

## **The Aga Khan University Hospital Clinical Laboratories Update**

### **Chromosomal Analysis on Chorionic Villi**

#### **Introduction**

Chromosomal anomalies in the developing fetus can occur in any pregnancy and lead to death prior to or shortly after birth or to costly lifelong disabilities. Early detection of fetal chromosomal aneuploidies, an atypical number of certain chromosomes, can help physician and couple to evaluate their pregnancy options. Current diagnostic methods include maternal serum sampling or nuchal translucency testing, which are minimally invasive diagnostics, but lack sensitivity and specificity. The gold standard, karyotyping, requires amniocentesis or chorionic villus sampling, which are highly invasive technique to early diagnosis. A number of risk factors can increase the probability of chromosomal aberrations in pregnancy, maternal age being the most important. Other risk factors are a history of infertility and a family history of chromosomal disorders. CVS sampling from the developing placenta is usually performed between 10 and 12 weeks of gestation.

#### **Principle**

The purpose of performing prenatal diagnosis via chorionic villi sampling is to provide early detection of fetal chromosomal abnormalities. Prenatal diagnosis of chromosomal disorders is carried out by means of culturing cells obtained from CVS at approximately 10 to 12 weeks of gestation.

#### **Indications for performing CVS include:**

- Advanced maternal age (35 years or older at time of delivery)
- Parental chromosome rearrangement
- Previous pregnancy or birth of a child with a chromosomal abnormality or neural tube defect.
- Increased risk of chromosomal abnormality or neural tube defect by maternal serum screening
- Fetal malformation detected on ultrasound
- Family history of genetic disorders

**Specimen Collection:**

10-20mg of Chorionic villi tissues

(Note: CVS specimens should be collected into sterile bottles containing Transport medium)

**Schedule:** Performed Monday through Sunday; Report 10 Working Days

