sampling and **must not be frozen**.

3 Test charges

Please refer to the attached price list or call 3505 1557 for details.