

How to transport the specimens?

- a. Keep specimen cool, NOT frozen during transport to the laboratory.
- b. Do not immerse the specimen tube or container in water to avoid contamination.
- c. Samples should be sent as soon as possible to the Cytogenetics Laboratory with same-day or overnight transport preferable.
- d. The laboratory accepts blood samples and other tissue for chromosomal studies Mondays to Fridays from 8AM to 3PM.
- e. Bone marrow aspirate sample is accepted Mondays to Thursdays only. The cutoff time is 3PM.

What are the result turnaround time?

Blood results	: 2 to 3 weeks
Bone Marrow results	: approximately 1 month
Tissue	: approximately 1 month
FISH	: 2 to 3 weeks

THE LARGEST PROVIDER OF GENETICS SERVICES IN THE COUNTRY



The **INSTITUTE OF HUMAN GENETICS** provides services through its Genetics Clinic, Specialized Laboratories and Research Unit

- Component Units include:
- 1. Cytogenetics Laboratory
 - 2. Molecular Genetics Laboratory
 - 3. Biochemical Genetics Laboratory
 - 4. Microarray Core Laboratory
 - 5. Hemoglobinopathy Reference Unit
 - 6. Clinical Genetics at PGH
 - 7. Newborn Screening Center - NIH

For inquiries on services, please contact:



Institute of Human Genetics
National Institutes of Health
University of the Philippines, Manila

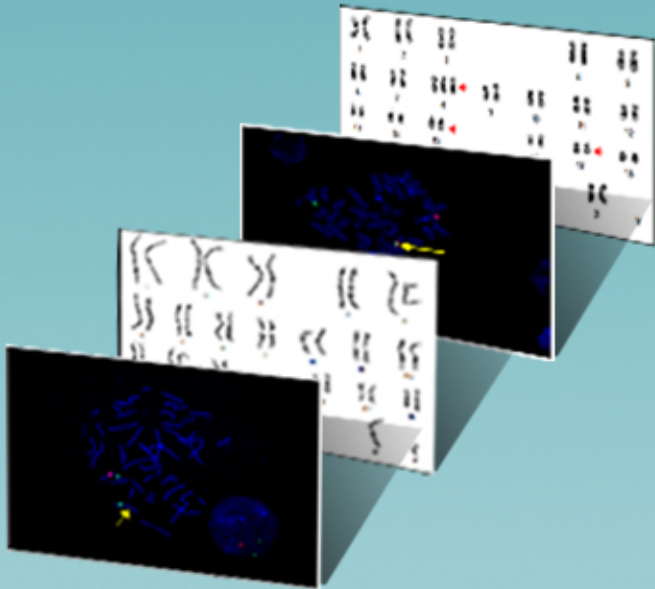
Pedro Gil Street, Malate, Manila
Tel No.: (632) 3101780
Fax No.: (632) 5269997
Website: <http://ihg.upm.edu.ph>

For further inquiries, contact the Cytogenetics laboratory-NIH at telephone numbers: (02) 310-1780 / 310-0788 local 107, Hotline: 0928-5060963
Email: cytogenetics-ihg@upm.edu.ph

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Institute of Human Genetics
National Institutes of Health Building
University of the Philippines Manila
Pedro Gil Street, Ermita, Manila 1000

CYTOGENETICS LABORATORY

The Philippines' Leading Cytogenetics Laboratory



The Institute of Human Genetics is ISO 9001:2008 certified



CYTOGENETICS LABORATORY

In 1991, Cytogenetics Laboratory was started at the Medical Genetics Unit of the University of the Philippines College of Medicine. This unit eventually became the Institute of Human Genetics, National Institutes of Health, University of the Philippines Manila (IHG-NIH-UPM) in 1999 that provides comprehensive diagnostic services for postnatal congenital and neoplastic/oncologic disorders.

We offer:

1. Constitutional Diagnostic Services

- a. Peripheral Blood - Routine and High-Resolution Analysis
- b. Newborn Blood Chromosome Analysis
- c. Family Studies
- d. Mosaicism Analysis
- e. Solid Tissues - Products of Conception, Skin Biopsies
- f. Fragile X Screening
- g. C-banding
- h. AgNOR

2. Neoplastic Diagnostic Services

- a. Bone Marrow Aspirate/Leukemic Blood
- b. Solid Tumors/Lymph Nodes

3. Fluorescence in situ hybridization (FISH)

a. Microdeletion probes for:

- DiGeorge/Velo-Cardio-Facial syndrome
- Prader-Willi/Angelman syndrome
- Williams-Beuren syndrome

b. Translocation probes for BCR/ABL

FREQUENTLY ASKED QUESTIONS (FAQs)

What is Chromosomal Analysis or Karyotyping?

Chromosomal Analysis or Karyotyping is a test that is used to detect abnormalities in human chromosomes. These abnormalities may be present as numerical abnormalities (aneuploidy) or structural (translocation, deletion, insertion, duplication and inversion) rearrangements. Some chromosomal abnormalities/rearrangements are present from birth and in every cell of the body, these are called constitutional abnormalities and some are acquired aberrations which are commonly found in neoplastic and cancer cells, such as leukemia.

Why do we study chromosomes?

This can help clinicians with reaching a diagnosis, as an ECG is to a cardiologist or an EEG is to a neurologist, or an X-ray is to a pulmonologist. The diagnostic findings can affect counselling for families – e.g. Type of translocation in Down syndrome patients.

When is constitutional karyotyping done?

Constitutional karyotyping is done in children born with dysmorphic features, multiple congenital anomalies, developmental delay, unexplained mental retardation, autism spectrum disorders, growth retardation, learning disability or any combination of the above. It is also done in cancer patient for management, diagnosis, prognosis, monitoring of their conditions.

What is FISH analysis?

Chromosome aberrations are variable in size. Some aberrations can be detected in conventional chromosome analysis, while some are too small to be seen. One technique to detect microdeletion is FISH (Fluorescence In Situ Hybridization) Analysis.

FISH Analysis aids to detect chromosomal abnormalities in patients that appeared to be normal in conventional cytogenetic analysis. It uses specific probes that bind only to those parts of the chromosome with which they show a high degree of sequence similarity.

In majority of cases, patients suspected with microdeletion syndrome, a routine or high resolution cytogenetic analysis is recommended in conjunction with FISH. This is to confirm or rule out another chromosome etiology for the clinical phenotype seen..

How to collect the specimens?

- a. Peripheral blood - 4 ml for adult and 1-2 ml for newborn/infant in a sterile sodium heparin tube (green top). Ensure that sample is NOT CLOTTED.
- b. Bone Marrow Aspirate – minimum of 3 ml (preferred) in a sterile sodium heparin tube (green top) or transport medium provided by the laboratory. Ensure that sample is NOT CLOTTED.
- c. Fetal Tissue, Placental Villi or Skin Biopsy – 1 to 2 cm³ biopsy samples obtained in an aseptic manner using sterile instruments and placed in transport medium provided by the laboratory. DO NOT PLACETISSUES IN FORMALIN OR WATER! If storing overnight prior to shipping, please refrigerate! Notify the laboratory before collecting/sending any solid tissue sample.
- d. Each specimen must clearly labeled with at least two patient identifiers such as patient name and birth date.
- e. Each specimen must be accompanied by doctor's request with the following information: First and last name, birth date, gender, physician's name & contact number, originating hospital or laboratory, clinical indication and tests ordered, and date of collection.