

## **XI. Genetic & Metabolic Unit**

### **Services Offered**

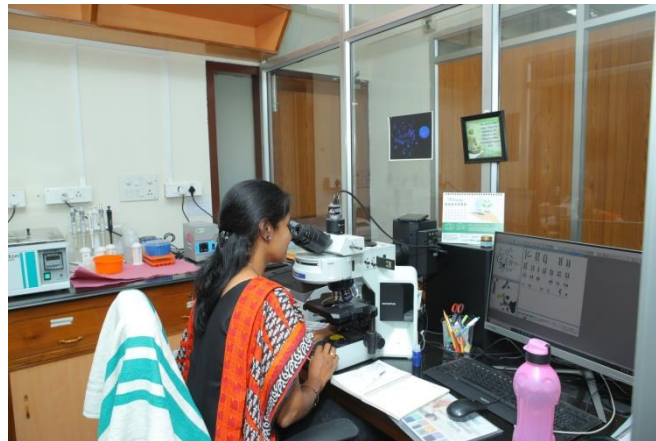
The Genetic and Metabolic unit offers advanced laboratory diagnostic facilities to identify and screen the developmental issues in children. The various diagnostic services include biochemical analysis, cytogenetic analysis and molecular biology techniques. The detailed infrastructure and procedures available at the unit is summarised below.

### **Bio-chemistry Section**

The Bio-chemistry section of the laboratory is equipped with diagnostic devices like Automated Chemiluminescence Immunoassay Analyser for doing thyroid function test in children and adolescents.



## Human Cytogenetics



The Human Cytogenetic Laboratory is involved in genetic diagnostic services to the patients attending Medical College and SAT Hospital, Thiruvananthapuram. Cytogenetic study is carried out in children with suspected chromosomal disorders (Intellectual Disability/ Dysmorphism/ Multiple malformation/ Down syndrome/ Short stature/ Ambiguous genitalia) and in couples with recurrent pregnancy loss, infertility etc. The lab follows stringent sterile conditions for sample collection and culture procedures. The documentation and reporting of chromosomal analysis follows the International standard for Chromosomal Nomenclature (ISCN- 2013). The laboratory is equipped with computerised karyotyping system (Applied Spectral Imaging Software) allowing a substantial shortening of turnaround time of the results. The results will be verified by a qualified Medical Geneticist and genetic counselling will be provided to the needy patients.

Standardizing and diagnosis based on FISH (Fluorescent Insitu Hybridization), the first of its kind in Government sector in Kerala was started. The introduction of this new testing facility will help in diagnosing new diseases and to rule out Aneuploidy conditions and Microdeletion syndromes prevailing among children and adolescents.

## Molecular Genetics

This section of the laboratory is involved in research activities which include an on-going multicentric collaborative study on lysosomal storage disorder. The lab is also dealing with mutation analysis of rare diseases. Blood samples are collected from SAT as well as from other collaborative centres. DNA isolation followed by PCR, visualization by Gel Documentation system and finally sequencing are being done.

## **Clinical Research Laboratory of Genetic & Metabolic Unit, CDC**

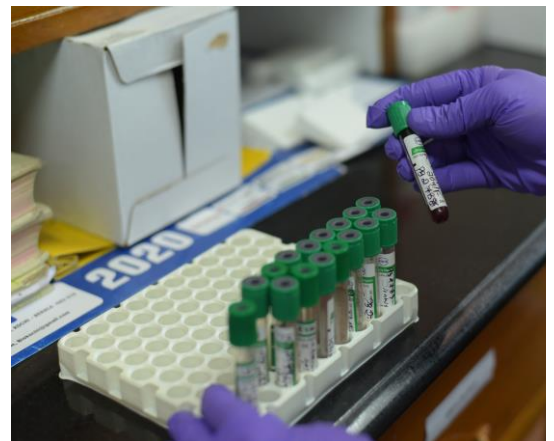


**Dr.Santhi S., Cytogeneticist  
doing Chromosomal Analysis Test at CDC**



**Ms.Haritha Dasaradh, Senior Lab. Technician  
Doing Biochemical Analysis at CDC**

**Molecular Analysis**



**Blood Collection**

**Sample verification**

## XII. Imageology Division- Sradha Project- Foetal Medicine Clinic

1. As part of the Reduction of childhood disability initiatives of CDC a project titled "SRADHA project was started during the year 2020. Dr.Pio James, Assistant Professor, Obstetrics & Gynecology, Govt. Medical College, Ernakulam is the consultant of the clinic.
- 2.
3. The "Sradha project" aims at reduction of childhood disability through various antenatal intervention including anomaly scanning using the 4D Ultrasonography machine and advanced equipments available at Genetic & Metabolic unit of CDC.
4. The major services offered are First trimester scan (11-14 weeks), Early Anomaly Scan - 16 weeks), Target Anomaly Scan (18 to 27 weeks), Target Anomaly scan with fetal ECHO, Growth scan with fetal Doppler

5



