



## CHROMOSOME DIAGNOSTIC FACILITY

### Indications:

- Dysmorphic features/Congenital malformations
- Syndromic and non-syndromic birth defects
- Trisomy/monosomy syndromes (eg. Down syndrome, Edwards syndrome, Patausyndrome, Klinefeltersyndrome, Turner syndrome)
- Delayed developmental milestones/growth retardation
- Abnormal developmental of the genitalia/ambiguous genitalia
- Delayed puberty/lack or late onset of secondary sexual characters
- Infertility
- Mental retardation
- Recurrent miscarriages
- Primary amenorrhoea
- Poor gonadal development
- Premature menopause/premature ovarian failure
  
- Prader-Willi/Angelman syndrome
- Williams syndrome
- DiGeorge syndrome
- SRY gene
- Androgen receptor
- Miller Diecker
- Kallman Probe
- BCR/ABL

- Prenatal testing in case where there is
  - Prior affected child
  - Maternal age factor
  - Change in routine ultrasound pattern

**From peripheral blood**

2-3 ml heparinised blood (green cap vacutainers)

**From prenatal samples**

**Amniotic fluid**

20-30 ml amniotic fluid in sterile centrifuge tubes for chromosome analysis

**Aneuploidy FISH in Uncultured amniocytes and Chorionic Villus sample (CVS)**

**Fetal cord blood**

1-2 ml heparinised fetal cord blood

**Cost of chromosome-based diagnostic services/sample**

Peripheral blood	: Rs 2000/-
Fetal cord blood	: Rs 3000/-
Other prenatal samples	: Rs 4500/-

**Fluorescence based chromosome analysis**

Conventional probes (whole chromosome paint)	: Rs 5000/-
Locus Specific Probes and	<b>Aneuploidy FISH: Rs 5000/-</b>

## **Reporting time**

- 2-3 weeks for blood samples.
- 3-4 weeks for prenatal samples
- 1 week for Aneuploidy FISH

## **Transport of samples**

The samples should reach us within 24-48 hrs in cold conditions (packed with ice or cool packs). Samples received within 24 hrs are good because the viability of the cells is lost when the sample is sent late which affects the quality of chromosomes.

**CYTOGENETIC ANALYSIS IS NOT POSSIBLE IN FROZEN SAMPLES.**

Dr.T.Karthik Bhardawaj  
Principal Clinical Geneticist  
MD (Pediatrics),DNB (Medical Genetics)  
Medical Biotechnology Complex (MBC)  
Centre for Cellular and Molecular Biology  
CCMB) Annex-II  
Council of Scientific and Industrial Research (CSIR)  
Uppal Road, Hyderabad 500 007. INDIA  
Tel: 00-91-40-27195612 (Off) 00-91-40-27195568 (Lab)