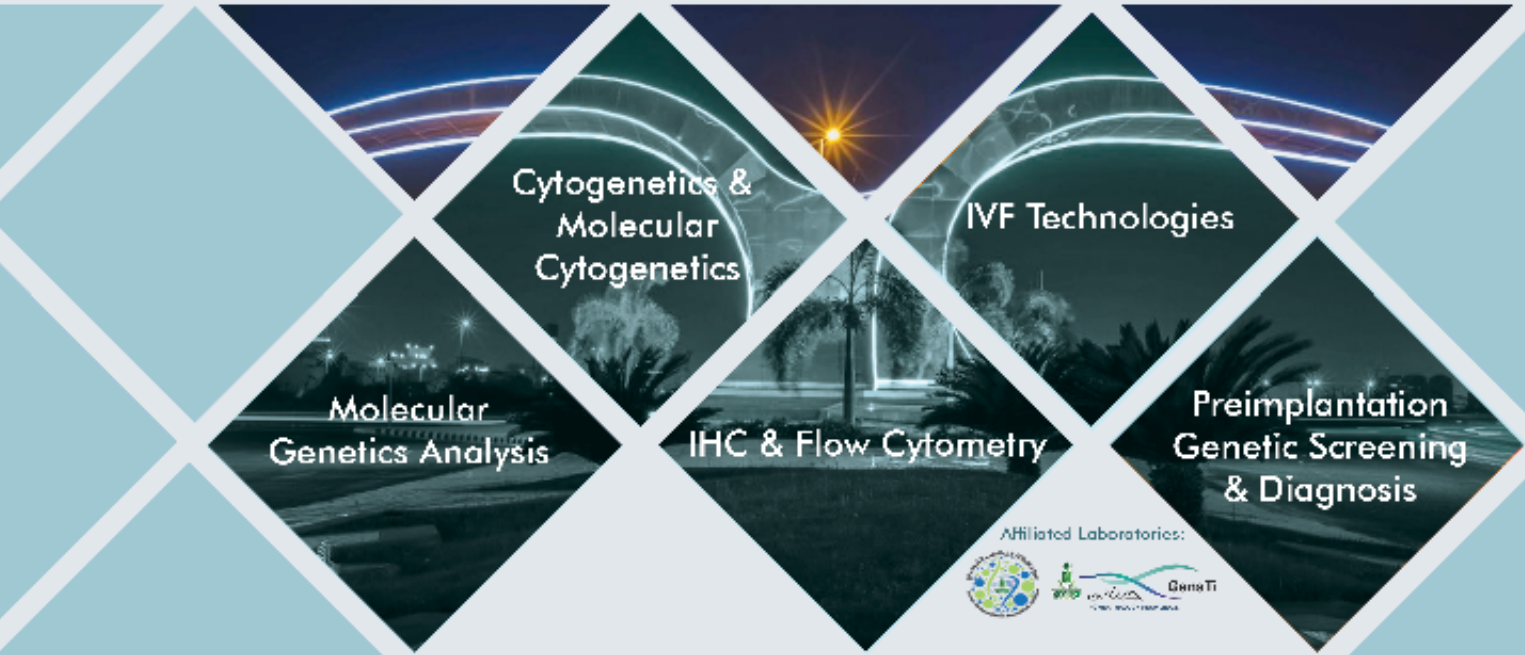
Ro'ya

Specialized Medical Laboratories
Laboratory Test Directory
2018 Edition



Affiliated Laboratories:

Ro'ya

Specialized Medical Laboratories

Laboratory Test Directory

2018 Edition

Published by Ro'ya Specialized Medical Laboratories - King Abdulaziz University

"Apart from the comprehensive tests and services offered in this catalogue, our experts are capable of customizing a variety of laboratory tests to suit your needs"

Introduction

Ro'ya Specialized Medical Laboratories at King Abdulaziz University is an initiative supported by the Kingdom of Saudi Arabia's Vision 2030 that led to establishment a group of specialized medical laboratories operating within the organization of the Medical Center at King Abdulaziz University. This initiative aims to deliver its services to all health sectors in the Kingdom in order to achieve self-sustainability as one of the main objectives of Vision 2030 at the university.

Mission

To help the Saudi society and beyond by providing high quality and internationally accredited diagnostic laboratory services to support the heath care sector.

Vision

Achieve par excellence and become renowned for our quality and variety of diagnostic services in the kingdom of Saudi Arabia.

Objectives

- Work towards accomplishing the vision of the kingdom of Saudi Arabia 2030 by utilizing full physical and human potentials available.
- Adopt best medical laboratory practices according to optimum internationally known standards and criteria.
- Provide specialized medical laboratory services at competitive price for both Saudis and residents alike and report within shortest turnaround time.
- Market medical laboratory services and capabilities available to both private and public sectors.
- Achieve self-sustainable income for the university.
- Professionally operate all specialized medical laboratories at their full potential with precise monitoring to guarantee smooth functioning.

General Instructions for Specimen Collection and Handling

Collection and Shipment of Specimens:

All samples will be shipped / delivered and received at our main receiving area at King Fahad Medical Research Center (KFMRC), Jeddah. Ro'ya will provide a special designated container for picking up and delivery of specimen upon request. Courier services are available for transporting specimens to Ro'ya from many locations throughout the Kingdom. The service provides overnight delivery of specimens (including frozen) under controlled condition. Any special requirements will be arranged accordingly. Contact us for shipment arrangement and special requests at: roya-info@kau.edu.sa

Hours of Operation

Saturday - Thursday.

After hours operation, please contact us at: +966 54 9779685

For CIPM hours operation, please contact us to schedule an appointment at: +966 53 3537257

Policies

To avoid specimen rejection, each specimen must be accompanied with a complete REQUEST FORM that provides:

- The patient full name, medical record number, hospital name, date of birth and gender (If forename has not been established for a new-born baby, the request form must contain surname and name of his/her parents)
- Specimen type, time/date of specimen collection and test desired
- Clinical indications, history and therapy (for oncology)
- Referring physicians name and contact number
- Specimen container, transportation temperature and transport medium must be checked

A blood sample will be rejected if:

- Clotted
- Haemolysed or low in volume
- Unlabeled or labelled improperly
- Wrong container
- Improperly stored or transported

Other specimens may have a prolonged turnaround times because of lack of necessary ancillary specimens or patient information. You will be notified of rejected or problem specimens upon receipt.

Cancellation of Tests:

Request for cancellation must be made via phone or email and will be honored at no charge to the sender if received prior to test setup. Cancellation request after test setup cannot be honored. A report will be issued automatically and charged appropriately.

Additional Tests (Add-ons):

Requests for additional orders on specimens received by Ro'ya must be confirmed via email. Call to check on sample requirements and availability to avoid any rejections. Repeated or additional tests will take additional turnaround time.

Policies

Test Result call back and confidentiality:
Results will be sent to a pre-assigned email address or given to the patient.
Ro'ya endeavors to maintain the confidentiality of all patients' information.

Unsatisfactory Analytical Result:
Ro'ya endeavors to perform all tests to the highest standards. Tests generating unsatisfactory or inconclusive results will not be charged to the sender.

Urgent orders:
All referred orders will be treated as routine and will be accommodated according to the laboratory working hours. Urgent requests must be approved by an authorised member of staff at the sending facility prior to submitting the order and sending the specimens. Processing urgent orders will waiver any contractual discounts and will be charged at the full listed price of the test(s).

Unlisted tests:
New procedures are developed throughout the year; therefore, some tests are not listed on our request form, booklet or website.
For information about the unlisted tests, contact Ro'ya general inquiry.

Supplies:
Special specimen collection containers and kits, sterile vials and request forms are supplied upon request.

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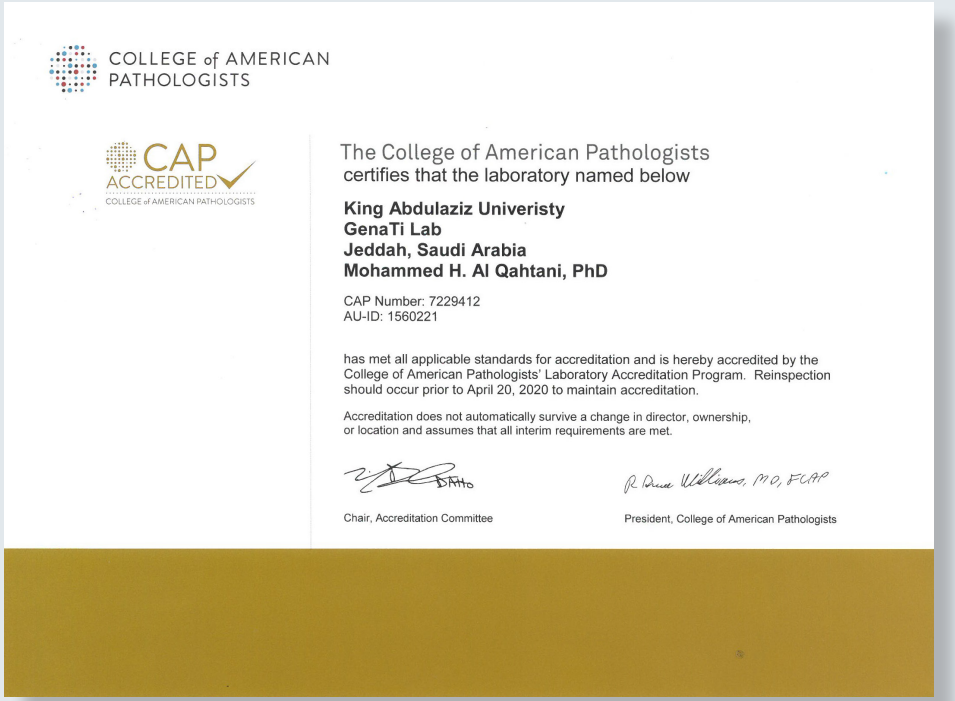
International Accreditations



COLLEGE of AMERICAN PATHOLOGISTS

CAP ACCREDITED
COLLEGE of AMERICAN PATHOLOGISTS

The College of American Pathologists
certifies that the laboratory named below

King Abdulaziz Univeristy
GenaTi Lab
Jeddah, Saudi Arabia
Mohammed H. Al Qahtani, PhD

CAP Number: 7229412
AU-ID: 1560221

has met all applicable standards for accreditation and is hereby accredited by the College of American Pathologists' Laboratory Accreditation Program. Reinspection should occur prior to April 20, 2020 to maintain accreditation.

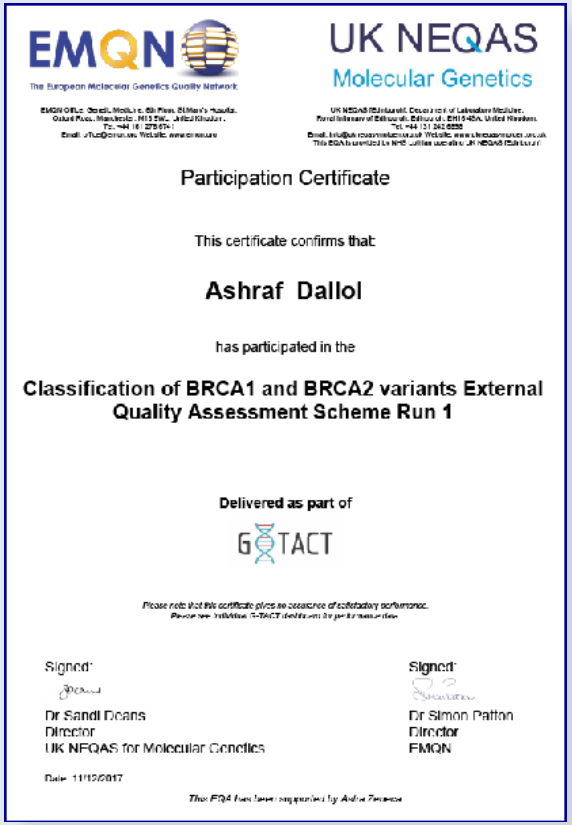
Accreditation does not automatically survive a change in director, ownership, or location and assumes that all interim requirements are met.

Chair, Accreditation Committee

R. Dirk Williams, M.D., FCAP

President, College of American Pathologists

International Accreditations



EMQN
The European Molecular Genetics Quality Network

UK NEQAS
Molecular Genetics

Participation Certificate

This certificate confirms that:

Ashraf Dallol

has participated in the

Classification of BRCA1 and BRCA2 variants External Quality Assessment Scheme Run 1

Delivered as part of

GENTACT

Please note that this certificate gives no assurance of satisfactory performance.

Signed:

Dr Sandi Deans
Director
UK NEQAS for Molecular Genetics

Signed:

Dr Simon Patton
Director
EMQN

Date: 11/12/2017

This EQA has been supported by Ardea Biosciences

Cytogenetics

Amniotic Fluid Chromosomal Analysis

CODE GT001

This test determines the chromosomal status of the fetus. Numerical and structural chromosomal abnormalities can be diagnosed. Sex and the presence of more than one cell line (mosaicism) can also be determined. Indications include diagnosis of chromosome abnormalities in fetuses of women who are of advanced maternal age; had a previous child with a chromosome abnormality; parental carrier of a balanced translocation, inversion or marker chromosome and parental mosaicism.

SPECIMEN REQUIREMENTS	Use sterile techniques to transfer 30 ml in 3 different sterile plastic conical tube for transport
TEMPERATURE	Maintain at room temperature. Send to the Lab ASAP
SETUP	Saturday to Thursday
TURN AROUND TIME	20 - 30 days working days

Chorionic Villi Chromosomal Analysis

CODE GT002

Numerical and structural chromosomal abnormalities can be diagnosed. Sex and presence of more than one cell line (mosaicism) can also be determined. Indications include diagnosis of chromosome abnormalities in fetuses of women who are of advanced maternal age; had a previous child with a chromosome abnormality; parental carrier of a balanced translocation, inversion or marker chromosome and parental mosaicism.

SPECIMEN REQUIREMENTS	5–25mg in 2 screw top 15 ml sterile conical tubes filled with sterile transport media
TEMPERATURE	Maintain at room temperature. Send to the Lab ASAP
SETUP	Saturday to Thursday
TURN AROUND TIME	14 - 21 days working days

Fetal or Cord Blood Chromosomal Analysis

CODE GT003

Numerical and structural chromosomal abnormalities can be diagnosed. Sex and presence of more than one cell line (mosaicism) can also be determined. Indications include diagnosis of chromosome abnormalities in fetuses with dysmorphic features; multiple congenital anomalies; parental carrier of a balanced translocation, inversion or marker chromosome and parental mosaicism.

SPECIMEN REQUIREMENTS	2 ml in Sodium Heparin tube
TEMPERATURE	Maintain at room temperature. Send to the Lab ASAP
SETUP	Saturday to Thursday
TURN AROUND TIME	7-10 working days

Product of Conception /Stillbirths/Abortuses Chromosomal Analysis

CODE GT004

For the diagnosis of chromosome abnormalities present in the fetus during the first trimester of pregnancy. Chromosome aberrations observed in the CVS specimen but not in the fetus (confined placental mosaicism) occurs in approximately 1% of cases.

SPECIMEN REQUIREMENTS	5 mm of the tissue in 15 ml or 50 ml conical sterile tube with sterile culture media. The preferred tissue types include kidney, thymus, heart, lung muscle
TEMPERATURE	Maintain at room temperature. Send to the Lab ASAP
SETUP	Saturday to Thursday
TURN AROUND TIME	14 - 21 working days

Peripheral Blood Chromosomal Analysis

CODE GT005

Chromosomal analysis of peripheral blood is used to detect chromosomal abnormalities when a blood is suspected of having a chromosomal abnormality, when an infant has congenital abnormalities, when a woman experiences miscarriages or infertility, and when an adult shows signs of a genetic disorder. It may also be ordered to detect the presence of a chromosomal abnormality in family members when it has been detected in a child or in another family member.

SPECIMEN REQUIREMENTS	3-5 ml Peripheral blood in Sodium Heparin tube
TEMPERATURE	Maintain at room temperature. Send to the Lab ASAP
SETUP	Saturday to Thursday
TURN AROUND TIME	7-10 working day

Peripheral Blood, Constitutional mosaicism Chromosomal Analysis

CODE GT006

Extended Chromosomal analysis by karyotyping for low degree chromosomal mosaicism. Some low degree cases may require FISH analysis if a specific abnormality is to be excluded.

SPECIMEN REQUIREMENTS	3-5 ml Peripheral blood in Sodium Heparin tube
TEMPERATURE	Maintain at room temperature. Send to the Lab ASAP
SETUP	Saturday to Thursday
TURN AROUND TIME	7-10 working days

Peripheral Blood-Breakage Study (Ataxia Telangiectasia) Chromosomal Analysis

CODE GT007

This analysis evaluates the amount of chromosome breakage in blood cells and will not identify any specific mutation in the DNA. This test will reliably detect affected individuals but is not an appropriate test for unaffected carriers of the disorder.

SPECIMEN REQUIREMENTS	3-5 ml Peripheral blood in Sodium Heparin tube
TEMPERATURE	Maintain at room temperature. Send to the Lab ASAP
SETUP	Saturday to Thursday
TURN AROUND TIME	7-10 working days

Peripheral Blood-Breakage Study (Fanconi Anaemia) Chromosomal Analysis

CODE GT008

This analysis evaluates the amount of chromosome breakage in cells and will not identify any specific mutation in the DNA. It will reliably detect affected individuals but is not an appropriate test for unaffected carriers of the disorder. This test does not rule out numeric or structural chromosomal abnormalities. Some individuals with characteristics suggestive of Fanconi Anaemia (FA) may have normal results in the Diepoxybutane induced DEB chromosome breakage test, therefore, a negative DEB study does not rule out FA in all cases.

SPECIMEN REQUIREMENTS	3-5 ml Peripheral blood in Sodium Heparin tube
TEMPERATURE	Maintain at room temperature. Send to the Lab ASAP
SETUP	Saturday to Thursday
TURN AROUND TIME	7-10 working days

Peripheral Blood- Unstimulated, Hematological Disorder, Chromosomal Analysis

CODE GT009

Cytogenetic analysis of peripheral blood (if 10% or more blasts present) can identify numerical and structural chromosomal aberrations that are diagnostic and/or prognostic for some types of leukemia and lymphoma. Chromosome analysis is often employed for staging, monitoring treatment and predicting relapse. Leukemias and lymphomas can be distinguished by specific chromosome abnormalities that can aid in precise diagnosis, disease etiology, patient prognosis and disease management.

SPECIMEN REQUIREMENTS	3-5 ml Peripheral blood in Sodium Heparin tube
TEMPERATURE	Maintain at room temperature. Send to the Lab ASAP
SETUP	Saturday to Thursday
TURN AROUND TIME	7-10 working days

Bone Marrow, Hematological Disorders, Chromosomal Analysis

CODE GT010

Cytogenetics analysis of hematologic disorders attempts to define and interpret chromosomal aberrations that occur in neoplastic cells associated with leukemia, lymphoma and other hematologic malignancies. Chromosome abnormalities in cancer cells of patients with malignant hematologic disorders including acute and chronic myeloid and lymphoid leukemias, myelodysplastic and myeloproliferative disorders, lymphomas and unexplained anemias may correlate with the diagnosis, prognosis, treatment and etiology of disease.

SPECIMEN REQUIREMENTS	Minimum 2 ml of Bone marrow in Sodium Heparin tube
TEMPERATURE	Maintain at room temperature. Send to the Lab ASAP
SETUP	Saturday to Thursday
TURN AROUND TIME	10-14 working days

Solid Tumor Chromosomal Analysis

CODE GT011

Many solid tumors, especially soft tissue sarcomas, have precise structural chromosomal abnormalities that are important for precise diagnosis. Cancer cytogenetic studies can identify numerical and structural chromosomal abnormalities that are diagnostic and/or prognostic for various types of solid tumors.

SPECIMEN REQUIREMENTS	Collect viable and non-necrotic tissue measuring 0.5cm or 1-5 g in sterile conical tubes filled with sterile transport media
TEMPERATURE	Maintain at room temperature. Send to the Lab ASAP
SETUP	Saturday to Thursday
TURN AROUND TIME	14 – 21 working days

Molecular Cytogenetics- Flourecence in Situ Hybridization (FISH)

Amniotic Fluid Prenatal Chromosomes 13, 18, 21, X, Y Aneuploidy Detection by Rapid FISH

CODE GT012

This test allows for the detection of aneuploidy for chromosomes 13, 18, 21, X & Y, via (FISH) assay in interphase cells from prenatal samples. In addition, it can rule out Patau Syndrome, Edward Syndrome, Down syndrome and determine the sex of the fetus. All FISH analysis can be confirmed by routine cytogenetic analysis if requested.

SPECIMEN REQUIREMENTS	Use sterile techniques to transfer 30 ml in 3 different sterile plastic conical tube for transport
TEMPERATURE	Maintain at room temperature. Send to the Lab ASAP
SETUP	Saturday to Thursday
TURN AROUND TIME	3-5 working days

Amniotic Fluid Prenatal Chromosome X/Y/18 Aneuploidy Screening & Sexing by Rapid FISH

CODE GT013

Detection of aneuploidy for chromosomes 18 to rule out Edward Syndrome and sex related disorders by screening sex chromosome X & Y, via (FISH) assay in interphase cells from prenatal samples. All FISH analysis can be confirmed by routine cytogenetic analysis if requested.

SPECIMEN REQUIREMENTS	Use sterile techniques to transfer 30 ml in 3 different sterile plastic conical tube for transport
TEMPERATURE	Maintain at room temperature. Send to the Lab ASAP
SETUP	Saturday to Thursday
TURN AROUND TIME	3-5 working days

Amniotic Fluid Prenatal Chromosomes X/Y Aneuploidy Screening & Sexing by Rapid FISH

CODE GT014

Detection of aneuploidy for sex chromosome X & Y, via fluorescence in situ hybridization (FISH) assay in interphase cells from prenatal samples. All FISH analysis can be confirmed by routine cytogenetic analysis if requested.

SPECIMEN REQUIREMENTS	Use sterile techniques to transfer 30 ml in 3 different sterile plastic conical tube for transport
TEMPERATURE	Maintain at room temperature. Send to the Lab ASAP
SETUP	Saturday to Thursday
TURN AROUND TIME	3-5 working days

Postnatal Blood - Common Chromosomal Aneuploidy (13, 18, 21, X, Y)

CODE GT015

Detection of aneuploidy for chromosomes 13, 18, 21, X & Y, via (FISH) assay in interphase cells from postnatal samples. To rule out Patau Syndrome, Edward Syndrome, Down syndrome and determine the sex of the fetus. All FISH analysis can be confirmed by routine cytogenetic analysis if requested.

SPECIMEN REQUIREMENTS	3-5 ml Peripheral blood in Sodium Heparin tube
TEMPERATURE	Maintain at room temperature. Send to the Lab ASAP
SETUP	Saturday to Thursday
TURN AROUND TIME	3-5 working days

Postnatal Blood - Sex Chromosome Aneuploidy X/Y

CODE GT016

Detection of aneuploidy for sex chromosome X & Y, via (FISH) assay in interphase cells from postnatal samples. All FISH analysis can be confirmed by routine cytogenetic analysis if requested.

SPECIMEN REQUIREMENTS	3-5 ml Peripheral blood in Sodium Heparin tube
TEMPERATURE	Maintain at room temperature. Send to the Lab ASAP
SETUP	Saturday to Thursday
TURN AROUND TIME	3-5 working days

MICRODELETION SYNDROMES:

1p36 Deletion Syndrome

CODE GT017

Chromosome 1p microdeletion syndrome is associated with a spectrum of dysmorphic features and mental retardation. The syndrome can be suspected in overweight patients with mental retardation, heart defects, and finger abnormalities. Facial features include micro-cephaly (small head), short neck, malformed ears, and small deep-set eyes. The phenotype is variable and depends on the size of the deletion.

SPECIMEN REQUIREMENTS	3-5 ml Peripheral blood in Sodium Heparin tube
TEMPERATURE	Maintain at room temperature. Send to the Lab ASAP
SETUP	Saturday to Thursday
TURN AROUND TIME	7-10 working days

Cri-Du-Chat Syndrome (5p15.2)

CODE GT018

Individuals affected with Cri-du-chat are characterized by dysmorphic facial features, microcephaly, growth deficiency, mental retardation, speech delay and a characteristic "cat-like" cry due to a deletion on chromosome 5p15.2.

SPECIMEN REQUIREMENTS	3-5 ml Peripheral blood in Sodium Heparin tube
TEMPERATURE	Maintain at room temperature. Send to the Lab ASAP
SETUP	Saturday to Thursday
TURN AROUND TIME	7-10 working days

DiGeorge/ Velo-Cardio-Facial Micro Deletion Syndrome (22q11)

CODE GT019

DiGeorge and Velocardiofacial syndromes (VCFS) are caused by 22q11 deletions. Fetus with congenital heart defect on fetal ultrasound. Individuals affected with DiGeorge syndrome are characterized by congenital heart defects, an absent or hypoplastic thymus, hypocalcemia, cleft lip and/or palate, microcephaly, immune deficiency, renal anomalies, psychiatric problems and learning difficulties.

SPECIMEN REQUIREMENTS	3-5 ml Peripheral blood in Sodium Heparin tube
TEMPERATURE	Maintain at room temperature. Send to the Lab ASAP
SETUP	Saturday to Thursday
TURN AROUND TIME	7-10 working days

Kallmann Syndrome/(STS) (Xp22.3)

CODE GT020

Kallmann syndrome 1 consists of congenital, isolated, idiopathic hypogonadotropic hypogonadism and anosmia

SPECIMEN REQUIREMENTS	3-5 ml Peripheral blood in Sodium Heparin tube
TEMPERATURE	Maintain at room temperature. Send to the Lab ASAP
SETUP	Saturday to Thursday
TURN AROUND TIME	7-10 working days

Miller-Dieker Syndrome (17p13.3)

CODE GT021

Individuals affected with Miller-Dieker syndrome are characterized by lissencephaly, mental retardation and a distinct facial appearance. The test is used to detect microdeletion of chromosome 17p13.3.

SPECIMEN REQUIREMENTS	3-5 ml Peripheral blood in Sodium Heparin tube
TEMPERATURE	Maintain at room temperature. Send to the Lab ASAP
SETUP	Saturday to Thursday
TURN AROUND TIME	7-10 working days

Prader-willi/ Angelmen Syndrome (15q-q13)

CODE GT022

Diagnostic criteria for PWS may include: hypotonia, failure to thrive, rapid weight gain between 12 months and 6 years, characteristic facial features, hypogonadism and mild to moderate mental retardation. Diagnostic criteria for Angelman Syndrome may include feeding problems in the first few months of life, developmental delay, seizures, microcephaly, movement and balance disorders, hand-flapping, frequent outbursts of inappropriate laughter. The test is used to detect microdeletion of chromosome 15q11-q13.

SPECIMEN REQUIREMENTS	3-5 ml Peripheral blood in Sodium Heparin tube
TEMPERATURE	Maintain at room temperature. Send to the Lab ASAP
SETUP	Saturday to Thursday
TURN AROUND TIME	7-10 working days

Smith Magenis Syndrome (17p11.2)

CODE GT023

Smith-Magenis syndrome in children and adults children is characterized by a flattened mid-face, down-turned mouth, hypotonia, short, broad hands, mental retardation, chronic sleep disturbance and self-injurious behavior. Individuals affected with Smith-Magenis syndrome have a normal life expectancy.

SPECIMEN REQUIREMENTS	3-5 ml Peripheral blood in Sodium Heparin tube
TEMPERATURE	Maintain at room temperature. Send to the Lab ASAP
SETUP	Saturday to Thursday
TURN AROUND TIME	7-10 working days

SRY (Yp11.3)

CODE GT024

This test is to detect microdeletions of chromosome Yp11.3 associated with several disorders of sex developments/ sex reversal disease.

SPECIMEN REQUIREMENTS	3-5 ml Peripheral blood in Sodium Heparin tube
TEMPERATURE	Maintain at room temperature. Send to the Lab ASAP
SETUP	Saturday to Thursday
TURN AROUND TIME	7-10 working days

Williams Syndrome (7q11.23)

CODE GT025

Williams's syndrome is a genetic disorder characterized by "elfin" facial features, mental retardation, growth deficiency, hypercalcemia and cardiovascular disease.

SPECIMEN REQUIREMENTS	3-5 ml Peripheral blood in Sodium Heparin tube
TEMPERATURE	Maintain at room temperature. Send to the Lab ASAP
SETUP	Saturday to Thursday
TURN AROUND TIME	7-10 working days

Wolf-Hirsch Horn Syndrome (4p16.2)

CODE GT026

Individuals with Wolf-Hirschhorn syndrome are characterized by microcephaly, growth deficiency, mental retardation and characteristic facial features.

SPECIMEN REQUIREMENTS	3-5 ml Peripheral blood in Sodium Heparin tube
TEMPERATURE	Maintain at room temperature. Send to the Lab ASAP
SETUP	Saturday to Thursday
TURN AROUND TIME	7-10 working days

HEMATOLOGY AND ONCOLOGY:

AML/ETO Translocations t(21q22.12;8q21.3)

CODE GT027

The (8;21) translocation is the most frequently observed karyotypic abnormality associated with Acute Myeloid Leukemia (AML), subtype M2. Patients with this type of AML have a high remission rate with conventional chemotherapy.

SPECIMEN REQUIREMENTS	3-5 ml Peripheral blood and minimum 2 ml of Bone marrow in Sodium Heparin tube
TEMPERATURE	Maintain at room temperature. Send to the Lab ASAP
SETUP	Saturday to Thursday
TURN AROUND TIME	3-6 days for uncultured specimen, 10-14 working days for cultured specimen

BCL6 (3q27.3)

CODE GT028

For the detection of translocations involving the chromosomal region 3q27.3 harboring the BCL6 gene. Chromosomal rearrangements of the BCL6 gene region were found to occur in different types of non-Hodgkin lymphoma (NHL), including diffuse large B-cell lymphoma (DLBCL) and follicular lymphoma (FL). DLBCL which are positive for both BCL6 and MYC rearrangements have been shown to have an extremely poor prognosis.

SPECIMEN REQUIREMENTS	• 3-5 ml Peripheral blood and minimum 2 ml of Bone marrow in Sodium Heparin tube • FFPE: Formalin-Fixed Paraffin-embedded tissue (4-5 µm thickness): sterile conditions/or container.
TEMPERATURE	Maintain at room temperature. Send to the Lab ASAP
SETUP	Saturday to Thursday
TURN AROUND TIME	3-6 days for uncultured specimen, 10-14 working days for cultured specimen and paraffin slides

BCR/ABL Translocation t(9;22)(q34;q11.2)

CODE GT029

The test is designed for the detection of the specific translocations involving the chromosomal region 9q34.12 harboring the ABL1 gene, and the chromosomal region 22q11.23, harboring the BCR gene. Rearrangements involving t(9;22)(q34.1;q11.2) are observed in approx. 90% of patients with chronic myeloid leukemia (CML) and in approx. 25% of adults with acute lymphoblastic leukemia (ALL).

SPECIMEN REQUIREMENTS	3-5 ml Peripheral blood and minimum 2 ml of Bone marrow in Sodium Heparin tube
TEMPERATURE	Maintain at room temperature. Send to the Lab ASAP
SETUP	Saturday to Thursday
TURN AROUND TIME	3-6 days for uncultured specimen, 10-14 working days for cultured specimen

CBFB Break Apart (16p13.11/16q22.1)

CODE GT030

CBFB encodes the beta subunit of the CBFA/CBFB transcription factor complex involved in myeloid differentiation. The chromosomal aberrations inv(16) (p13.1q22.1) and the related translocation t(16;16)(p13.1;q22.1), which have been detected in about 10% of patients with AML (acute myeloblastic leukemia), lead to the fusion of the CBFB gene with the MYH11 (smooth muscle myosin heavy chain) gene on 16p13.1. The resulting CBFB-MYH11 fusion gene is involved in leukemic transformation. AML patients with these genetic rearrangements have a favorable prognosis. Inv(16) may sometimes be difficult to identify using conventional cytogenetic analysis.

SPECIMEN REQUIREMENTS	3-5 ml Peripheral blood and minimum 2 ml of Bone marrow in Sodium Heparin tube
TEMPERATURE	Maintain at room temperature. Send to the Lab ASAP
SETUP	Saturday to Thursday
TURN AROUND TIME	3-6 days for uncultured specimen, 10-14 working days for cultured specimen

IgH (14q32.33)

CODE GT031

The IgH (14q32) Break Apart probe is designed to detect chromosomal rearrangements involving the immunoglobulin heavy chain (IGH) gene on chromosome 14q32. To date, 43 different IGH chromosomal gene arrangement pairings have been identified.
Multiple Myeloma (MM) and Non-Hodgkins lymphomas (NHL) are both characterized by recurrent chromosomal gene arrangements involving the IGH gene1, 2. The incidence of IGH gene rearrangements have been linked to MM pathogenesis and contribute to both the clinical and morphological features associated with NHL subtypes1,2.

SPECIMEN REQUIREMENTS	• 3-5 ml Peripheral blood and minimum 2 ml of Bone marrow in Sodium Heparin tube • FFPE: Formalin-Fixed Paraffin-embedded tissue (4-5µm thickness): sterile conditions/or container.
TEMPERATURE	Maintain at room temperature. Send to the Lab ASAP
SETUP	Saturday to Thursday
TURN AROUND TIME	3-6 days for uncultured specimen, 10-14 working days for cultured specimen and paraffin slides

c-MYC Rearrangements (8q24.21)

CODE GT032

The MYC (8q24) Break Apart FISH probe is designed to detect chromosomal rearrangements involving the MYC gene on Chromosome 8q24. The MYC gene belongs to a family of transcription factors and under normal conditions it controls cell cycle progression1. However in multiple cancer indications, MYC is considered a Proto-oncogene1. MYC gene deregulation is identified in multiple malignancies such as Burkitt's lymphoma, diffuse large B-cell lymphoma, and B-cell lymphoma. MYC gene amplifications, rearrangements, and/or point mutations are considered the underlining mechanisms that induce MYC gene deregulation. Specifically, a MYC gene rearrangement is considered a prognostic marker in several cancer subtypes.

SPECIMEN REQUIREMENTS	• 3-5 ml Peripheral blood and minimum 2 ml of Bone marrow in Sodium Heparin tube • FFPE: Formalin-Fixed Paraffin-embedded tissue (4-5µm thickness): sterile conditions/or container.
TEMPERATURE	Maintain at room temperature. Send to the Lab ASAP
SETUP	Saturday to Thursday
TURN AROUND TIME	3-6 days for uncultured specimen, 10-14 working days for cultured specimen and paraffin slides

Her2/neu Amplification

CODE GT033

The detection of HER-2/neu gene amplification by FISH analysis is linked with rapid cancer cell proliferation, decreased disease-free survival and poor overall survival in both node-negative and node-positive ductal breast cancers. In patients with advanced breast carcinoma, HER-2 amplification predicts responsiveness to transtuzumab (Herceptin®) therapy and poor response to standard chemotherapy.

SPECIMEN REQUIREMENTS	FFPE: Formalin-Fixed Paraffin-embedded tissue (4-5µm thickness): sterile conditions/or container.
TEMPERATURE	Maintain at room temperature. Send to the Lab ASAP
SETUP	Saturday to Thursday
TURN AROUND TIME	10-14 working days

Myelodysplastic Syndrome 5q-/-5, 7q-/-7

CODE GT034

To detect chromosomal abnormalities on chromosome 5 and 7, to help diagnose and classify MDS, guide treatment, and evaluate prognosis.

SPECIMEN REQUIREMENTS	• 3-5 ml Peripheral blood and minimum 2 ml of Bone marrow in Sodium Heparin tube • FFPE: Formalin-Fixed Paraffin-embedded tissue (4-5µm thickness): sterile conditions/or container.
TEMPERATURE	Maintain at room temperature. Send to the Lab ASAP
SETUP	Saturday to Thursday
TURN AROUND TIME	3-6 days for uncultured specimen, 10-14 working days for cultured specimen

PML/RARA Translocation 15q24.1/17q21.1-q21.1

CODE GT035

It is used to help diagnose acute promyelocytic leukemia (APL) in which the PML-RARA gene sequence is present, to guide treatment, to monitor response to treatment, and to monitor for disease recurrence. (FISH) may be used to help diagnose APL and/or help to determine the percentage of a person's blood or bone marrow cells that contain the abnormal, fused PML-RARA gene. This test method uses fluorescent dye-labeled probes to "light up" the PML-RARA gene sequence when it is present. FISH can also be used to detect the variant translocations involving RARA and genes other than PML. This may help identify drug-resistant (ATRA-resistant) rearrangements.

SPECIMEN REQUIREMENTS	3-5 ml Peripheral blood and minimum 2 ml of Bone marrow in Sodium Heparin tube
TEMPERATURE	Maintain at room temperature. Send to the Lab ASAP
SETUP	Saturday to Thursday
TURN AROUND TIME	3-6 days for uncultured specimen, 10-14 working days for cultured specimen

TP53 Aberrations (17p13.1)

CODE GT036

This TP53 FISH probe localizes to the chromosome region 17p13 allowing it to detect chromosomal aberrations to the TP53 gene. Somatic mutation of the TP53 tumor suppressor gene is the most common genetic alteration seen in human cancers. Mutations in TP53 gene usually correlate with poor outcome and early recurrence in cancer.

SPECIMEN REQUIREMENTS	• 3-5 ml Peripheral blood and minimum 2 ml of Bone marrow in Sodium Heparin tube • FFPE: Formalin-Fixed Paraffin-embedded tissue (4-5µm thickness): sterile conditions/or container
TEMPERATURE	Maintain at room temperature. Send to the Lab ASAP
SETUP	Saturday to Thursday
TURN AROUND TIME	3-6 days for uncultured specimen, 10-14 working days for cultured specimen and paraffin slides

FOR VALIDATION OF CONVENTIONAL CYTOGENETIC ANALYSIS:

Arm-Specific Painting Probe

CODE GT037

Used for validation of chromosomal abnormalities detected by conventional cytogenetic

SPECIMEN REQUIREMENTS	3-5 ml Peripheral blood in Sodium Heparin tube
TEMPERATURE	Maintain at room temperature. Send to the Lab ASAP
SETUP	Saturday to Thursday
TURN AROUND TIME	7-10 working days

Centromeric Probes for Chromosomes 13, 18, 21, X and Y

CODE GT038

Used to rule out common aneuploidy on cultured specimen.

SPECIMEN REQUIREMENTS	3-5 ml Peripheral blood in Sodium Heparin tube
TEMPERATURE	Maintain at room temperature. Send to the Lab ASAP
SETUP	Saturday to Thursday
TURN AROUND TIME	7-10 working days

Telomeric Probe Analysis

CODE GT039

Submicroscopic abnormalities at the ends of chromosomes are a significant cause of idiopathic mental retardation. Indications for sub-telomere analysis include individuals with moderate to severe mental retardation of unknown etiology or parents of a child found to have subtelomere abnormalities. Subtelomere analysis also has application in the investigation of recurrent miscarriage and autistic disorders.

SPECIMEN REQUIREMENTS	3-5 ml Peripheral blood in Sodium Heparin tube
TEMPERATURE	Maintain at room temperature. Send to the Lab ASAP
SETUP	Saturday to Thursday
TURN AROUND TIME	7-10 working days

Whole Chromosome Painting

CODE GT040

Used for validation of chromosomal abnormalities detected by conventional cytogenetic.

SPECIMEN REQUIREMENTS	3-5 ml Peripheral blood in Sodium Heparin tube
TEMPERATURE	Maintain at room temperature. Send to the Lab ASAP
SETUP	Saturday to Thursday
TURN AROUND TIME	7-10 working days

Chromosomal Microarray Analysis

Hematologic Disorders and Tumor (Fresh or Frozen) Chromosomal Microarray

CODE GT041

Detection and characterization of clonal copy number imbalance and loss of heterozygosity associated with malignancies. Assisting in the diagnosis and classification of certain cancer type. Evaluating the prognosis for patients with certain malignancies. Genomic characterization of tumor for copy number imbalances and loss of heterozygosity.

For hemato-oncology this test is used for the detection of aneuploidy in different diseases including: CLL, Low risk MDS, Hypoplastic MDS, Aplastic Anemia and Myeloma

SPECIMEN REQUIREMENTS	• 3-5 ml Peripheral blood and minimum 2 ml of Bone marrow in EDTA tube • FFPE: Formalin-Fixed Paraffin-embedded tissue (4-5µm thickness): sterile conditions/or container
TEMPERATURE	Maintain at room temperature. Send to the Lab ASAP
SETUP	Saturday to Thursday
TURN AROUND TIME	10-14 working days

Constitutional (Postnatal) Chromosomal Microarray

CODE GT042

This is the highest diagnostic yield of any single clinically available test for children with global developmental delay, intellectual disability, autism spectrum disorders, and multiple congenital anomalies. High resolution detection of chromosome segments involved in deletions, duplications, and long continuous stretches of homozygosity. The results will assist in clinical management and genetic counseling.

SPECIMEN REQUIREMENTS	3-5 ml Peripheral blood in EDTA tube
TEMPERATURE	Maintain at room temperature. Send to the Lab ASAP
SETUP	Saturday to Thursday
TURN AROUND TIME	10-14 working days

Family Screening Chromosomal Microarray

CODE GT043

This is the highest diagnostic yield of any single clinically available test for children with global developmental delay, intellectual disability, autism spectrum disorders, and multiple congenital anomalies. Investigation of inheritance pattern is determined by screening parent samples. The test involves high-resolution detection of chromosome segments involved in deletions, duplications, and long continuous stretches of homozygosity. The result will assist in clinical management and genetic counseling.

SPECIMEN REQUIREMENTS	3-5 ml Peripheral blood in EDTA tube
TEMPERATURE	Maintain at room temperature. Send to the Lab ASAP
SETUP	Saturday to Thursday
TURN AROUND TIME	10-14 working days

Molecular Genetics Analysis

AML/ETO t(8;21) Qualitative Analysis

CODE GT044

Acute myelogenous leukemia (AML) with t(8;21)(q22;q22) is an acute myelogenous leukemia generally showing maturation in the neutrophil lineage. The test Detects AML/ETO fusions arising from t(8;21) in acute myeloid leukemia (AML). Clinical uses of the test are: diagnose acute myeloid leukemia (AML) with t(8;21) chromosome translocation, monitor treatment response, monitor minimal residual disease (MRD) and predict early relapse.

SPECIMEN REQUIREMENTS	3-5 ml Peripheral blood and minimum 2 ml of Bone marrow in EDTA tub
TEMPERATURE	Maintain at room temperature. Send to the Lab ASAP
SETUP	Saturday to Thursday
TURN AROUND TIME	5-7 working days

AML/ETO t(8;21) Quantitative Analysis

CODE GT045

Acute myelogenous leukemia (AML) with t(8;21)(q22;q22) is an acute myelogenous leukemia generally showing maturation in the neutrophil lineage. The test Detects and quantitate AML/ETO fusions arising from t(8;21) in acute myeloid leukemia (AML). Clinical uses of the test are: diagnose acute myeloid leukemia (AML) with t(8;21) chromosome translocation, monitor treatment response, monitor minimal residual disease (MRD) and predict early relapse.

SPECIMEN REQUIREMENTS	3-5 ml Peripheral blood and minimum 2 ml of Bone marrow in EDTA tube
TEMPERATURE	Maintain at room temperature. Send to Lab ASAP
SETUP	Saturday to Thursday
TURN AROUND TIME	10-14 working days

Argininosuccinate Lyase (ASL) Mutation Analysis

CODE GT046

The urea cycle consists of six consecutive enzymatic reactions that convert waste nitrogen into urea. Deficiencies of any of these enzymes of the cycle result in urea cycle disorders (UCDs). Argininosuccinate lyase (ASL) catalyzes the fourth reaction in this cycle, resulting in the breakdown of argininosuccinic acid to arginine and fumarate. ASL deficiency (ASL) is the second most common UCD.

SPECIMEN REQUIREMENTS	3-5 ml Peripheral blood in EDTA tube
TEMPERATURE	Maintain at room temperature. Send to Lab ASAP
SETUP	Saturday to Thursday
TURN AROUND TIME	5-7 working days

BCR/ABL t(9;22) Qualitative Analysis

CODE GT047

Detects the presence of cells in blood or bone marrow containing the BCR/ABL fusions arising from t(9;22). Major and minor Philadelphia Chromosome translocations are associated with CML, AML and ALL.

SPECIMEN REQUIREMENTS	3-5 ml Peripheral blood and minimum 2 ml of Bone marrow in EDTA tube
TEMPERATURE	Maintain at room temperature. Send to Lab ASAP
SETUP	Saturday to Thursday
TURN AROUND TIME	5-7 working days

BCR/ABL t(9;22) Quantitative Analysis

CODE GT048

Detects and quantitate the presence of cells in blood or bone marrow containing the BCR/ABL fusions arising from t(9;22). Major and minor Philadelphia Chromosome translocations are associated with CML, AML and ALL. This technique can be used to quantitatively monitor minimal residual disease.

SPECIMEN REQUIREMENTS	3-5 ml Peripheral blood and minimum 2 ml of Bone marrow in EDTA tube
TEMPERATURE	Maintain at room temperature. Send to Lab ASAP
SETUP	Saturday to Thursday
TURN AROUND TIME	10-14 working days

Beta Thalassemia, HBB Gene Mutation Analysis

CODE GT049

There are currently over 800 hemoglobin variants catalogued, of which approximately 500 are due to mutations in the beta-globin gene. In addition, approximately 95% of beta-thalassemia is caused by point mutations. This test, which sequences the coding regions and introns of the beta-globin gene (HBB) in both directions, identifies hemoglobin variants that are not easily diagnosed by electrophoresis/HPLC and can determine the cause of beta-thalassemia and sickle cell anemia. It can also be used to determine carrier states.

SPECIMEN REQUIREMENTS	3-5 ml Peripheral blood in EDTA tube
TEMPERATURE	Maintain at room temperature. Send to Lab ASAP
SETUP	Saturday to Thursday
TURN AROUND TIME	7-10 working days

Blood Coagulation and Cardiovascular Disease (Multigene Panel)

CODE GT050

Genes included: KLKB1, KNG1, coagulation factor XI(F11), coagulation factor IX(F9), coagulation factor VIII (F8), coagulation factor III, tissue factor (F7), coagulation factor X(F10), coagulation factor V (F5), coagulation factor II, thrombin (F2), fibrinogen alpha chain 9(FGA) FGG, coagulation factor XIII A chain(F13B), THBD, protein C, inactivator of coagulation factors Va and VIIIa (PROC), PROS1, plasminogen activator, tissue type (PLAT), PLG, serpin, SERPIND1, CPB2, and SERPINC1.

SPECIMEN REQUIREMENTS	• 3-5 ml Peripheral blood in EDTA tube • For additional samples requirements, see Appendix
TEMPERATURE	Maintain at room temperature. Send to Lab ASAP
SETUP	Saturday to Thursday
TURN AROUND TIME	Up to 8 weeks

BRCA1 and BRCA2 Germline Mutation Analysis

CODE GT051

This test is useful for determining the degree of risk for breast cancer particularly in the presence of positive family history for the disease. This test is also indicated prior to prescribing PARP inhibitor treatment for breast and ovarian cancer.

SPECIMEN REQUIREMENTS	3-5 ml Peripheral blood in EDTA tube
TEMPERATURE	Maintain at room temperature. Send to Lab ASAP
SETUP	Saturday to Thursday
TURN AROUND TIME	Up to 6 weeks

BRCA1 and BRCA2 Somatic Mutation Analysis

CODE GT052

This test is used to identify tumour-specific BRCA1 and BRCA2 mutations the presence of which can render breast or ovarian tumours sensitive to PARP inhibitor treatments.

SPECIMEN REQUIREMENTS	• 3-5 ml Peripheral blood in EDTA tube • FFPE: Formalin-Fixed Paraffin-embedded tissue (4-5µm thickness): sterile conditions/or container.
TEMPERATURE	Maintain at room temperature. Send to Lab ASAP
SETUP	Saturday to Thursday
TURN AROUND TIME	21 working days

B-Raf Proto-Oncogene, Serine/Threonine Kinase (BRAF- V600E) Mutation Analysis

CODE GT053

The most common BRAF mutation, V600E (c.1799T>A) is particularly prevalent in malignant melanoma, papillary thyroid and colorectal cancers. In addition, patients with Langerhans cell histiocytosis commonly have this BRAF V600E mutation.

SPECIMEN REQUIREMENTS	• 3-5 ml Peripheral blood and minimum 2 ml of Bone marrow in EDTA tube • FFPE: Formalin-Fixed Paraffin-embedded tissue (4-5µm thickness): sterile conditions/or container.
TEMPERATURE	Maintain at room temperature. Send to Lab ASAP
SETUP	Saturday to Thursday
TURN AROUND TIME	7-10 working days

Calreticulin (CALR -Exon 8 and Exon 9) Mutation Analysis

CODE GT054

The CALR mutation test is used to help diagnose and classify bone marrow disorders that lead to the production of too many blood cells. These disorders are known as myeloproliferative neoplasms (MPNs).A positive CALR mutation test means that the person likely has a MPN, specifically essential thrombocythemia (ET) or primary myelofibrosis (PMF).

SPECIMEN REQUIREMENTS	3-5 ml Peripheral blood and minimum 2 ml of Bone marrow in EDTA tube
TEMPERATURE	Maintain at room temperature. Send to Lab ASAP
SETUP	Saturday to Thursday
TURN AROUND TIME	7-10 working days

CBFB/MYH11 inv(16)/t(16;16) Qualitative Analysis

CODE GT055

This test is for detection of CBFB-MYH11 fusion transcripts resulting from inv(16)/t(16;16) in patients with acute leukemia specifically Acute Myeloid Leukemia (AML).

SPECIMEN REQUIREMENTS	3-5 ml Peripheral blood and minimum 2 ml of Bone marrow in EDTA tube
TEMPERATURE	Maintain at room temperature. Send to Lab ASAP
SETUP	Saturday to Thursday
TURN AROUND TIME	10-14 working days

CBFB/MYH11 inv(16)/t(16;16) Quantitative analysis

CODE GT056

This test is for detection and quantification of CBFB-MYH11 fusion transcripts resulting from inv(16)/t(16;16) in patients with acute leukemia specifically Acute Myeloid Leukemia (AML).

SPECIMEN REQUIREMENTS	3-5 ml Peripheral blood and minimum 2 ml of Bone marrow in EDTA tube
TEMPERATURE	Maintain at room temperature. Send to Lab ASAP
SETUP	Saturday to Thursday
TURN AROUND TIME	10-14 working days

Clinical Exome Sequencing

CODE GT057

Exome analysis is indicated when a clear clinical diagnosis cannot be ascertained or the gene(s) responsible for the disease are overlapping in function and nature. The results are useful for providing clear-cut diagnosis and therefore better clinical management of the disease. The information obtained can be utilized for the provision of better genetic counselling.

SPECIMEN REQUIREMENTS	• 3-5 ml Peripheral blood in EDTA tube • For additional samples requirements, see Appendix
TEMPERATURE	Maintain at room temperature. Send to Lab ASAP
SETUP	Saturday to Thursday
TURN AROUND TIME	Up to 8 weeks

Cystic Fibrosis (CFTR) Mutation Analysis

CODE GT058

Cystic fibrosis is an inherited disease that affects mainly the lungs, pancreas, and sweat glands. It leads to the production of thick, sticky mucus and can cause recurrent respiratory infections and impaired function of the pancreas. The CF gene mutations screening detects the common mutations in the cystic fibrosis transmembrane conductance regulator (CFTR) gene on chromosome 7.

SPECIMEN REQUIREMENTS	3-5 ml Peripheral blood in EDTA tube
TEMPERATURE	Maintain at room temperature. Send to Lab ASAP
SETUP	Saturday to Thursday
TURN AROUND TIME	7-10 working days

DNA Methyltransferase 3 Alpha (DNMT3A) Mutation Analysis

CODE GT059

The DNMT3A gene provides instructions for making an enzyme called DNA methyltransferase 3 alpha. This enzyme is involved in DNA methylation. DNA methylation is important in many cellular functions. These include determining whether the instructions in a particular segment of DNA are carried out or suppressed (gene silencing), regulating reactions involving proteins and fats, and controlling the processing of chemicals that relay signals in the nervous system (neurotransmitters). DNA methyltransferase 3 alpha is particularly important for establishing DNA methylation patterns during development before birth. Mutations in the DNMT3A gene are associated with a form of blood cancer known as cytogenetically normal acute myeloid leukemia (CN-AML).

SPECIMEN REQUIREMENTS	3-5 ml Peripheral blood and minimum 2 ml of Bone marrow in EDTA tube
TEMPERATURE	Maintain at room temperature. Send to Lab ASAP
SETUP	Saturday to Thursday
TURN AROUND TIME	7-10 working days

Duchenne / Becker Muscular Dystrophy (DMD)

CODE GT060

Duchenne muscular dystrophy (DMD) is a severe X-linked recessive neuromuscular disorder. The molecular defect in most patients is deletion within the dystrophin gene, and duplications as well. Becker muscular dystrophy (BMD) is a milder allelic form, characterized by primarily deletion and duplication.

SPECIMEN REQUIREMENTS	3-5 ml Peripheral blood in EDTA tube
TEMPERATURE	Maintain at room temperature. Send to Lab ASAP
SETUP	Saturday to Thursday
TURN AROUND TIME	7-10 working days

Factor II Prothrombin (G20210A) Mutation Analysis

CODE GT061

Thrombophilia is a multifactorial disease due to the interaction of genetic and environmental risk factors. Some of the recently discovered genetic risk factors, such as factor Factor V Leiden (R506Q), Prothrombin (G20210A) and MTHFR (C677T) are common in the population. Genetic testing of thrombophilic disorders can help in determining the duration of anticoagulant therapy, risk stratification for primary or secondary prophylaxis and genetic counselling.

SPECIMEN REQUIREMENTS	3-5 ml Peripheral blood in EDTA tube
TEMPERATURE	Maintain at room temperature. Send to Lab ASAP
SETUP	Saturday to Thursday
TURN AROUND TIME	7-10 working days

Factor V (F5)-Leiden (R506Q) Mutation Analysis

CODE GT062

Thrombophilia is a multifactorial disease due to the interaction of genetic and environmental risk factors. Some of the recently discovered genetic risk factors, such as factor Factor V Leiden (R506Q), Prothrombin (G20210A) and MTHFR (C677T) are common in the population. Genetic testing of thrombophilic disorders can help in determining the duration of anticoagulant therapy, risk stratification for primary or secondary prophylaxis and genetic counselling.

SPECIMEN REQUIREMENTS	3-5 ml Peripheral blood in EDTA tube
TEMPERATURE	Maintain at room temperature. Send to Lab ASAP
SETUP	Saturday to Thursday
TURN AROUND TIME	7-10 working days

Familia Hemophagocytic Lymphohistiocytosis,Type 5, STXBP2 Gene

CODE GT063

It is a disorder in which the immune system produces too many activated immune cells (lymphocytes) called T cells, natural killer cells, B cells, and macrophages (histiocytes). Excessive amounts of immune system proteins called cytokines are also produced. This overactivation of the immune system causes fever and damages the liver and spleen, resulting in enlargement of these organs.

SPECIMEN REQUIREMENTS	3-5 ml Peripheral blood in EDTA tube
TEMPERATURE	Maintain at room temperature. Send to Lab ASAP
SETUP	Saturday to Thursday
TURN AROUND TIME	7-10 working days

Familial Cancer Genetic Screening by NGS

CODE GT064

Genes included: ABL1, AKT1, ALK, APC, ATM, BMPR1A, BRAF, BRCA1, BRCA2, BRIP1, CDK4, CDKN2A, CHEK2, CSF1R, CTNNB1, EGFR, ERBB2, ERBB4, ERCC6, EZH2, FBXW7, FGFR1, FGFR2, FGFR3, EZH2, FBXW7, EZH2, FBXW7, FGFR1, FGFR2, FGFR3, FLT3, G protein, GNA11, G protein, GNAQ, GNAS, HNF1A, HRAS, IDH1, IDH2 JAK2, JAK3, KDR, KIT, KRAS, MEN1, MET, MLH1, MPL, MSH2, MSH6, MUTYH, NBN, ARTN, NOTCH1, NPAT, NPM1, NRAS, PALB2, PDGFRA, PIK3CA, PMS1, PMS2, PTEN, PTPN11, RAD51C, RAD51D, RET, SDHB, SDHD, SMAD4, SMARCB, SMO, SRC, STK11, SUFU, TP53, TSC1, TSC2, VHL, WT1. Familial breast, colon and brain cancers can be screened using this panel.

SPECIMEN REQUIREMENTS	• 3-5 ml Peripheral blood in EDTA tube • For additional samples requirements, see Appendix
TEMPERATURE	Maintain at room temperature. Send to Lab ASAP
SETUP	Saturday to Thursday
TURN AROUND TIME	14 - 21 working days

Fms Related Tyrosine Kinase 3 (FLT3 -ITD) Mutation Analysis

CODE GT065

A mutation in the FLT3 gene on chromosome 13 results from internal tandem duplications (ITD) in exons 14 and 15 of the juxta membrane portion of the gene and causes activation of the FLT3 protein. Approximately 20-30% of patients with acute myeloid leukemia have this mutation which has been associated with adverse prognosis.

SPECIMEN REQUIREMENTS	3-5 ml Peripheral blood and minimum 2 ml of Bone marrow in EDTA tube
TEMPERATURE	Maintain at room temperature. Send to Lab ASAP
SETUP	Saturday to Thursday
TURN AROUND TIME	7 - 10 working days

Fms Related Tyrosine Kinase 3 (FLT3 TKD -D835) Mutation Analysis

CODE GT066

This assay detects mutations in the FLT3 gene at codons D835 of the tyrosine kinase domain (FLT3 TKD). Evaluation for FLT3 point mutations in the TKD is indicated at diagnosis of acute myeloid leukemia and may provide prognostic information and direct treatment.

SPECIMEN REQUIREMENTS	3-5 ml Peripheral blood and minimum 2 ml of Bone marrow in EDTA tube
TEMPERATURE	Maintain at room temperature. Send to Lab ASAP
SETUP	Saturday to Thursday
TURN AROUND TIME	7-10 working days

Frank-Ter Haar Syndrome (SH3PXD2B)

CODE GT067

Frank-Ter Haar syndrome (FTHS) is an autosomal-recessive disorder characterized by skeletal, cardio-vascular, and eye abnormalities. The syndrome appears to be due to mutations in the SH3PXD2B gene on chromosome 5q35.

SPECIMEN REQUIREMENTS	3-5 ml Peripheral blood in EDTA tube
TEMPERATURE	Maintain at room temperature. Send to Lab ASAP
SETUP	Saturday to Thursday
TURN AROUND TIME	7-10 working days

Glucose 6 Phosphate Deficiency (G6PD) Mutation Analysis

CODE GT068

Glucose-6-phosphate dehydrogenase deficiency is a genetic disorder that occurs almost exclusively in males. This condition mainly affects red blood cells, which carry oxygen from the lungs to tissues throughout the body. In affected individuals, a defect in an enzyme called glucose-6-phosphate dehydrogenase causes red blood cells to break down prematurely.

SPECIMEN REQUIREMENTS	3-5 ml Peripheral blood and minimum 2 ml of Bone marrow in EDTA tube
TEMPERATURE	Maintain at room temperature. Send to Lab ASAP
SETUP	Saturday to Thursday
TURN AROUND TIME	7-10 working days

Hearing Impairment (GJB2) Mutation Analysis

CODE GT069

GJB2 encodes the protein CONNEXIN 26 and GJB6 encodes the protein CONNEXIN 30. Variants in these genes are responsible for autosomal recessive non-syndromic hearing loss at the DFNB1 locus. Variants in GJB2 can also cause autosomal dominant non-syndromic hearing loss at the DFNA3 locus and keratitis-ichthyosis-deafness syndrome (KID).

SPECIMEN REQUIREMENTS	3-5 ml Peripheral blood in EDTA tube
TEMPERATURE	Maintain at room temperature. Send to Lab ASAP
SETUP	Saturday to Thursday
TURN AROUND TIME	7-10 working days

Hearing Impairment (Multigene Panel Testing)

CODE GT070

There are over 90 genes implicated in hearing impairment. Multigene panel sequencing allows for the determination of the genetic mutations responsible for the disease in the absence of GJB2 mutations.

SPECIMEN REQUIREMENTS	• 3-5 ml Peripheral blood in EDTA tube • For additional samples requirements, see Appendix
TEMPERATURE	Maintain at room temperature. Send to Lab ASAP
SETUP	Saturday to Thursday
TURN AROUND TIME	Up to 8 weeks

HRAS Proto-Oncogene, GTPase (HRAS) Mutation Analysis

CODE GT071

This test detects mutations in the HRAS gene, which includes some codons. Acquired HRAS mutations at these codons have been observed in colon cancer and melanomas. This test can normally detect a heterozygous mutation if it is present in more than about 5% of the cells in the sample.

SPECIMEN REQUIREMENTS	• 3-5 ml Peripheral blood and minimum 2 ml of Bone marrow in EDTA tube • FFPE: Formalin-Fixed Paraffin-embedded tissue (4-5µm thickness): sterile conditions/or container. • For additional samples requirements, see Appendix
TEMPERATURE	Maintain at room temperature. Send to Lab ASAP
SETUP	Saturday to Thursday
TURN AROUND TIME	7-10 working days

Isocitrate Dehydrogenase ($NADP^+$), Cytosolic (IDH1-R132 and IDH2-R172) Mutation Analysis

CODE GT072

IDH1 and IDH2 mutations occur in about 15% of acute myeloid leukemias (AML) and in up to 70% of WHO grade II gliomas. IDH mutations are also frequently observed secondary glioblastoma multiforme (GBM), but rarely in primary GBM. Testing for IDH mutations may be useful for diagnosis and prognosis in glioma, and for risk stratification in AML. This test detects mutations in exons 4 of the IDH1 and IDH2 genes, which includes codons 132 (IDH1) and 172 (IDH2).

SPECIMEN REQUIREMENTS	• 3-5 ml Peripheral blood and minimum 2 ml of Bone marrow in EDTA tube • FFPE: Formalin-Fixed Paraffin-embedded tissue (4-5µm thickness): sterile conditions/or container. • For additional samples requirements, see Appendix
TEMPERATURE	Maintain at room temperature. Send to Lab ASAP
SETUP	Saturday to Thursday
TURN AROUND TIME	7-10 working days

Inborn Errors of Metabolism (Multigene Panel Testing)

CODE GT073

Diseases investigated include 3-hydroxymethyl-3-methylglutaryl-CoA lyase deficiency (also called HMG-CoA lyase deficiency), Alkaptonuria, Alpha-ketoglutarate dehydrogenase deficiency, Aminoacylase 1 deficiency, Argininosuccinic aciduria, Autoimmune Addisson disease, Beta-ketothiolase deficiency, Carbamoyl phosphate synthetase I deficiency, Carnitine palmitoyltransferase I deficiency, Congenital hypothyroidism, Cystinuria D-2-hydroxyglutaric aciduria (D-2- HGA) type I, Galactosemia, Gamma-aminobutyric acid transaminase deficiency, Glutamate formiminotransferase deficiency, Glutaric acidemia type II, Histidinemia, Homocystinuria, Hyperammonemia, Hyperlysinemia, Hypermethioinemia, Hypomagnesemia. Genes include: ACAT1, ETFA, ETFB, SLC5A5, TPO, LRPPRC, PC, D2HGDH, SUMF1, ACADS, PMM2, GUSB, HAL, CYP27B1, MAT1A, FAH, CPT1A, GALT, DBT, BCKDHB, BCKDHA, PEX1, LTC4S, AASS, ABAT, TRPM6, MOCS1, PEX12, NDUFS2.

SPECIMEN REQUIREMENTS	• 3-5 ml Peripheral blood in EDTA tube
TEMPERATURE	Maintain at room temperature. Send to Lab ASAP
SETUP	Saturday to Thursday
TURN AROUND TIME	Up to 8 weeks

Janus Kinase 2 (JAK2 V671F and Exon12) Mutation Analysis

CODE GT074

The somatic point mutation V617F in the JAK2 tyrosine kinase gene (JAK2V617F) has been associated with several chronic myeloproliferative disorders, including polycythemia vera (PV), essential thrombocythemia (ET), primary myelofibrosis (PMF) and BCR-ABL negative myeloprolifrative neoplasm. Some cases of polycythemia vera (PV) have the V617F activating mutation in the JAK2 tyrosine kinase. However, <5% of PV cases have various other mutations in JAK2 exons 12. JAK2 V617F has been also detected in some rare cases of chronic myelomonocytic leukemia, myelodysplastic syndrome, systemic mastocytosis and chronic neutophilic leukemia.

SPECIMEN REQUIREMENTS	3-5 ml Peripheral blood and minimum 2 ml of Bone marrow in EDTA tube
TEMPERATURE	Maintain at room temperature. Send to Lab ASAP
SETUP	Saturday to Thursday
TURN AROUND TIME	7-10 working days

KRAS Proto-Oncogene, GTPase (KRAS) Mutation Analysis

CODE GT075

The KRAS gene is the most commonly mutated oncogene in all human cancer. They are activating mutations that result in continual signal transduction, stimulating downstream signaling pathways involved in cell growth, proliferation, invasion, and metastasis. Mutations in KRAS are determintal for drug response in colon cancer treatment and testing is indicated prior to administration of e.g Imatinib in gastrointestinal tumors.

SPECIMEN REQUIREMENTS	• 3-5 ml Peripheral blood and minimum 2 ml of Bone marrow in EDTA tube • FFPE: Formalin-Fixed Paraffin-embedded tissue (4-5µm thickness): sterile conditions/or container. • For additional samples requirements, see Appendix
TEMPERATURE	Maintain at room temperature. Send to Lab ASAP
SETUP	Saturday to Thursday
TURN AROUND TIME	7-10 working days

MPL Proto-Oncogene, Thrombopoietin Receptor (MPL-Exon 9 and Exon10) Mutation Analysis

CODE GT076

MPL, found at chromosome 1p34, encodes the thrombopoietin receptor that works in concert with thrombopoietin for platelet production. Acquired MPL mutations sever anaemia, myelofibrosis and essential thrombocythemia.

SPECIMEN REQUIREMENTS	3-5 ml Peripheral blood and minimum 2 ml of Bone marrow in EDTA tube
TEMPERATURE	Maintain at room temperature. Send to Lab ASAP
SETUP	Saturday to Thursday
TURN AROUND TIME	7-10 working days

MTHFR (C677T) Mutation Analysis

CODE GT077

Thrombophilia is a multifactorial disease due to the interaction of genetic and environmental risk factors. Some of the recently discovered genetic risk factors, such as factor Factor V Leiden (R506Q), Prothrombin (G20210A) and MTHFR (C677T) are common in the population. Genetic testing of thrombophilic disorders can help in determining the duration of anticoagulant therapy, risk stratification for primary or secondary prophylaxis and genetic counseling.

SPECIMEN REQUIREMENTS	3-5 ml Peripheral blood in EDTA tube
TEMPERATURE	Maintain at room temperature. Send to Lab ASAP
SETUP	Saturday to Thursday
TURN AROUND TIME	7-10 working days

Multiple Mitochondrial Dysfunction Syndrome (ISCA2)

CODE GT078

The protein encoded by this gene is an A-type iron-sulfur cluster (ISC) protein found in mitochondria. The encoded protein appears to be involved in the maturation of mitochondrial iron-sulfur proteins.

SPECIMEN REQUIREMENTS	3-5 ml Peripheral blood in EDTA tube
TEMPERATURE	Maintain at room temperature. Send to Lab ASAP
SETUP	Saturday to Thursday
TURN AROUND TIME	7-10 working days

Nucleophosmin 1 (NPM1- Exon12) Mutation Analysis

CODE GT07

Insertion mutations in exon 12 of the NPM1 gene on chromosome 5 cause abnormal cytoplasmic localization of the NPM1 protein and have been identified in 35-50% of adult acute myeloid leukemia (AML) and in 50-60% of AML cases having normal karyotype (AML-NK). In the absence of FLT3 internal tandem duplication (ITD) mutations, the presence of NPM1 mutations in AML-NK has been associated with better response to induction therapy and favorable overall survival.

SPECIMEN REQUIREMENTS	3-5 ml Peripheral blood and minimum 2 ml of Bone marrow in EDTA tube
TEMPERATURE	Maintain at room temperature. Send to Lab ASAP
SETUP	Saturday to Thursday
TURN AROUND TIME	7-10 working days

NRAS Proto-Oncogene, GTPase (NRAS) Mutation Analysis

CODE GT080

This test detects mutations in the NRAS gene. In colorectal cancer, acquired NRAS mutations are associated with resistance to drugs that target the epidermal growth factor receptor (including cetuximab and panitumumab).

SPECIMEN REQUIREMENTS	• 3-5 ml Peripheral blood and minimum 2 ml of Bone marrow in EDTA tube • FFPE: Formalin-Fixed Paraffin-embedded tissue (4-5μm thickness): sterile conditions/or container. • For additional samples requirements, see Appendix
TEMPERATURE	Maintain at room temperature. Send to Lab ASAP
SETUP	Saturday to Thursday
TURN AROUND TIME	7-10 working days

Obesity (FTO Gene Polymorphism)

CODE GT081

Many variants of the Fat mass and obesity (FTO) gene, clustered within intron 1, have been widely connected to human obesity. In addition to body weight regulation, some variants of the FTO gene have also been associated with common diseases including human adiposity and metabolic disorders.

SPECIMEN REQUIREMENTS	3-5 ml Peripheral blood in EDTA tube
TEMPERATURE	Maintain at room temperature. Send to Lab ASAP
SETUP	Saturday to Thursday
TURN AROUND TIME	7-10 working days

Phosphatidylinositol-4,5-Bisphosphate 3-Kinase Catalytic Subunit Alpha (PIK3CA) Mutation Analysis

CODE GT082

PIK3CA mutation has been associated with poor prognosis in endometrial, breast and colorectal cancers. Mutations in exons 9 and 20 of PIK3CA also been associated with resistance to cetuximab therapy in patients with colorectal cancer.

SPECIMEN REQUIREMENTS	• 3-5 ml Peripheral blood and minimum 2 ml of Bone marrow in EDTA tube • FFPE: Formalin-Fixed Paraffin-embedded tissue (4-5µm thickness): sterile conditions/or container. • For additional samples requirements, see Appendix
TEMPERATURE	Maintain at room temperature. Send to Lab ASAP
SETUP	Saturday to Thursday
TURN AROUND TIME	7-10 working days

PML/RARA t(15;17) Qualitative Analysis

CODE GT083

This test is for the detection of a t(15;17)(q22;q12-21) which fuses the Promyelocytic gene (PML) on chromosome band 15q22 to the Retinoic acid receptor alpha (RARA) gene at 17q12-21. This PML / RARA fusion is associated with a good response to all- transretinoic acid therapy and identified in majority of cases of Acute Promyelocytic Leukemia (APL).

SPECIMEN REQUIREMENTS	3-5 ml Peripheral blood and minimum 2 ml of Bone marrow in EDTA tube
TEMPERATURE	Maintain at room temperature. Send to Lab ASAP
SETUP	Saturday to Thursday
TURN AROUND TIME	5-7 working days

PML/RARA t(15;17) Quantitative Analysis

CODE GT084

This test is for the detection and quantification of a t(15;17)(q22;q12-21) which fuses the Promyelocytic gene (PML) on chromosome band 15q22 to the Retinoic acid receptor alpha (RARA) gene at 17q12-21. This PML / RARA fusion is associated with a good response to all- transretinoic acid therapy and identified in majority of cases of Acute Promyelocytic Leukemia (APL).

SPECIMEN REQUIREMENTS	3-5 ml Peripheral blood and minimum 2 ml of Bone marrow in EDTA tube
TEMPERATURE	Maintain at room temperature. Send to Lab ASAP
SETUP	Saturday to Thursday
TURN AROUND TIME	10-14 working days

RET Proto-Oncogene (RET) Mutation Analysis

CODE GT085

This test can be used to identify genetic variations in the RET gene that are causative for MEN2. Germline mutations in the RET gene on chromosome 10 are causative for multiple endocrine neoplasia, type 2 (MEN2), a monogenic, autosomal-dominant hereditary cancer syndrome. The vast majority (>95%) of MEN2 cases have RET gene mutations in exons 10, 11, 13, 14, 15, or 16.

SPECIMEN REQUIREMENTS	• 3-5 ml Peripheral blood and minimum 2 ml of Bone marrow in EDTA tube • FFPE: Formalin-Fixed Paraffin-embedded tissue (4-5µm thickness): sterile conditions/or container. • For additional samples requirements, see Appendix
TEMPERATURE	Maintain at room temperature. Send to Lab ASAP
SETUP	Saturday to Thursday
TURN AROUND TIME	7-10 working days

Sickle Cell Anemia, HBB Gene Mutation Analysis

CODE GT086

Mutation in the HBB gene changes the normal shape of the red blood cells. The red blood cells become rigid and sickled (C-shaped). The abnormally shaped red blood cells can get stuck in small blood vessels. Blood vessels are like pipes. When they are clogged, blood can't flow through the body as well.

SPECIMEN REQUIREMENTS	3-5 ml Peripheral blood in EDTA tube
TEMPERATURE	Maintain at room temperature. Send to Lab ASAP
SETUP	Saturday to Thursday
TURN AROUND TIME	7-10 working days

Spinal Muscular Atrophy (SMA), SMN Deletions

CODE GT087

SMA causes progressive muscular weakness and degeneration by loss of anterior horn cells in the brain stem and spinal cord. Approximately 95-98% of individuals with SMA are homozygous for a deletion of exons 7 and 8 of the SMN1 gene. The remaining 2%-5% of individuals with SMA are compound heterozygotes for a deletion in combination with a point mutation within the SMN1 gene. Increases in SMN2 gene copy number often modify the phenotype.

SPECIMEN REQUIREMENTS	3-5 ml Peripheral blood in EDTA tube
TEMPERATURE	Maintain at room temperature. Send to Lab ASAP
SETUP	Saturday to Thursday
TURN AROUND TIME	7-10 working days

Thrombophilia Genetic Susceptibility (Multigene Panel)

CODE GT088

Genes included: KLKB1, KNG1, coagulation factor XI(F11), coagulation factor IX(F9), coagulation factor VIII (F8), coagulation factor III, tissue factor (F7), coagulation factor X(F10), coagulation factor V (F5), coagulation factor II, thrombin (F2), fibrinogen alpha chain 9(FGA) FGG, coagulation factor XIII A chain(F13B), THBD, protein C, inactivator of coagulation factors Va and VIIIa (PROC), PROS1, plasminogen activator, tissue type (PLAT), PLG, serpin, SERPIND1, CPB2, and SERPINC1.

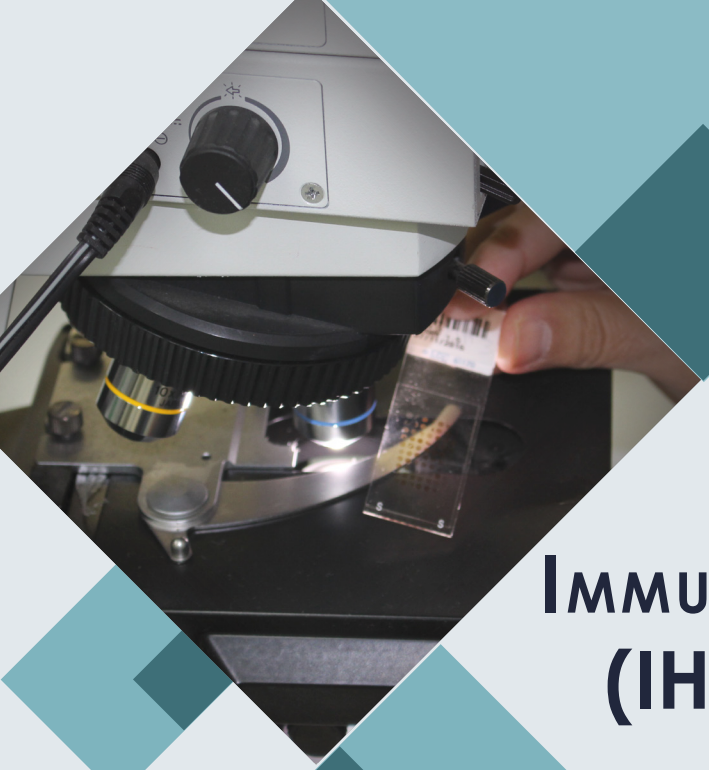
SPECIMEN REQUIREMENTS	• 3-5 ml Peripheral blood in EDTA tube • For additional samples requirements, see Appendix
TEMPERATURE	Maintain at room temperature. Send to Lab ASAP
SETUP	Saturday to Thursday
TURN AROUND TIME	Up to 8 weeks

Tumor Genetic Mutations Profiling

CODE GT089

Genes included: ABL1, AKT1, ALK, APC, ATM, BMPR1A, BRAF, BRCA1, BRCA2, BRIP1, CDK4, CDKN2A, CHEK2, CSF1R, CTNNB1, EGFR, ERBB2, ERBB4, ERCC6, EZH2, FBXW7, FGFR1, FGFR2, FGFR3, EZH2, FBXW7, EZH2, FBXW7, FGFR1, FGFR2, FGFR3, FLT3, G protein, GNA11, G protein, GNAQ, GNAS, HNF1A, HRAS, IDH1, IDH2 JAK2, JAK3, KDR, KIT, KRAS, MEN1, MET, MLH1, MPL, MSH2, MSH6, MUTYH, NBN, ARTN, NOTCH1, NPAT, NPM1, NRAS, PALB2, PDGFRA, PIK3CA, PMS1, PMS2, PTEN, PTPN11, RAD51C, RAD51D, RET, SDHB, SDHD, SMAD4, SMARCB, SMO, SRC, STK11, SUFU, TP53, TSC1, TSC2, VHL, WT1. Mutational profiling of tumors can aid in determining drug response and identifying actionable mutations.

SPECIMEN REQUIREMENTS	• 3-5 ml Peripheral blood and minimum 2 ml of Bone marrow in EDTA tube • FFPE: Formalin-Fixed Paraffin-embedded tissue (4-5µm thickness): sterile conditions/or container. • For additional samples requirements, see Appendix
TEMPERATURE	Maintain at room temperature. Send to Lab ASAP
SETUP	Saturday to Thursday
TURN AROUND TIME	7-10 working days



Immunohistochemistry (IHC) Analysis

Immunohistochemistry (IHC) Analysis

CODE GT090

The (IHC) laboratory employs state-of-the-art automation technology to provide outstanding IHC staining for known markers or on demand marker(s) on human tissues. The IHC lab perform prognostic and diagnostic services of solid tumors on fresh, frozen and/or formalin-fixed, paraffin-embedded tissue on single tissue sections or Tissue MicroArray (TMA) sections prepared by our histology laboratory or provided by the investigator. The majority of these studies are performed on Ventana automated immunostainers.
DNA probes for Bright Field Double in Situ Hybridization (BDISH) technique in frozen and FFPE tissue samples are also offered.

Markers available:

Diagnostic Markers			
Cytokeratins (CKs)	SMMHC	E-cadherin	GCDFP-15
SMA, calponin	PSA	CD56	CD10
P63	S100		
Prognostic Markers			
Ki-67	uPA	Thyroglobulin	p53
TTF-1	COX2	AgNOR	MMR
Bcl2	PAI-1	CAM	(MLH1, MSH2, MSH6 and PMS2)
LCA	MMPs		
Predictive Markers			
HER2/neu	PR	EGFR	c-KIT
ER	VEGF	PDL-1	SLAMF7

Flow Cytometry Analysis

Flow Cytometry Analysis

CODE GT091

Diagnostic flow cytometry analysis is performed on the BD FACSAria III platform and offers support in cellular and molecular analysis of extracellular and intracellular proteins in normal and patient samples as well as sorting of individual cell populations from biological samples. We offer a complete staining and analysis of samples using a list of diagnostic-grade antibodies. Custom analysis using alternative antibodies is also available upon request.

Condition tested:

- Leukemia/ Lymphoma Immunophenotyping
- Leukemia DNA index & ploidy test
- Stem Cell and Umbilical Cord blood Enumeration
- Cell Cycle analysis

Collection and Shipment of Specimens

All samples will be shipped / delivered and received at our Main receiving area in King Fahad Medical Research Center (KFMRC), Jeddah.

SPECIMEN REQUIREMENTS for CYTOGENETICS (Karyotyping/FISH):

- Amniotic fluid: 30 ml directly in 3 different 15 ml conical sterile plastic tube
- Blood: Adult: 3-5 ml, Pediatric 2ml minimum, Fetal blood: 2 ml minimum in Sodium Heparin tube. It is important that a first draw, spicule-rich sample be collected. Invert immediately upon completion of sample collection to prevent the formation of clots. The FISH analysis for un-cultured sample only applicable for urgent cases. Usually referred orders will be treated as routine and will be accommodated according to the laboratory working hours
- Bone marrow (BM): 2 ml in sodium Heparin tube. (It is important that a first draw, spicule-rich specimen to be collected)
- Chorionic villus sample (CVS): Collect 5–25 mg in 15 ml in sterile conical tube filled with sterile transport medium
- Cord blood: 2 ml (minimum) in Sodium Heparin tube
- FFPE: Formalin-Fixed Paraffin-embedded tissue: FFPE Slice (3 slices 10 micrometer in thickness)/FFPE Slide or FFPE blocks in sterile conditions
- Lymph Node: 2–3 mm thick center slice or wedge of lymph node or other Lymphomatous tissue in sterile container containing sterile tissue culture media
- Product of conception (POC): Collect specimen into sterile plastic container filled with transport medium or sterile saline
- Solid tumor: Collect viable and non-necrotic tissue measuring 0.5 cm or 1-5 g in transport medium
- Unstimulated peripheral blood (Leukemic blood): 3-5 ml in Sodium Heparin tube

SPECIMEN REQUIREMENTS for Chromosomal Microarray Analysis:

- Blood: Adult: 3-5 ml, Pediatric 2ml minimum, Fetal blood: 2 ml minimum in Sodium Heparin tube. It is important that a first draw, spicule-rich sample be collected. Invert immediately upon completion of sample collection to prevent formation of clots
- Bone marrow (BM): 2 ml in sodium Heparin tube. (It is important that a first draw, spicule-rich specimen to be collected)
- FFPE: Formalin-Fixed Paraffin-embedded tissue: FFPE Slice (3 slices 10 micrometer in thickness)/FFPE Slide or FFPE blocks in sterile conditions

Collection and Shipment of Specimens

SPECIMEN REQUIREMENTS for Molecular Genetic Analysis:

- Blood: Adult: 3-5 ml, Pediatric 2ml minimum, Fetal blood: 2 ml minimum in EDTA tube. It is important that a first draw, spicule-rich sample be collected. Invert immediately upon completion of sample collection to prevent formation of clots
- Bone marrow (BM): 2 ml in EDTA tube. (It is important that a first draw, spicule-rich specimen to be collected)
- FFPE: Formalin-Fixed Paraffin-embedded tissue: FFPE Slice (3 slices 10 micrometer in thickness)/FFPE Slide or FFPE blocks in sterile conditions
- Fresh tissue in 15ml sterile tube filled with RNA later solution
- Saliva: Freshly collected saliva in Oragene container per kit's specific instructions

SPECIMEN REQUIREMENTS for NGS:

- Blood: Next generation sequencing panels and clinical exome sequencing require 6-10 ml (adult), 5 ml (pediatric) minimum, SNP arrays require 1 ml minimum in EDTA tube. Blood samples not accepted for patients that have undergone an allogenic transplant (e.g. bone marrow or peripheral stem cell)
- Chemotherapy patients: DNA quality may be affected if the patient has received chemotherapy within the last 120 days. Clients will be contacted to provide additional specimen if DNA quality is insufficient
- DNA: 1 µg of DNA in TE (10mM Tris-Cl pH 8.0, 1mM EDTA); preferred 100 µl at ~50 ng/µl concentration for multi-gene/NGS panels. DNA OD 260/280 ratio (preferred 1.7-1.9). Please send agarose picture with high molecular weight genomic DNA, if available
- Saliva: Freshly collected saliva in Oragene container per kit's specific instructions

Policies

A blood sample will be rejected if:

- Clotted
- Haemolysed or low in volume
- Unlabeled or labelled improperly
- Wrong container
- Improperly stored or transported

Other specimens may have a prolonged turnaround times because of lack of necessary ancillary specimens or patient information.

You will be notified of rejected or problem specimens upon receipt.

To avoid specimen rejection, each specimen must be accompanied with a complete REQUEST FORM that provides:

- The patient full name, medical record number, and hospital name, date of birth and gender (If forename has not been established for a new-born baby, the request form must contain surname and names of the parents)
- Specimen type, time/date of specimen collection and test desired
- Clinical indications, history and therapy (for oncology)
- Referring physicians name and contact number
- Three generations pedigree should be provided for all patients, where indicated for hereditary disorders
- Specimen container, transportation temperature and transport medium must be checked and labelled appropriately

Cancellation of Tests:

Request for cancellation must be made via phone or email and will be honored at no charge to the sender if received prior to test setup. Cancellation request after test setup cannot be honored. A report will be issued automatically and charged appropriately.

Additional Tests (Add-ons):

Requests for additional orders on specimens received by Ro'ya must be confirmed via email. Call to check on sample requirements and availability to avoid any rejections. Repeated or additional tests will take additional turnaround time.

Policies

Test Result call back and confidentiality:
Results will be sent to a pre-assigned email address or given to the patient.
Ro'ya endeavors to maintain the confidentiality of all patients' information.

Unsatisfactory Analytical Result:
Ro'ya endeavors to perform all tests to the highest standards. Tests generating unsatisfactory or inconclusive results will not be charged to the sender.

Urgent orders:
All referred orders will be treated as routine and will be accommodated according to the laboratory working hours. Urgent requests must be approved by an authorised member of staff at the sending facility prior to submitting the order and sending the specimens. Processing urgent orders will waiver any contractual discounts and will be charged at the full listed price of the test(s).

Unlisted tests:
New procedures are developed throughout the year; therefore, some tests are not listed on our request form, booklet or website.
For information about the unlisted tests, contact Ro'ya general inquiry.

Supplies:
Special specimen collection containers and kits, sterile vials and request forms are supplied upon request.

Andrology Lab

Semen Analysis

CODE IP001

Initial macroscopic and microscopic examination. Semen analysis should begin with a simple inspection soon after liquefaction, preferably at 30 minutes, but no longer than 1 hour after ejaculation.

SPECIMEN REQUIERMENTS	The sample should be collected after a minimum of 3 days of sexual abstinence, in a private room near the laboratory, in a sterile container labelled with the patient ID, MRN,NAME, time of receiving the sample should written in the container.
TEMPERATURE	Maintain at 37^{o} C temperature for 30 Min.
SETUP	Before 3 pm
TURN AROUND TIME	Same day

Sperm Viability Using Eiosin-Nigrosin

CODE IP002

Initial macroscopic examination should be done after 30 minutes of sample collection. Sperm vitality test should be assessed as soon as possible after liquefaction of the semen sample, preferably at 30 minutes, but in any case within 1 hour of ejaculation, by using (Eiosin-Nigrosin) stain.

SPECIMEN REQUIERMENTS	The sample should be collected after a minimum of 3 days of sexual abstinence, in a private room near the laboratory, in a sterile container labelled with the patient ID, MRN, NAME, , time of receiving the sample should written in the container.
TEMPERATURE	Room temperature
SETUP	Before 3 pm
TURN AROUND TIME	Same day

Sperm Freezing

CODE IP003

Initial macroscopic and microscopic examination should be done after 30 minutes of sample collection. Labelling straws or vials is essential by patient ID, MRN, NAME. Sample should be double-checked by two people and evidence of this checking witnessed in the laboratory records. Ideally a technician should process only one semen sample at any given time.

SPECIMEN REQUIERMENTS	The sample should be collected after a minimum of 3 days of sexual abstinence, in a private room near the laboratory, in a sterile container labelled with the patient ID, MRN, NAME, , time of receiving the sample should written in the container.
TEMPERATURE	Maintain at -196^{0}C after treatment (Deep freeze)
SETUP	Before 3 pm
TURN AROUND TIME	Same day

Peroxidase Test (SPERMAR)

CODE IP004

Initial macroscopic and microscopic examination should be done after 30 minutes of sample collection. The test useful in distinguishing polymorphonuclear leukocytes from multinucleated spermatids, which are peroxidase-free. Assessing peroxidase-positive cell number in the hemocytometer chambers.

SPECIMEN REQUIERMENTS	The sample should be collected after a minimum of 3 days of sexual abstinence, in a private room near the laboratory, in a sterile container labelled with the patient ID, MRN, NAME, , time of receiving the sample should written in the container.
TEMPERATURE	Room temperature
SETUP	Before 3 pm
TURN AROUND TIME	Same day

Sperm Viability Test Using Hypo-Osmotic Swelling (HOS)

CODE IP005

Initial macroscopic and microscopic examination should be done after 30 minutes of sample collection, As an alternative to dye exclusion, the hypo-osmotic swelling (HOS) test may be used to assess vitality. This is useful when staining of spermatozoa must be avoided, when choosing spermatozoa for ICSI.

SPECIMEN REQUIERMENTS	The sample should be collected after a minimum of 3 days of sexual abstinence, in a private room near the laboratory, in a sterile container labelled with the patient ID, MRN, NAME, , time of receiving the sample should written in the container.
TEMPERATURE	Maintain at 37° C temperature.
SETUP	Before 3 pm
TURN AROUND TIME	Same day

Anti-Sperm Antibody Test (ASA TEST)

CODE IP006

Initial macroscopic and microscopic examination should be done after 30 minutes of sample collection. ASAs in semen belong almost exclusively to two immunoglobulin classes: IgA and IgG, Two direct tests are assessed; the mixed antiglobulin reaction (MAR) test and the immunobead (IB) test.

SPECIMEN REQUIERMENTS	The sample should be collected after a minimum of 3 days of sexual abstinence, in a private room near the laboratory, in a sterile container labelled with the patient ID, MRN, NAME, time of receiving the sample should written in the container.
TEMPERATURE	Room Temperature
SETUP	Before 3 pm
TURN AROUND TIME	Same day

Intra Uterine Insemination (IUI)

CODE IP007

Intra Uterine Insemination(IUI) involves the injection of treated sperms directly into the uterus with a close proximity to the fallopian tubes. Usually, it is an option for couples suffering from infertility rendered not so severe as to resort to IVF or ICSI. The sample is processed at the lab where the best sperm are selected and additional materials are added to boost them and better prepare them for fertilization. Injection of sperm inside the womb is performed using a simple method similar to internal examination and without causing pain. Usually no sedation is required. Afterwards, the wife stays for about half an hour at the hospital before leaving, and she can travel distances by car or plane if she wishes.
There are no restrictions on sexual intercourse after administering the injection.
The wife is required to undergo a pregnancy blood test after two weeks of the procedure.

SPECIMEN REQUIERMENTS	The wife's ovulation is monitored which starts between the sixth and tenth day after her period depending on the treatment used to enhance ovulation. In some cases no treatment to enhance ovulation is used. The sample should be collected after a minimum of 3 days of sexual abstinence, in a private room near the laboratory, in a sterile container labelled with the patient ID, MRN, NAME, time of receiving the sample should written in the container.
TEMPERATURE	Room temperature
SETUP	Sunday to Thursday
TURN AROUND TIME	Same day
INDICATIONS	• Lack of natural conception is related to the husband such as the decreased number of sperm or its mobility or shape. • If the lack of natural conception is the incapability of the sperm to pass through the cervix. • If many years passed after marriage without pregnancy with no reason hindering conception from either partner.

IVF Technologies

Intra Cytoplasmic Sperm Injection (ICSI)

CODE IP008

Injection of the sperm within the egg near the nucleus is a modern approach which requires accurate sensitive technology and skillful laboratory work. At our Assisted Reproductive Unit we provide the best with our team who has extensive knowledge and experience in implementing this procedure.

SPECIMEN REQUIERMENTS	The sample should be collected after a minimum of 3 days of sexual abstinence, in a private room near the laboratory, in a sterile container labelled with the patient ID, MRN, NAME, time of receiving the sample should written in the container.
TEMPERATURE	Room temperature
SETUP	Sunday to Thursday
INDICATIONS	• Husbands who suffer from severe weakness in sperm quality, number, movement, or all of the above. • Husbands suffering from absence of transfer canals for sperm, or natural occlusion, or suffering from reverse ejaculation of sperm towards the bladder. • Sterile cases with unknown causes.

Assisted Hatching

CODE IP009

This procedure performed on embryo(s) the aim of which is to improve implantation of embryo(s) thus achieving a pregnancy. It is initially involves making a hole using a guided laser beam.

INDICATIONS	• Certain studies have shown that making a hole in the surrounding membrane would facilitate the process of implantation. • Currently, assisted hatching is not offered as routine for all patients. It is usually offered for repeated failure cases. • Thick or hard shell (Zona).

Our IVF unit seeks to offer medical service of the highest international standards for its patients. Complete cryopreservation treatments and other state of art technologies are available upon request.

Preimplantation Genetic Screening (PGS) and Preimplantation Genetic Diagnosis (PGD)

Preimplantation Genetic Screening (PGS)

CODE IP010

PGS, or preimplantation genetic screening, is a genetic test performed on embryos produced through IVF. PGS evaluates embryos for chromosomal abnormalities to help your care team select the best embryo for transfer and improve your chance of achieving a successful pregnancy.

SPECIMEN REQUIERMENTS	1-2 blastomeres in 0.2µl sterile tube
TEMPERATURE	Shipped frozen on ice/dry ice. Send to the Lab ASAP
SETUP	Sunday to Wednesday before 12 PM
TURN AROUND TIME	24-48 hours after set up

Contact us for more information: +966 53 3537257
+966-12-6401000 Ext: 72141

Preimplantation Genetic Diagnosis (PGD)

CODE IP011

PGD or pre-implantation genetic diagnosis has helped many couples with serious genetic diseases in the family conceive healthy babies. We have a genetic team dedicated to helping patients who are at risk of inherited conditions and can provide you with information about these risks, and support you with any decisions you make.

SPECIMEN REQUIERMENTS	If you know or suspect you have a genetic or chromosomal abnormality please book an appointment.
SETUP	Sunday to Thursday before 4 PM

Contact us for more information: +966 53 3537257
+966-12-6401000 Ext: 72141

PGD for Single Gene Disorder

CODE IP012

Disorders caused by the inheritance of a single defective gene are known as monogenic diseases or single gene disorders. Monogenic diseases fall into two main categories. Firstly, there are 'recessive' diseases, which do not produce any symptoms unless a defective copy of the gene is passed on by both the Mother and the Father. The second category is comprised of disorders that are said to be 'dominant', which only require one defective copy of the gene to be inherited in order to occur.
Another category of monogenic diseases has an X-linked inheritance. Most couples at risk for an X-linked condition are identified by review of the family history or the birth of an affected child.

SPECIMEN REQUIERMENTS	If you know or suspect you have a genetic or chromosomal abnormality please book an appointment.
SETUP	Sunday to Thursday before 4 PM

Contact us for more information: +966 53 3537257
+966-12-6401000 Ext: 72141

PGD for HLA Matching

CODE IP013

PGD of single gene disorders, combined with HLA matching, represents one of the most recent applications in reproductive medicine. In such cases, PGD is used not only to avoid the birth of affected children, but also to conceive healthy children who may also be potential HLA-identical donors of haematopoietic stem cells (HSC) for transplantation in siblings with a life-threatening disorder. At delivery, HSC from the newborn umbilical cord blood are collected and used for the haematopoietic reconstruction of the affected sibling that without stem cell transplant is likely to die.

SPECIMEN REQUIERMENTS	If you know or suspect you have a genetic or chromosomal abnormality please book an appointment.
SETUP	Sunday to Thursday before 4 PM

Contact us for more information: +966 53 3537257
+966-12-6401000 Ext: 72141

PGD for Inherited Cancer

CODE IP014

With current progress in understanding of the molecular basis of cancers, and sequencing of the genes involved in malignancy, inherited cancer predisposition became one of the emerging indications for PGD. Using a standard IVF procedure, oocytes or embryos can be tested for different mutations predisposing to cancer, preselecting and transferring only mutation-free embryos back to the patients.

SPECIMEN REQUIERMENTS	If you know or suspect you have a genetic or chromosomal abnormality please book an appointment.
SETUP	Sunday to Thursday before 4 PM

Contact us for more information: +966 53 3537257
+966-12-6401000 Ext: 72141

PGD for Late-Onset Disorders

CODE IP015

One of the uses of PGD are the identification of embryos at risk for late-onset diseases. Late-onset Disorders are complex and have both genetic and environmental causes. Genetic tests may indicate a susceptibility or predisposition for these diseases. Diseases like Alzheimer's, Huntington's and heart disease caused by single genes that also are seen later in life.

SPECIMEN REQUIERMENTS	If you know or suspect you have a genetic or chromosomal abnormality please book an appointment.
SETUP	Sunday to Thursday before 4 PM

Contact us for more information: +966 53 3537257
+966-12-6401000 Ext: 72141

PGD for Structural Chromosomal Abnormalities

CODE IP016

Chromosomes are very important for healthy growth and development. Embryos with the incorrect structure of chromosomes typically do not result in a successful pregnancy or may lead to the birth of a child with a genetic condition. The lab uses the most advanced embryo screening technology available, providing the most complete picture of chromosomal health.

SPECIMEN REQUIERMENTS	If you know or suspect you have a genetic or chromosomal abnormality please book an appointment.
SETUP	Sunday to Thursday before 4 PM

Contact us for more information: +966 53 3537257
+966-12-6401000 Ext: 72141

Collection and Shipment of Specimens

All samples will be shipped / delivered and received at our Main receiving area in King Fahd Medical Research Center (KFMRC), Jeddah.

Policies

Unacceptable specimens:

Semen Sample will be rejected if:

- Unlabeled or labelled improperly
- Wrong container
- Improperly stored or transported
- Received after more than 30 mins of the ejaculation
- Contaminated Samples

You will be notified of rejected or problem specimens upon receipt.

To avoid specimen rejection, each specimen must be accompanied with a complete REQUEST FORM that provides:

- The patient full name, medical record number, date of birth and gender
- Specimen type, time and date of specimen collection and test desired
- Clinical indications, history and therapy (for infertility)
- Referring physicians name and contact number
- Specimen must contain full name and medical record number and patient ID
- Specimen container, transportation temperature and transport medium must be checked
- Frozen samples should be received in liquid nitrogen container with freezing-thawing consent

Cancellation of Tests:

Request for cancellation must be via phone or email and will be honored at no charge if received prior to test setup. Cancellation request after test setup cannot be honored. A report will be issued automatically and charged appropriately. Complete IVF procedure will be cancelled if the husband failed to give a fresh ejaculate/ no frozen sample is available.

Policies

Additional Tests (Add-ons):
Requests for additional orders on specimens received by CIMP must be confirmed via call or email. Call to check on sample requirements and availability to avoid any rejections. Repeated or additional tests will take additional turnaround time.

Test Result call back and confidentiality:
Results will be sent to a pre assigned email address or given to the patient.
CIPM endeavors to maintain the confidentiality of all patients' information.

Unsatisfactory Analytical Result:
There will be no charge incurred if CIPM is unable to obtain a satisfactory analytical result.

Urgent orders:
All referred orders will be treated as routine and will be accommodated according to the laboratory working hours. Urgent processing request must be approved by a designated person before submitting the order and sending the specimens. Processing approved urgent orders will waiver any contractual discounts and will be charged by the full listed price of the order.

Unlisted test:
New procedures are developed throughout the year; therefore, some tests are not listed on our request form, booklet or website.
For information about the unlisted test, contact CIPM general inquiry.

Supplies:
Special specimen collection containers and kits, sterile vials and request forms are supplied upon request.

Index

Location

Ro'ya Specialized Medical Laboratories

King Fahd Medical Research Center

King Abdulaziz University

Jeddah, Kingdom of Saudi Arabia

Ro'ya Specialized Medical Laboratories
King Fahd Medical Research Center, King Abdulaziz University
Jeddah, Kingdom of Saudi Arabia

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