BION
Genetic Lab

Edition A - April 2023

Where science meets innovation

Contents

BION Genetic Laboratory

MOH NO: 167/2023

BION Genetic Laboratory

Cancer Panels

Cancer Panels

Genetic testing for hereditary and somatic cancers can provide life-changing results in affected patients and their relatives, accompanied by potential actionable steps for genetic-related cancers. With many different applications of genetic testing to detect and care for cancer, we can guide you in selecting the right options to enhance the treatment of your patients suffering from hereditary cancers. Having identified genetic variants associated with oncological diseases in more than 200 different genes, we can provide a comprehensive range to foster cancer diagnosis, prognosis, treatment selection, and monitoring.

1. **Panel BRCA1, BRCA2 (N+M)**
   Breast cancer is the most common type of cancer in woman constituting around 25% of all females' cases.

   No. of genes: 2
   TAT: 21 days
   Coverage: ≥99.50% ≥20x
   Details: Sequencing and MLPA: BRCA1, BRCA2

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Cancer Panels

Breast and Ovarian Cancer Panel

2. Breast cancer is one of the most common cancers in the world affecting ~12.5% of women during their lifetime. 5–10% of these patients have a hereditary form.
Mutations in the **BRCA1** and **BRCA2** genes are the most common hereditary cause. However, other genes such as **ATM, BRIP1, CHEK2, PALB2, RAD51, etc.** have also been associated with increased risk.

No. of genes: 28
TAT: 21 days
Coverage: ≥99.50% ≥20x
Details: NGS including CNV analysis

COMMON SYNDROMES AND DISORDERS COVERED

. Breast cancer
. Ovarian cancer

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3. Cancer Panel [70]

It has been carefully selected based on its risk potential in the development of one or more of the following cancers: breast, ovarian, colorectal, gastric, thyroid, endometrial, pancreatic,melanoma, renal, and prostate. This panel is appropriate for patients with positive personal history early-onset cancer, rare cancer, bilateral cancer, or multiple primary cancers.

No. of genes:70
TAT:21 days
Coverage:99.50% ≥20x
Details: NGS including CNV analysis

COMMON SYNDROMES AND DISORDERS COVERED

- Breast cancer
- Colorectal cancer
- Endometrial cancer
- Familial adenomatous polyposis
- Gastric cancer
- Gastrointestinal stromal tumor
- Melanoma
- Ovarian cancer
- Pancreatic cancer
- Prostate cancer
- Renal cancer
- Skin cancer
- Thyroid cancer
- Uterine cancer

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4. GI cancer panel
GI cancer panel detects genes that are associated with colon, pancreatic, and gastric cancer.

No. of genes: 33
TAT: 21 days
Coverage: ≥99.50% ≥20x
Details: NGS including CNV analysis

COMMON SYNDROMES AND DISORDERS COVERED

. Colorectal cancer
. Familial adenomatous polyposis
. Gastric cancer
. Hereditary nonpolyposis colorectal cancer
. Pancreatic cancer

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5. Comprehensive Cancer Panel
This panel is a specific and sensitive panel with all known highly penetrant cancer genes. Each gene has been carefully selected based on its risk potential in the development of one of the following cancers: breast, ovarian, colorectal, gastric, bowel, endometrial, pancreatic, melanoma, renal and prostate cancer.

No. of genes: 118
TAT: 21 days
Coverage: ≥99.00% ≥20x
Details: NGS including CNV analysis

COMMON SYNDROMES AND DISORDERS COVERED

. Beckwith-Wiedemann syndrome
. Breast cancer
. Colorectal cancer
. Endometrial cancer
. Familial adenomatous polyposis
. Gastric cancer
. Gastrointestinal stromal tumor
. Hereditary Paraganglioma/ Pheochromocytoma
. Melanoma
. Ovarian cancer
. Pancreatic cancer
. Paragangliomas/Pheochromocytoma/
. Gastrointestinal stromal
. Prostate cancer
. Renal cancer
. Retinoblastoma
. Rothmund-Thomson syndrome (Type 2)
. Skin cancer
. Thyroid cancer
. Uterine cancer

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BION Cancer
Early detection(Ed)

BION Cancer early detection

What?

A prescription blood test that can help early detection of multiple cancers in asymptomatic individual based on presence of Circulating Tumor Cells (CTCs).

How?

Blood samples are evaluated for presence of circulating tumor cells (CTCs) which may be further analysed to determine the organ or tissue of origin.

Why?

Screening is not available for all cancers. Where available, it may be an inconvenient procedure that necessitates a visit to an advanced healthcare facility. Bion cancer early detection is critical for successful cure with minimum chances of recurrence.

Sample type:
Peripheral blood as per protocol.

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BION Cancer
Early detection(Ed)

	BION Cancer (Ed)	Mammography
Sensitivity	88.2%	86.9%
Specificity	100.0%	88.9%

Sensitivity: Ability to designate an individual with disease as positive
Specificity: Ability to designate an individual who does not have a disease as negative

This test is a safe and reliable test that can help detect cancers more effectively, which can asist physicians too make timely life-saving diagnostic and clinical management decisions.

DOES A 'NEGATIVE' RESULT MEAN THAT I AM CANCER FREE AT PRESENT?

- Within the limits of error, a 'negative' result is an indication of undetectable cancer at the time of the test. For other cancers not covered by the test, no inference should be drawn from a 'negative' result.
  A 'negative' result means that at the time of the test there were no detectable cancer cells suggestive of cancer circulating in the blood.
  This test is recommended to be performed annually.

IF MY RESULT IS 'POSITIVE', HOW DO I FIND THE LOCATION OF CANCER?

- The test indicates the organ affected by cancer with high accuracy in most types of cancers / cases. In all cases of 'positive' results, the Report should be interpreted by your physician who can guide you on the further course of action.

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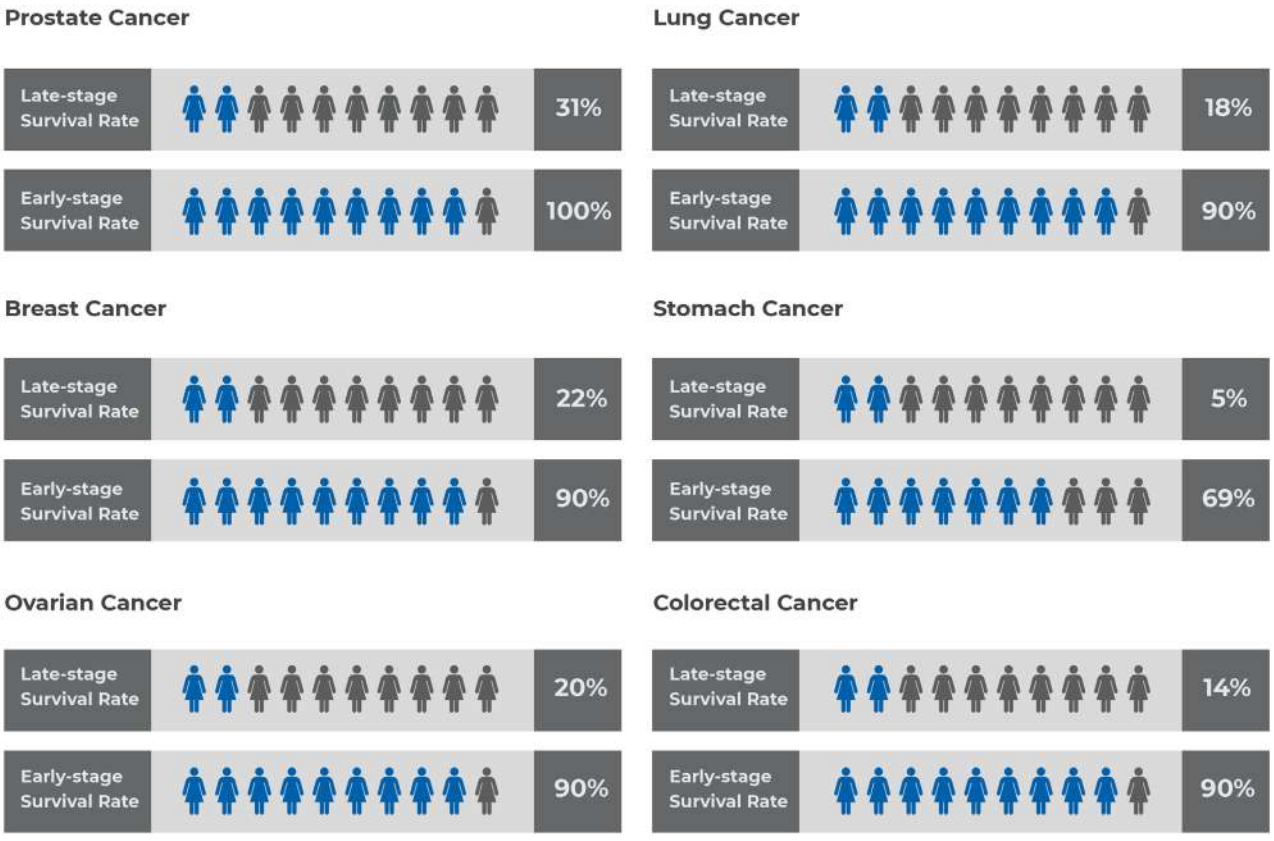
BION Cancer
Early detection(Ed)

Late-Stage Survival Rate compared to Early-Stage Survival Rate:

BION Cancer(Ed) is a blood test that can enable early detection of multiple cancers in asymptomatic individuals.



Three most common Cancers:

Men

1. Prostate
2. Lung
3. Colorectal

Women

1. Breast and ovarian
2. Lung
3. Colorectal

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BION Cancer
Early detection(Ed)

It includes 5 different tests:

1. Golden test
It is complete test and covers Melanoma, Head and Neck, Salivary Gland, Thyroid, Lung, Breasts, Liver, Biliary Tract, Gastro-Intestinal, Soft Tissue and Osteosarcomas, Mesothelioma, Urinary Tract, Gynaecological, Male Cancers including Prostate, CNS, Thymus, Adrenal, Skin.

2. Silver Test
The test is intended for early detection of Lung, Stomach, Colon, Pancreas, Prostate, Breast, and Ovarian cancers.

3. Diabetes can detect 6 types of Diabetes Associated Cancers
Diabetes can help in early detection of cancers that originate in the Liver, Gallbladder, Pancreas, Kidney, Bladder and Colorectum.

4. Breast test the most common cancer in the women

5. Prostate test the most common cancer in the men

IT is a safe and reliable test that can help detect cancers more effectively, which can assist physicians to make timely life-saving diagnostic and clinical management decisions.

HOW FREQUENTLY SHOULD THE TEST BE REPEATED?
- It is a non-invasive blood test, there is no disadvantage in frequent testing. However, we advise that test should be performed every 12 months.

DOES T THE TEST DETECT 'EXISTENCE OF CANCER' OR 'POSSIBILITY OF CANCER'?
- It detects 'existence of cancer'.

CAN THE TEST BE USED TO MONITOR RECURRENCE AFTER CANCER TREATMENT?
- No

IS THE TEST RECOMMENDED FOR A PERSON SUSPECTED TO HAVE/HAS SYMPTOMS OF CANCER?
- No, such individuals should seek expert medical advice without delay.

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BION Cancer
NRAS, KRAS, BRAF

1. NRAS, KRAS, BRAF

Why Get Tested? To determine whether a cancer, usually a large bowel (colorectal) cancer is positive for KRAS or NRAS gene mutation, which **helps to guide treatment and determine outcome.** RAS gene mutation analysis is also used in the assessment of some other cancer types including head and neck cancer.

When To Get Tested?
If you have been diagnosed with a cancer and your doctor wants to determine whether the **KRAS** and NRAS genes are mutated in the tumour. If the KRAS or NRAS genes are mutated, the cancer will not be responsive to treatment with RAS targeted therapy.

Sample Required?
A sample of cancer tissue obtained during a biopsy. Generally, this test is done on the biopsy taken for initial diagnosis and a second biopsy is not necessary.

"All-RAS" testing (testing for both KRAS and NRAS) is currently the only additional test that oncologists will routinely request if metastatic disease is found

TAT: 10 days
Details: NRAS: exon 2 and 3; KRAS: exon 2,3, and 4 ;BRAF exon 15
Sanger Sequencing (100%)

2. JAK2
A gene that makes a protein that sends signals in cells to promote cell growth and helps control the number of red blood cells, white blood cells, and platelets that are made in the bone marrow. Mutated (changed) forms of the JAK gene have been found in some types of blood conditions, including polycythemia vera, essential thrombocythemia, and primary myelofibrosis. These changes may cause the body to make too many blood cells.

TAT: 8 days
Details: Sanger Sequencing (100%)

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Cardiology panel

Cardiology panel

This panel includes the most relevant genes for arrhythmias, congenital heart disease, and cardiomyopathies. Syndromes included: Long and short QT, Brugada syndrome, catecholaminergic polymorphic ventricular tachycardia, cardiomyopathies dilated and hypertrophic, and congenital heart defects. In addition, this panel includes vascular abnormalities, such as dolichoectasia and hereditary hemorragic telangiectasia.

No. of genes: 327
TAT: 4-6 weeks
Coverage: ≥99.00% ≥20x

COMMON SYNDROMES AND DISORDERS COVERED

- Arrhythmogenic right ventricular cardiomyopathy
- Brugada syndrome
- Catecholaminergic polymorphic ventricular tachycardia
- Congenital heart defects
- Dilated cardiomyopathy
- Dolichoectasia
- Hereditary arrhythmia syndromes
- Hereditary hemorrhagic telangiectasia
- Heterotaxy syndrome
- Hypertrophic cardiomyopathy
- Hypomagnesemia
- Long QT syndrome
- Short QT syndrome

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Connective tissue and related Disorders panel

Connective tissue and related disorders panel

Our connective tissue and related disorders panel provides a profound one-step evaluation of several genes to detect different disorders with similar phenotypes, such as Marfan Syndrome, Loeys-Dietz, cutis laxa, Ehlers-Danlos, Stickler syndrome, and Familial thoracic aortic aneurysm and dissection.

No. of genes: 76
TAT: 4-6 weeks
Coverage: ≥99.00% ≥20x
Details: NGS including CNV analysis

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COMMON SYNDROMES AND DISORDERS COVERED

- Cutis laxa
- Ehlers-Danlos syndrome
- Familial thoracic aortic aneurysm and dissection
- Loeys-Dietz syndrome
- Marfan syndrome
- Osteogenesis imperfecta
- Stickler syndrome

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SKIN disorders panel

Skin disorders panel

BION Skin is our diagnostic test for patients displaying skin disorders. Our panel includes genes for hypotricosis, epidermoly- sis bullosa, and congenital ichthyosis, among others. In addition, CentoSkin tests for albinism and other conditions with similar pigmentation abnormalities such as Hermasky-Pudlak syndrome, Griscelli snydrome and Waardenburg syndrome. For melanoma, please check our oncology section.

No. of genes: 152
TAT: 4-6 weeks
Coverage: ≥99.00% ≥20x
Details: NGS including CNV analysis

COMMON SYNDROMES AND DISORDERS COVERED

- Albinism oculocutaneous
- Chediak-Higashi syndrome
- Congenital ichthyosis
- Cutis laxa
- Epidermolysis bullosa
- Griscelli syndrome
- Hermasky-Pudlak syndrome
- Ichthyosis extended
- Non-syndromic hypotrichosis
- Waardenburg syndrome

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Congenital adrenal hyperplasia (CAH) panel

Congenital adrenal hyperplasia (CAH) panel

Our congenital adrenal hyperplasia **(CAH) panel** is designed for patients suspected of having CAH. CAH is a group of inherited disorders characterized by improper functioning of the adrenal glands, leading to abnormal production of steroid hormones, such as a cortisol or aldosterone. Our panel includes the analysis of the **CYP21A2** gene, which codes for the enzyme **21-hydroxylase.** More than 90.0% of CAH cases are caused by a deficiency of this enzyme.

No. of genes: 12
TAT: 4-6 weeks
Coverage: ≥99.00% ≥20x
Details: NGS including CNV analysis,
MLPA: CYP21A2, Sanger sequencing: CYP21A2

COMMON SYNDROMES AND DISORDERS COVERED

. Congenital adrenal hyperplasia

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Diabetes and Obesity panel

Diabetes and obesity panel

Our diabetes and obesity panel is recommended for patients with abnormalities in glucose metabolism, such as hyperinsulinemic hypoglicemia, diabetes neonatal, **MODY**, diabetes in adults, and familial hypercholesterolemia, as well as for patients displaying insulin resistance, from mild to the severe spectrum (Donohue syndrome), and for patients with familial hyperinsulinism. Disorders caused by imprinting errors or uniparental disomy, such as **6q24-**related transient neonatal diabetes mellitus and Beckwith Wiedemann syndrome, are not detected with this panel.

No. of genes: 265
TAT: 4-6 weeks
Coverage: ≥99.00% ≥20x
Details: NGS including CNV analysis

COMMON SYNDROMES AND DISORDERS COVERED

- Bardet-Biedl syndrome
- Congenital glycosylation disease
- Congenital hyperinsulinism
- Congenital hypothyroidism
- Diabetes insipidus
- Growth hormone deficiency
- Familial hypercholesterolemia
- Hypoglycemia
- Maturity onset diabetes of the young
- Neonatal diabetes
- Obesity

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Hearing loss panel

Hearing loss panel

Hearing loss is a common condition in children, affecting 1 in 100 live births. In more than 50.0 % of cases, there is a genetic cause for this disorder, of which 70.0 % experience non-syndromic hearing loss. This panel includes genes associated with syndromic and non-syndromic hearing loss. Both autosomal recessive and dominant genes are included in the panel. In addition, This panel includes several other syndromes, such as Alport, Pendred, Waardenburg, Usher, and branchio-oto-renal, among others.

No. of genes: 196
TAT: 4-6 weeks
Coverage: ≥99.00% ≥20x
Details: NGS including CNV analysis

COMMON SYNDROMES AND DISORDERS COVERED

- Alport syndrome
- Coffin-Lowry snydrome
- Deafness autosomal recessive and dominant
- Non-syndromic hearing loss
- Pendred syndrome
- Perrault syndrome
- Pfeiffer syndrome
- Sensorineural hearing loss
- Stickler syndrome
- Syndromic hearing loss
- Usher syndrome
- Waardenburg syndrome
- Wolfram syndrome

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Hereditary thrombophilia

IMPACT ON WOMEN'S HEALTH

Thrombophilia has important implications for women's health, particularly contraceptive therapy, fertility, and pregnancy.

- **Hormonal Contraceptive/Replacement Therapy**

Use of combination oral contraceptive pills, especially those containing third-generation progestins, has been associated with at least a 3-fold increased risk of VTE. **Use of combination oral contraceptive pills in patients with thrombophilia such as factor V Leiden heterozygosity is associated with at least a 30-fold increase in risk of VTE.** The increased risk of VTE appears to be highest around the time of oral contraceptive pill initiation and within the first 6 months.

- **Infertility**

In a subset of women, thrombophilia results in infertility, which may manifest as difficulty with conception, recurrent pregnancy loss, or both.
The mechanism by which thrombophilia causes infertility does not appear to be limited to a hypercoagulable state but may also include abnormalities of trophoblast differentiation and placentation. **Thrombophilias are associated with both early and late pregnancy loss.**

- **Thrombophilia and Pregnancy**

Thrombophilias also increase the risk of pregnancy-related complications, including VTE. **For example, the relative risk increase for VTE ranges from 9-fold in women with heterozygosity for factor V Leiden to 34-fold in those with homozygosity for the mutation.** However, the absolute risk increase in pregnant women with factor V Leiden is 0.2%. Therefore, although the relative risk of VTE resulting from thrombophilia in pregnancy is high, the absolute risk is low.

The risk of other pregnancy-related complications such as preeclampsia and placental abruption is also increased in the presence of thrombophilia.

MOH NO: 167/2023

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Hereditary thrombophilia

Hereditary thrombophilia

The Panel **analyzes genes that are associated with hereditary thrombophilia. Hereditary thrombophilia** is characterized by increased clotting tendency and increased risk for deep venous thrombosis and/or venous thromboembolism.

Specific genetic defects should be suspected when a thrombotic event has any of the following characteristics:

1. Spontaneous with no predisposing condition, such as prolonged immobilization or surgery
2. Patient suffers from more than one thrombotic event
3. Patient has a positive family history of thrombosis
4. Patient is <50 years old
5. Thrombosis occurs at an unusual site (eg, mesenteric or cerebral brain)

Disorders tested

- Factor V Leiden-related thrombophilia
- Prothrombin-related thrombophilia
- Protein C deficiency
- Hereditary fibrinogen abnormalities
- Protein S deficiency
- Antithrombin III deficiency
- Essential thrombocythemia

TAT: 5-7 days
Details: Real-Time PCR

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Blood coagulation panel

Blood coagulation panel

Our blood coagulation panel contains genes to diagnose thrombophilia, thrombocytopenia, hereditary hemorrhagic telangiectasia, ARC syndrome, Hermasky-Pudlak syndrome, coagulation factor disorders, hemophilia, and platelet related disorders.

No. of genes: 112
TAT: 4-6 weeks
Coverage: ≥99.00% ≥20x
Details: NGS including CNV analysis

MOH NO: 167/2023

COMMON SYNDROMES AND DISORDERS COVERED

- Afibrinogenemia
- Arthrogryposis-renal dysfunction-cholestasis syndrome
- Coagulation factor disorders
- Hemophilia
- Hereditary angioedema
- Hereditary hemorrhagic telangiectasia
- Hermasky-Pudlak syndrome
- Platelet related disorders
- Shwachman-Diamond syndrome
- Thrombocytopenia
- Thrombophilia

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Bone marrow failure / Anemia panel

Bone marrow failure / Anemia panel

Our bone marrow failure / Anemia panel is intended for patients with abnormalities in more than two blood cell types (red blood cell, white blood cell, and platelets) who present symptoms of lethargy, recurrent infections, excessive bleeding, abnormal pigmentation, enlarged spleen, and malignancies.
Some specific disorders detected with this panel are hemophagocytic lymphohistiocytosis, Seckel syndrome, thrombocytopenia, Fanconi anemia, dyskeratosis congenita, Shwachman Diamond syndrome as well as other types of anemias, such as thalassemia alpha and beta, sickle cell disease, spherocytosis, megaloblastic anemia, congenital sideroblastic, and dyserythropoietic anemia.

No. of genes: 214
TAT: 4-6 weeks
Coverage: ≥99.00% ≥20x
Details: NGS including CNV analysis

MOH NO: 167/2023

COMMON SYNDROMES AND DISORDERS COVERED

. Bleeding disorders
. Bone marrow failure syndrome
. Congenital dyserythropoietic anemia
. Congenital sideroblastic anemia
. Diamond-Blackfan anemia
. Fanconi anemia
. Hemolytic anemias
. Hemophagocytic lymphohistiocytosis
. Hereditary spherocytosis
. Megaloblastic anemia
. Seckel syndrome
. Sitosterolemia
. Thrombocytopenia

BION Genetic Laboratory

Immunology panel

Immunology panel

Immunology panel is our solution for immunodeficiency and severe combined immunodeficiency (SCID) disorders. Our panel includes genes targeting severe combined immunodeficiency, congenital neutropenia, primary antibody deficiency, common variable immune deficiency, chronic granulomatous disease, autoimmune lymphoproliferative, afibrinogenemia, hemolytic uremic syndrome, and agammaglobulinemia.

No. of genes: 330
TAT: 4-6 weeks
Coverage: ≥99.00% ≥20x
Details: NGS including CNV analysis

COMMON SYNDROMES AND DISORDERS COVERED

. Agammaglobulinemia
. Autoimmune lymphoproliferative syndrome
. B-cell-negative severe combined immunodeficiency
. B-cell-positive severe combined immunodeficiency
. Bare lymphocyte syndrome
. Chronic granulomatous disease
. Common variable immune deficiency
. Complement deficiency
. Congenital afibrinogenemia
. Congenital neutropenia syndromes
. Hermasky-Pudlak syndrome
. Hemolytic uremic syndrome
. Mendelian susceptibility to mycobacterial diseases
. Periodic fever syndrome
. Primary antibody deficiency
. Primary ciliary dyskinesia
. Primary immunodeficiencies (PID)
. Psoriasis
. Severe combined immunodeficiency

MOH NO: 167/2023

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Infertility panel

Our infertility panel is recommended for patients trying to conceive for one year or longer, with known fertility problems, who have experienced more than one miscarriage, with irregular or absent menstruation, with low sperm count, form, or movement, or with absence of development of secondary sexual features. Our panel includes the most important genes related to infertility in males and females. Knowing the exact cause of infertility allows for better diagnostic decisions and enables enhanced counseling for couples.

No. of genes: 276
TAT: 4-6 weeks
Coverage: ≥99.00% ≥20x
Details: NGS including CNV analysis
Sanger sequencing: CFTR; Repeat expansion analysis: FMR1;
Y chromosome microdeletions (according to order)

COMMON SYNDROMES AND DISORDERS COVERED

- Androgen insensitivity syndrome
- Female infertility
- Germline aneuploidy of chromosomes 13, 18, 21 and X
- Hypogonadotropic hypogonadism
- Klinefelter syndrome
- Male infertility
- Ovarian hyperstimulation syndrome
- Premature ovarian failure
- Primary ciliary dyskinesia
- Spermatogenic failure
- Turner Syndrome
- Y chromosome microdeletions

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Infertility panel (male)

Approach into male infertility panel

Indications	Genetic test recommendation
1. Men with infertility of unknown etiology and sperm concentrations < 10 million/mL who are candidates for ART	Y chromosome microdeletion and G-band karyotyping
2. Non-obstructive azoospermia in a male considering testicular sperm retrieval for ART	Y chromosome microdeletion and G-band karyotyping
3. Azoospermic or oligozoospermic men with the absence of at least one vas deferens at physical examination	CFTR gene mutation analysis
4. Azoospermic men with signs of normal spermatogenesis (e.g., obstructive azoospermia of unknown origin)	CFTR gene mutation analysis
5. History of recurrent miscarriage or personal/-familiar history of genetic syndromes	G-band karyotyping

ART= assisted reproductive techniques.
G-band karyotyping = Giemsa band karyotyping.
CFTR = cystic fibrosis transmembrane conductance regulator.

TAT: 10-14 days
Details: Y chromosome microdeletion, CFTR gene mutation analysis, Karyotyping

MOH NO: 167/ 2023

BION Genetic Laboratory

Ataxia/Spastic paraplegia comrpehensive panel

Ataxia / Spastic paraplegia comprehensive panel

Our Ataxia / Spastic paraplegia panel includes genes relevant to hereditary neurological disorders characterized by ataxia and spastic paraplegia, including spinocerebellar ataxia (dominant and recessive), cerebellar ataxia, episodic ataxia, and pontocerebellar ataxia. These disorders normally share overlapping symptoms and can only be clearly differentiated by molecular genetic testing.
Traditionally, ataxias and spastic paraplegia have been classified into separate categories. However, recent information shows that these diseases share genes, pathways and mechanisms and therefore our panel covers both syndromes and involves ataxia-spasticity disease spectrum.
Our Ataxia / Spastic paraplegia panel is not only the best option for patients displaying gait imbalance and uncoordinated walking, but also for patients displaying spastic gait impairment, spastic weakness, and hyperreflexia or any of the combinations.The most common forms of inherited ataxia are caused by repeat expansion mutations, therefore the comprehensive version of our panel includes repeat expansion analysis.

No. of genes: 493
TAT: 4-6 weeks
Coverage: ≥99.00% ≥20x
Details: NGS including CNV analysis
Repeat expansion analysis: ATN1, ATXN1, ATXN10, ATXN2, ATXN3, ATXN7, ATXN8OS, BEAN1, CACNA1A, FXN, NOP56, PPP2R2B, TBP

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Ataxia/Spastic paraplegia repeat expansion panel

Ataxia repeat expansion panel

No. of genes: 13
TAT: 4-6 weeks
Coverage: ≥100.00%
Details: Repeat expansion detection
Repeat expansion analysis: ATN1, ATXN1, ATXN10, ATXN2, ATXN3, ATXN7, ATXN8OS, BEAN1, CACNA1A, FXN, NOP56, PPP2R2B, TBP

COMMON SYNDROMES AND DISORDERS COVERED

. Cerebellar ataxia
. Episodic ataxia
. Pontocerebellar hypoplasia
. Spinocerebellar ataxia

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Neurology panel

Neurology panel

This panel is our largest panel, designed to detect an array of neurological disorders from neonatal ICU cases to dementia or movement disorders in adults.
This panel includes genes related to neurological diseases, such as amyotrophic lateral sclerosis, dementia, Parkinson's, neuromuscular diseases, Charcot-Marie-Tooth, dystonia, epilepsy, autism, intellectual disability, migraine, spastic paraplegia, ataxia, Leigh syndrome, peroxisomal diseases, epileptic encephalopathies, and movement disorders, among others. **Please consider that it does not include repeat expansion analysis.** If suspicion of neurological disorders caused by repeat expansions, we recommend that physicians orders one of our disease specific panels. **If there is high suspicion of Duchenne muscular dystrophy, we recommend that clinicians order deletion / duplication analysis by MLPA targeted to the DMD gene.**

No. of genes: 1902
TAT: 4-6 weeks
Coverage: ≥99.00% ≥20x
Details: NGS including CNV analysis

MOH NO: 167/2023

COMMON SYNDROMES AND DISORDERS COVERED

- Arthrogryposis multiplex congenita
- Ataxia
- Dementia
- Dolichoectasia
- Dystonia
- Epilepsy
- Familial hemiplegic migraine
- Frontotemporal dementia
- Hypogonadotropic hypogonadism
- Intellectual disability
- Joubert syndrome
- Kallman syndrome
- Leigh syndrome
- Leukodystrophy and peroxisome biogenesis disorders
- Meckel syndrome
- Mitochondrial encephalomyopathy
- Neonatal mitochondrial hepatopathies
- Neuromuscular disorders
- Parkinson´s disease
- Refsum disease
- Spastic paraplegia
- Tuberous sclerosis
- Zellweger syndrome

BION Genetic Laboratory

Epilepsy panel

Epilepsy panel

While some types of seizures are easily categorized (i.e., partial or generalized), others are not or might later develop into different types (i.e., partial seizures with secondary generalization) making targeted panel testing less likely to succeed at reaching a diagnosis. Our epilepsy panel is phenotype-directed and covers different types of seizure syndromes, covering Dravet syndrome, early infantile epileptic encephalopathy, epilepsy partial, epilepsy generalized, epilepsy absence, myoclonic epilepsy panel, and hypomagnesemia.
In addition, our panel includes mitochondrial and nuclear mitochondrial genes (i.e., genes causing myoclonic epilepsy with ragged red fibers –MERRF–).

No. of genes: 783
TAT: 4-6 weeks
Coverage: ≥99.50% ≥20x
Details: NGS including CNV analysis, Repeat expansion analysis: CSTB

MOH NO: 167/2023

COMMON SYNDROMES AND DISORDERS COVERED

- Aicardi-Goutieres syndrome
- Brain iron accumulation syndromes
- Congenital glycosylation disease
- Dravet syndrome
- Early infantile epileptic encephalopathy
- Epilepsy
- Epilepsy (absence) in childhood
- Epilepsy (generalized) with febrile seizures
- Epilepsy (partial)
- Epileptic encephalopathy
- Hypomagnesemia
- Leigh syndrome
- Leukodystrophy and peroxisome biogenesis disorders
- Lysosomal storage disease
- Mitochondrial DNA depletion
- Mitochondrial encephalomyopathy
- Muscular dystrophy-dystroglycanopathy
- Myoclonic epilepsy
- Urea cycle disorder

BION Genetic Laboratory

Intellectual disability panel

Intellectual disability panel

Our panel includes genes associated with intellectual disabilities covering all mechanisms of inheritance as well as syndromic and non-syndromic autism, microcephaly, neuronal migration disorders, developmental regression, and Aicardi Goutierres. Detection of Fragile X syndrome is possible, as our panel includes repeat expansion of FMR1.

No. of genes: 819
TAT: 4-6 weeks
Coverage: ≥99.00% ≥20x
Details: NGS including CNV analysis
Repeat expansion analysis: FMR1

COMMON SYNDROMES AND DISORDERS COVERED

- Aicardi-Goutieres syndrome
- Bardet-Biedl syndrome
- Epileptic encephalopathy
- Intellectual disability AD, AR, XL
- Micro syndrome
- Microcephaly
- Neurodevelopmental disorders
- Neuronal migration disorders
- Syndromic autism

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BION Genetic Laboratory

Neuromuscular panel

Neuromuscular panel

Our neuromuscular panel is ideal for patients with muscular diseases. It includes genes causing neurological diseases and covers disorders, such as metabolic myopathies, muscular dystrophies, Charcot-Marie-Tooth, congenital myasthenic syndromes, congenital myopathies, myofibrillar myopathies, nemaline myopathies, and other syndromes with hypotonia, myotonia, or weakness. Arthrogryposis is included for differential diagnosis of early-onset neuromuscular disorders.
If there is high diagnostic suspicion for Duchenne muscular dystrophy, we recommend that the clinician orders deletion/duplication analysis by MLPA targeted to the DMD gene as an additional service.

No. of genes: 354
TAT: 4-6 weeks
Coverage: ≥99.00% ≥20x
Details: NGS including CNV analysis
Repeat expansion analysis: DMPK
E7 homozygous deletion screening: SMN1

COMMON SYNDROMES AND DISORDERS COVERED

- Arthrogryposis
- Bethlem myopathy
- Charcot–Marie–Tooth disease
- Congenital myasthenic syndrome
- Congenital myopathy
- Dejerine-Sottas syndrome
- Hyperekplexia
- Hypotonia
- Malignant hyperthermia
- Metabolic myopathies
- Muscular dystrophy
- Muscular dystrophy-dystroglycanopathy type A
- Myofibrillar myopathy
- Myopathy-rhabdomyolysis syndrome
- Nemaline myopathy
- Non-dystrophic myotonia congenita
- Spinal muscular atrophy type 1
- Ullrich muscular dystrophy

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BION Genetic Laboratory

Non-Invasive Prenatal Testing

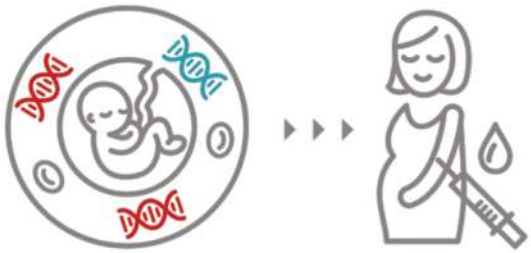
No Risk to Mother and Baby

Non-Invasive Prenatal Testing(NIPT)

Want to make sure that your baby is growing and developing normally during pregnancy? NIPT – Safe and accurate prenatal testing – Offers you information about the health and development of your child from as early as the 10th week of pregnancy.

Non-invasive prenatal testing (NIPT) is a new method of testing for common chromosomal abnormalities that can occur in a developing baby. While rare, these chromosome abnormalities can have profound consequences to the life and health of you and your child and it is important to find out as soon as possible.

Initial screening with non-invasive prenatal testing can help to avoid this potentially unnecessary and invasive testing. There is no risk to mother or baby and NIPT provides the earliest testing available.



FETAL DNA MATERNAL DNA

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Non-Invasive Prenatal Testing

No Risk to Mother and Baby

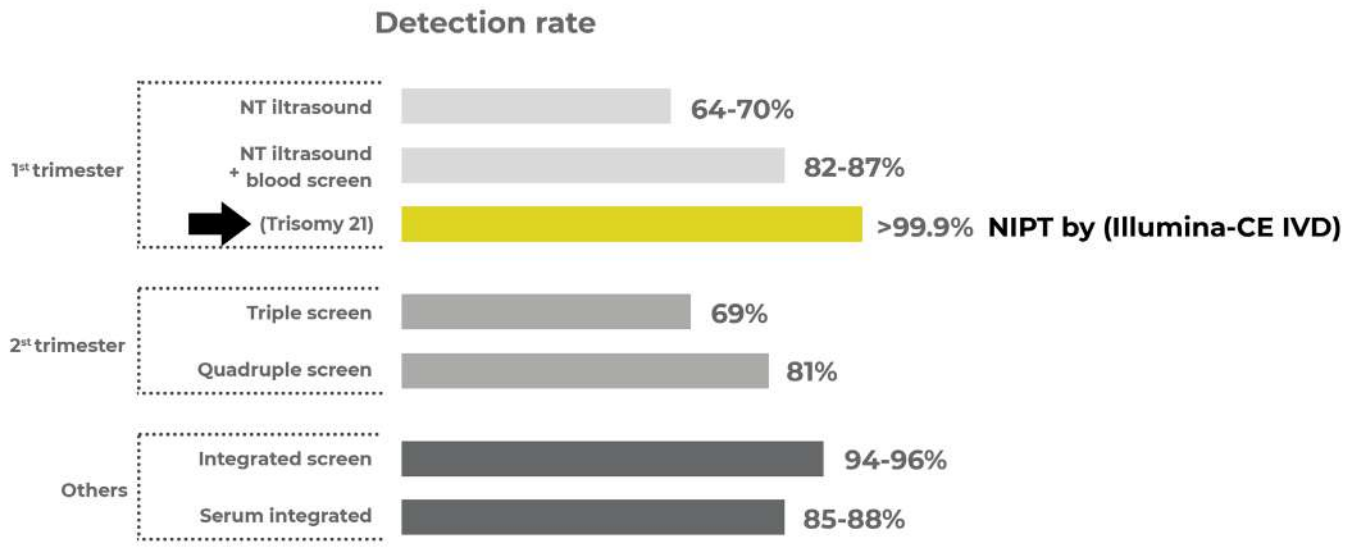
What Does NIPT Screen For?

1. Down syndrome (Trisomy 21), which is caused by an extra copy of chromosome 21. Mild/moderate mental retardation and risk of some birth defects (affects 1 in 1,000 live births).
2. Edwards syndrome (Trisomy 18), which is caused by an extra copy of chromosome 18. Severe mental retardation and risk of multiple birth defects (affects 1 in 3,000-6,000 live births).
3. Patau syndrome (Trisomy 13), which is caused by an extra copy of chromosome 13. Severe mental re tardation and risk of multiple birth defects (affects 1 in every 5,000 live births).

The test can also detect abnormalities of the sex chromosomes:

4. Turner syndrome (Monosomy X), which is caused by a missing X chromosome in females
5. Klinefelter syndrome (XXY), which is caused by an extra X chromosome in males
6. Jacobs syndrome (XYY), which is caused by an extra Y chromosome in males
7. Triple X syndrome (XXX), which is caused by an extra X chromosome in females

MOH NO: 167/2023



BION Genetic Laboratory

Inborn Errors of Metabolism (IEM)

Inborn Errors of Metabolism (IEM)

Inborn Errors of Metabolism (IEM) largely impact human diseases.

CentoIEM is a metabolic and liver disease gene panel that screens for an array of different disorders and contains genes responsible for diverse phenotypes, including intermediary metabolism, such as aminoacidopathies, organic acidurias, urea cycle disorders, sugar intolerance, mental disorders, and porphyrias, among others.

Genes linked to cytoplasmic and mitochondrial energetic processes and metabolism affecting cellular organelles, such as lysosomal, peroxisomal, glycosylation, and cholesterol synthesis are also included.

No. of genes: 744
TAT: 4-6 weeks
Coverage: ≥99.00% ≥20x
Details: NGS including CNV analysis

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Inborn Errors of Metabolism (IEM)

COMMON SYNDROMES AND DISORDERS COVERED

- Aicardi-Goutieres syndrome
- Autoimmune lymphoproliferative syndrome
- Ceroid lipofuscinosis
- Congenital glycosylation disease
- Familial hypercholesterolemia
- Fatty acid oxidation disorder
- Fatty liver disease
- Glycogen storage disease
- Hemophagocytic lymphohistiocytosis
- Hereditary hemochromatosis
- Hereditary spherocytosis
- Leigh syndrome and mitochondrial encephalopathy
- Leukodystrophy and peroxisome biogenesis disorders
- Lipodystrophy syndromes
- Lysosomal storage disease
- Maple syrup urine disease
- Methylmalonic acidemia
- Mucopolysaccharidosis
- Neurodegeneration with Brain Iron Accumulation
- Non-ketotic hyperglycinemia
- Organic acidemias
- Porphyria
- Refsum disease
- Urea cycle disorder

MOH NO: 167/2023

BION Genetic Laboratory

Dysmorphology

Dysmorphology

Congenital malformations **("birth defects")** remain a leading cause of infant mortality and childhood morbidity. We offer comprehensive and rapid testing options, including analysis of genome-wide copy number alterations. Testing can provide your patients with a clear diagnosis of inherited malformation and intellectual disability syndromes.

This panel is designed to help physicians diagnose patients that suffer from a dysmorphic syndrome. The panel includes craniosynostosis, craniofacial disorders, cleft / lip palate, holoprosencephaly, Waardenburg syndrome, Hirschsprung disease, lissencephaly, and brain malformation disorders, among others.

Additionally, this panel includes genes related to RASopathies. RASopathies are a group of genetic syndromes caused by germline mutations in genes that encode components or regulators of the RAS / mitogen-activated protein kinase (MAPK) pathway.
This panel includes genes related to neurofibromatosis type 1, Noonan syndrome, Noonan syndrome with multiple lentigines, capillary malformation-arteriovenous malformation syndrome, Costello syndrome, Cardio-Facio- Cutaneous syndrome, and Legius syndrome, among others. Tuberous sclerosis and McCune Albright syndromes.

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BION Genetic Laboratory

Dysmorphology

No. of genes: 776
TAT: 4-6 weeks
Coverage: ≥99.00% ≥20x
Details: NGS including CNV analysis

COMMON SYNDROMES AND DISORDERS COVERED

. Bardet-Biedl syndrome
. Cardiofaciocutaneous syndrome
. Cerebral cavernous malformations
. Ciliopathies
. Cleft lip and palate
. Coffin-Siris syndrome
. Cornelia de Lange syndrome
. Ciliopathic skeletal dysplasias
. Craniosynostosis and craniofacial disorders
. Heterotaxy syndrome
. Hirschsprung disease
. Holoprosencephaly
. Klippel-Feil syndrome
. Lissencephaly and brain malformation
. Meckel syndrome
. Metaphyseal dysplasia
. Micro syndrome
. Microphthalmia/anophthalmia/coloboma spectrum
. Multiple epiphyseal dysplasia
. Neurofibromatosis
. Noonan-RASopathies syndromes
. Seckel syndrome
. Skeletal dysplasia extended
. Stickler syndrome
. Tuberous sclerosis
. Waardenburg syndrome

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BION Genetic Laboratory

Enzymes and Biomarkers Test for New-born

Enzymes and Biomarkers Test for New-born

A genetic test alone may not be able to provide the information needed for a final diagnosis.

A) Enzyme assays use for detection of these disorders

1. Oligosaccharidosis and Sphingolipidoses

- Wolman disease (Acid lipase)
- Pompe disease (Alpha-glucosidase)
- Fucosidosis (Alpha-fucosidase)
- Fabry disease (Alpha-galactosidase)
- Alpha-mannosidosis (Alpha-mannosidase)
- Schindler/Kanzaki disease (Alpha-N-acetylgalactosaminidase)
- Gaucher disease (Beta-glucocerebrosidase)
- Tay-Sachs disease (Beta-hexosaminidase)
- Beta-mannosidosis (Beta-mannosidase)
- Sandhoff disease (Total-hexosaminidase)

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Enzymes and Biomarkers Test for New-born

2. Neuronal Ceroid Lipofuscinosis

- Santavuori-Haltia disease (Palmitoyl-protein- thioesterase)
- Jansky-Bielschowsky disease (Tripeptidyl-peptidase)

3. Mucopolysaccharidosis

- Hurler syndrome (MPS I) (Alpha-L-iduronidase)
- Hunter syndrome (MPS II) (Iduronate-2-sulfatase)
- Sanfilippo syndrome B (MPS III B) (Alpha-N-acetylglucosaminidase)
- Morquio syndrome A (MPS IV A) (N-acetylgalactosamine-6-sulfate-sulfatase)
- Morquio syndrome B (MPS IV B) (Beta-galactosidase)
- Maroteaux-Lamy syndrome (MPS VI) (Arylsulfatase B)
- Sly syndrome (MPS VII) (Beta-glucuronidase)

B) Biomarkers assays use for detection of these disorders

1. Gaucher disease (Glucosylsphingosine (lyso-Gb1))
2. Fabry disease (Lyso-ceramide trihexoside (lyso-Gb3))
3. Niemann-Pick disease type A/B/C (Lyso-SM-509)
4. Aromatic L-amino acid decarboxylase (AADC) deficiency (3-O-methyldopa (3-OMD))
5. Hereditary angioedema (HAE) (Complement C4-alpha peptide and Complement C1-INH pep tide

TAT: 21 working days

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BION Genetic Laboratory

Metabolic Panel

Metabolic Panel

Metabolic Panel was developed specifically for patients suspected of having a metabolic disorder or presenting complex, overlapping symptoms, a metabolic crisis, or neurological conditions of unknown etiology.
It provides short turnaround times –, targeting critically ill patients in NICU/PICU.
It leverages a multiomic approach by including enzyme -activity testing where applicable, as well as a proprietary selection of biomarkers that is continuously updated.

No. of genes: 206
TAT: 21 days
Coverage: ≥99.50% ≥20x
Details: NGS including CNV analysis

MOH NO: 167/ 2023

COMMON SYNDROMES AND DISORDERS COVERED

Congenital disorders of glycosylation and other disorders of protein modification
Defects in cholesterol and lipoprotein metabolism
Defects in hormone biogenesis or function

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Metabolic Panel

Metabolic Panel

- Disorder of phosphate, calcium and vitamin D metabolism
- Disorders in the metabolism of purines, pyrimidines and nucleotides
- Disorders in the metabolism of trace elements and metals
- Disorders in the metabolism of vitamins and (non-protein) cofactors
- Disorders of amino acid and peptide metabolism
- Disorders of carbohydrate metabolism
- Disorders of energy metabolism
- Disorders of fatty acid and ketone body metabolism
- Disorders of lipid and lipoprotein metabolism
- Disorders of neurotransmitter metabolism
- Disorders of porphyrin and heme metabolism
- Disorders of the metabolism of sterols
- Lysosomal disorders
- Peroxisomal disorders
- Porphyria and bilirubinemia

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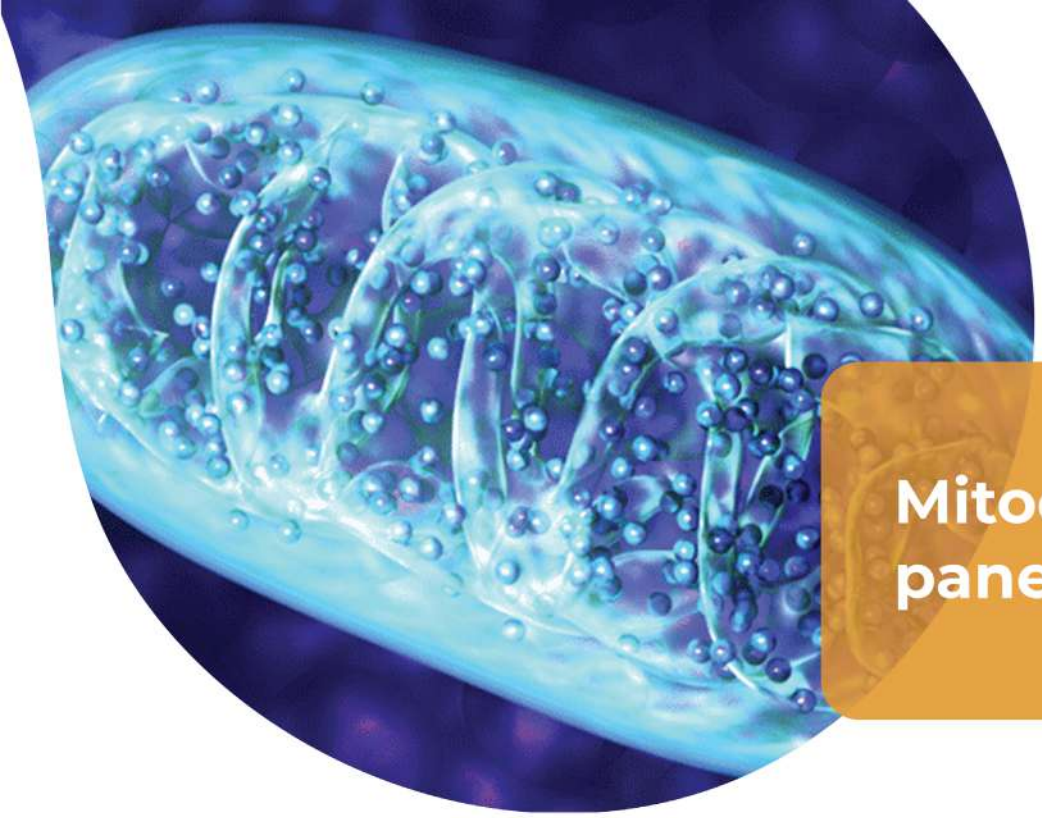

BION Genetic Laboratory

Mitochondrial panel

Mitochondrial panel

Mitochondrial panel includes mitochondrial genes. Nuclear mitochondrial genes are not included.

No. of genes: 37
TAT: 4-6 weeks
Coverage: ≥97.00% ≥200x
Details: NGS including CNV analysis

MOH NO: 167/2023

COMMON SYNDROMES AND DISORDERS COVERED

. Chronic progressive external ophthalmoplegia
. Kearns-Sayre syndrome
. Leber hereditary optic neuropathy
. Leigh-like syndrome
. Leigh syndrome
. Mitochondrial disorders
. NARP

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BION Genetic Laboratory

New born in ICU panel Comprehensive NGS panel

New born in ICU panel comprehensive NGS panel

It is a comprehensive NGS panel that includes more than 850 genes explicitly selected for the genetic testing of critically ill new-borns and children under 24 months in intensive care units (ICU). It is designed to address multiple genetic conditions that may be present in the new-born or early childhood period, with many having overlapping phenotypes and immediate implications for treatment initiation.

As you know, new-borns and children under 24 months presenting with life-threatening conditions need a fast and precise diagnosis to ensure rapid and efficient further diagnostic and therapeutic initiation. Up to one-third of all babies and children admitted to the ICU have a genetic disease. For many of them early identification can make the difference for their immediate and later health.

It is indicated for new-borns and children under 24 months admitted to the ICU and presenting with unclear symptomatology which can be part of a genetic condition, i.e.v

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New born in ICU panel
comprehensive NGS panel

. Bleeding dyathesis
. Blood abnormalities (anemia)
. Bone fragility
. Failure to thrive
. Heart abnormality/arrhythmia
. Hepatospenomegaly
. Hypotonia
. Ichthyosis/epidermolysis bullosa
. Metabolic abnormalities*
. Microcephaly
. Neutropenia
. Abnormal new-born screening results**
. Respiratory failure
. Skeletal abnormalities/craniosynostosis
. Skin fragility
. Unclear seizures

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* Abnormal acylcarnitine profile, amino acidemia/urea, hyperbilirubinemia, hyper-/hypoinsulinism, persistent hypoglycemia, organic acidemia/urea
** It includes genes to cover all ACMG core panel phenotypes for new-born screening except hearing loss

COMMON SYNDROMES AND DISORDERS COVERED

. Alagille Syndrome
. Alpha-Thalassemia
. Arginase Deficiency
. Beta-Thalassemia
. Biotinidase Deficiency
. Biotin-Thiamine-Responsive Basal Ganglia disease
. Carnitine Deficiency
. Congenital Hypothyroidism
. Cystic Fibrosis

BION Genetic Laboratory

New born in ICU panel
comprehensive NGS panel

. DYSTONIA DOPA RESPONSIVE
. FACTOR VII DEFICIENCY
. GLUCOSE TRANSPORTER 1 DEFICIENCY
. GLUTARIC ACIDEMIA TYPE 1
. HEREDITARY FRUCTOSE INTOLERANCE
. HOLOCARBOXYLASE SYNTHETASE DEFICIENCY
. MAPLE SYRUP URINE DISEASE (MSUD)
. NON KETOTIC HYPERGLICINEMIA
. PHENYLKETONURIA
. POMPE DISEASE
. PRIMARY COENZYME Q10 DEFICIENCY
. PYRIDOXAMINE 5 PHOSPHATE OXIDASE DEFICIENCY
. PYRIDOXINE-DEPENDENT EPILEPSY
. PYRUVATE CARBOXYLASE DEFICIENCY
. TUBEROUS SCLEROSIS COMPLEX
. TYROSINEMIA TYPE I
. VLCAD DEFICIENCY

*LIST DOES NOT INCLUDE ALL DISORDERS COVERED BY OUR PANEL COMMON SYNDROMES AND DISORDERS COVERED

No. of genes: 856
TAT: 21 working days
Coverage: ≥99.00% ≥20x

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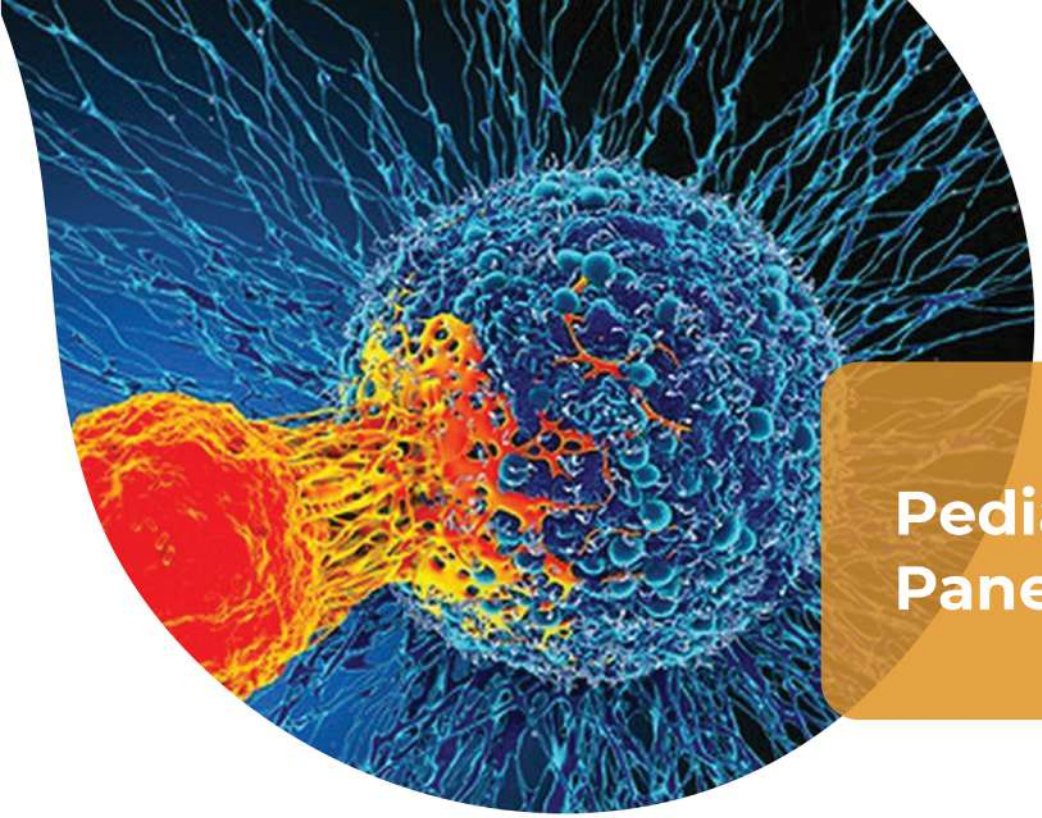

BION Genetic Laboratory

Pediatric Cancer Panel

Pediatric Cancer Panel

Cancer occurs in people of all ages, with some being nearly exclusively tied to childhood. Cento Pediatric Cancer Panel is our comprehensive solution to detect genes associated with pediatric cancer. The gene list has been carefully curated by internal and external experts to cover the most common forms of pediatric cancer. Germline mutations identified by these panel will help to define prognosis, differentiate patient/family risk, and guide treatment decisions. Spotting cancer early increases the chances of survival.

COMMON SYNDROMES AND DISORDERS COVERED

- Leukemia
- Malignant brain tumors
- Lymphomas
- Bone cancer
- Neuroblastoma
- Wilms tumor
- Rhabdomyosarcoma

No. of genes: 97
TAT: 21 days
Coverage: ≥99.00% 20x
Details: NGS including CNV analysis

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Carrier Screening with BION

Carrier Screening with BION

Most people can be carriers of a disease causing change without knowing it.

If both partners are carriers, they have a 25% risk of having an affected child with a recessive genetic disease and a 50% that the child will be a carrier like the parents. Getting tested early gives future parents the opportunity to make informed decisions and review the range of options available to guide pregnancy and family planning.

The results indicate a very high prevalence of consanguineous marriage in Oman, as more than half (52%) of marriages are consanguineous. First cousin unions are the most common type of consanguineous unions, constituting 39% of all marriages and 75% of all consanguineous marriages.

Many studies showed that consanguinity increases the prevalence of birth defects and other genetic disorders.
Carrier screening test helps the family to reduce incidence of genetic disorders.

No. of genes: 330
TAT: 4-6 weeks
Details: NGS including CNV analysis, Additional analyses for fragile X syndrome, spinal muscular atrophy, and congenital adrenal hyperplasia (FMR1, SMN1 and CYP21A2 genes respectively)

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BION Genetic Laboratory

Vision panel

Vision panel

Vision panel is carefully designed to find the genetic basis of eye diseases, including those that are the leading causes of blindness among infants (Leber congenital amaurosis), children (early-onset retinitis pigmentosa), and adults (pattern dystrophy). Our panel includes the most common ophthalmology diseases, such as congenital glaucoma, retinitis pigmentosa, Stargardt disease, Stickler syndrome, achromatopsia, and Usher syndrome, among others. It also screens for different types of albinism (oculocutaneous and ocular) as well as Hermasky-Pudlak syndrome.

No. of genes: 450
TAT: 4-6 weeks
Coverage: ≥99.00% ≥20x
Details: NGS including CNV analysis

MOH NO: 167/2023

COMMON SYNDROMES AND DISORDERS COVERED

- Achromatopsia
- Albinism
- Bardet-Biedl syndrome
- Cataract
- Cone-rod and cone dystrophy

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Vision panel

- Flecked retina
- Glaucoma
- Hermansky-Pudlak syndrome
- Leber congenital amaurosis
- Meckel syndrome
- Microphthalmia/anophthalmia/coloboma spectrum
- Oculomotor apraxia
- Optic atrophy
- Progressive external ophthalmoplegia
- Retinitis pigmentosa, autosomal dominant
- Retinitis pigmentosa, autosomal recessive
- Stargardt disease
- Stickler syndrome
- Usher syndrome
- Vitreoretinopathy
- Wagner syndrome

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BION Genetic Laboratory

Pulmonary panel

Pulmonary panel

Our pulmonary panel includes genes for the diagnosis of central hypoventilation, surfactant metabolism dysfunction, pulmonary hypertension among other pulmonary diseases.

No. of genes: 101
TAT: 4-6 weeks
Coverage: ≥99.00% ≥20x
Details: NGS including CNV analysis
Repeat expansion analysis: PHOX2B

MOH NO: 167/2023

COMMON SYNDROMES AND DISORDERS COVERED

- Central hypoventilation syndrome
- Comprehensive pulmonary disease
- Pulmonary hypertension
- Surfactant metabolism dysfunction

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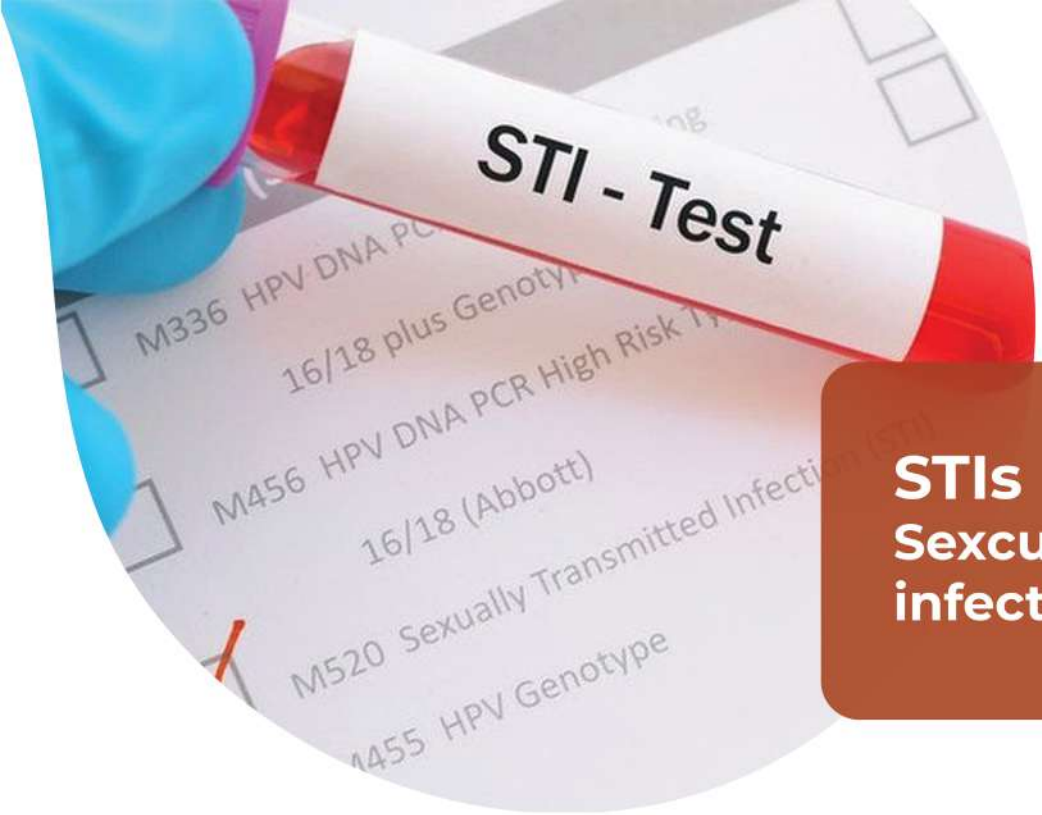

BION Genetic Laboratory

STIs
Sexcually transmitted infections

STIs:

Testing for specific STIs

These guidelines for specific STIs can help you decide if STI testing is right for you.

Chlamydia and gonorrhea

National guidelines recommend yearly screening for:

- Sexually active women under age 25
- Women older than 25 and at increased risk of STIs — such as having sex with a new partner or multiple partners
- Men who have sex with men
- People with HIV
- Transgender women who have sex with men
- People who have been forced to have intercourse or engage in sexual activity against their will

Health care providers screen people for chlamydia and gonorrhea using a urine test or swab. Swabs are taken inside the penis in men or from the cervix in women. The sample is then studied in a lab. Screening is important, because if you don't have symptoms, you may not know that you're infected.

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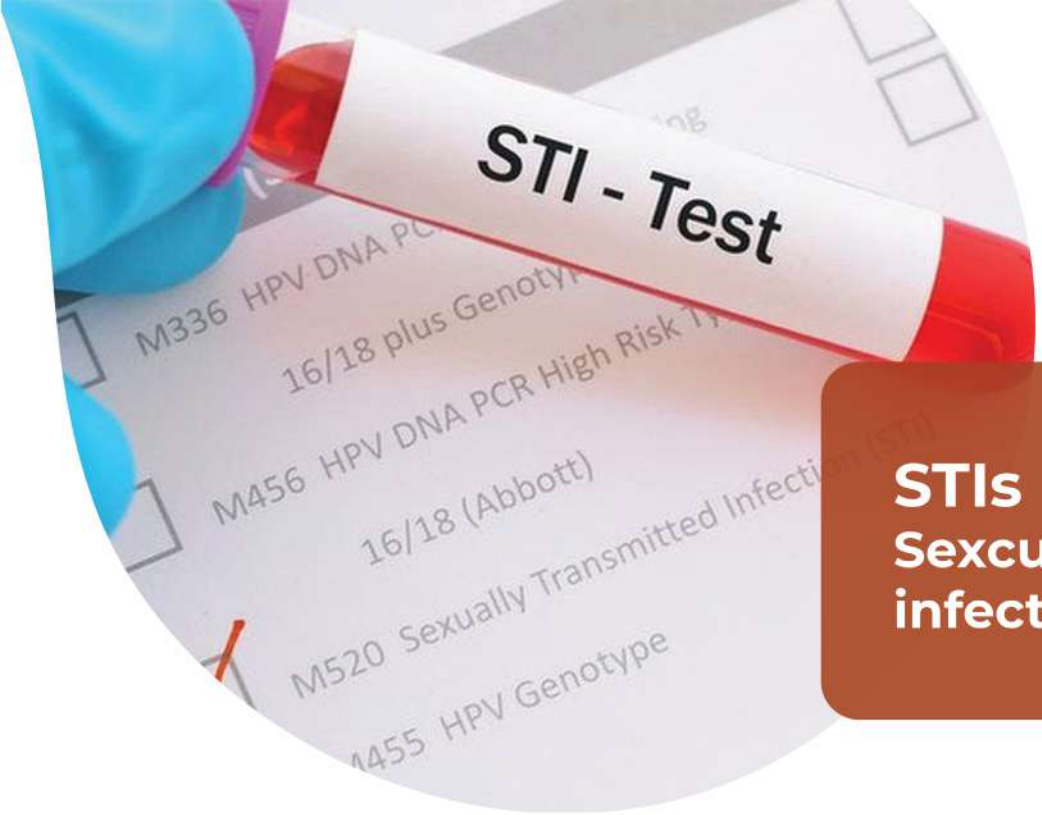

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STIs
Sexcually transmitted infections

HIV, syphilis and hepatitis

The U.S. Preventive Services Taskforce encourages HIV testing, at least once, as a routine part of health care if you're between the ages of 15 and 65. Younger teens or older adults should be tested if they have a high risk of an STI. The Centers for Disease Control and Prevention (CDC) advises at least yearly HIV testing if you're at high risk of infection.

National guidelines recommend hepatitis C screening for all adults ages 18 to 79. Vaccines are available for hepatitis A and B and are usually given at birth. Unvaccinated adults can be vaccinated if they are at high risk of getting hepatitis A or B.

If you have any of the following risk factors, talk to your health care provider about testing for HIV, syphilis or hepatitis:

- Symptoms of infection
- Positive test for another STI, which puts you at greater risk of other STIs
- Having more than one sexual partner (or if your partner has had multiple partners) since your last test
- Intravenous (IV) drug use
- Men who have sex with men
- Being pregnant or planning to become pregnant
- Being forced to have intercourse or engage in sexual activity against your will

Your health care provider tests you for syphilis by taking either a blood sample or a swab from any genital sores you might have. A lab specialist studies the sample in a lab. Your provider also takes a blood sample to test for HIV and hepatitis.

Genital herpes

Providers generally only recommend testing for genital herpes for people who have symptoms or other risk factors. But most people with herpes infection never have any symptoms but can still spread the virus to others. Your health care provider may take a tissue sample or culture of blisters or early ulcers, if you have them, and send them to a lab. But a negative test doesn't always mean you don't have herpes, especially if you have symptoms.

A blood test also may tell if you had a past herpes infection, but results aren't always reliable. Some blood tests can help providers see which of the two main types of the herpes virus you have. Type 1 is the virus that usually causes cold sores, although it can also cause genital sores.

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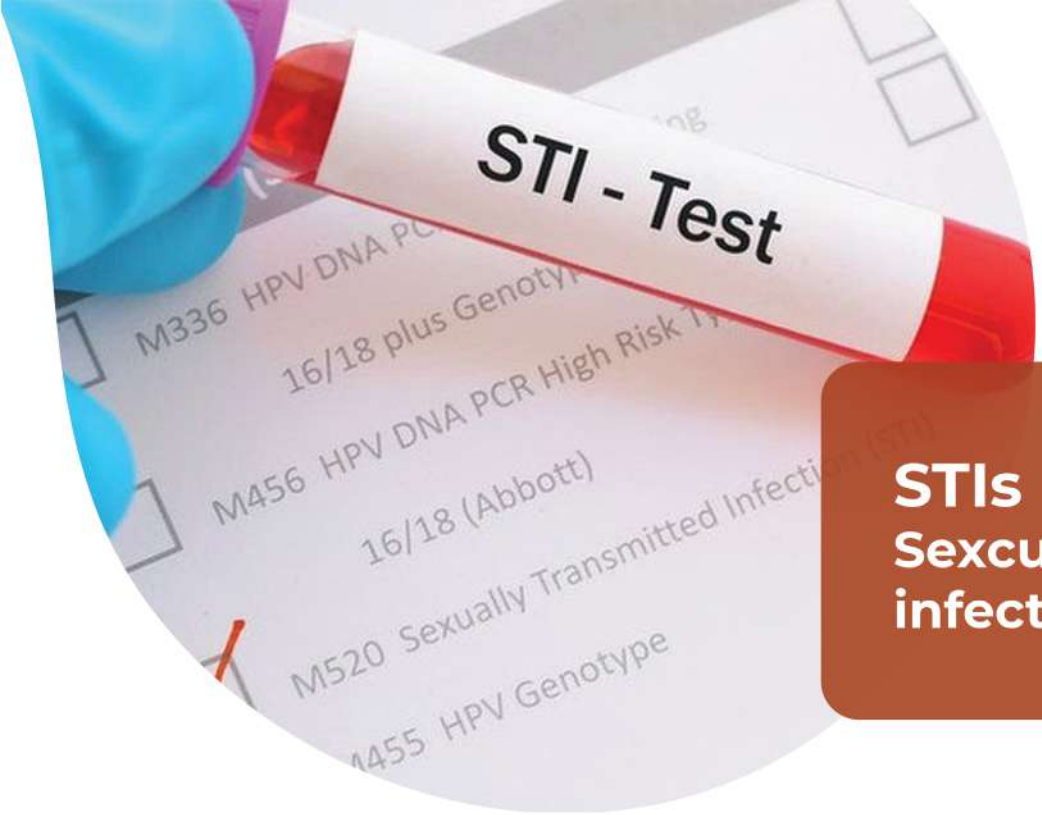

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STIs
Sexcually transmitted infections

Type 2 is the virus that causes genital sores more often. Still, the results may not be clear, depending on how sensitive the test is and the stage of the infection. False-positive and false-negative results are possible.

HPV

Certain types of human papillomavirus (HPV) can cause cervical cancer. Other types of HPV can cause genital warts. Many sexually active people get HPV at some point in their lives but never have symptoms. Most of the time, the virus goes away on its own within two years.

Regular HPV testing isn't recommended for men. Instead, health care providers may choose to test men who have symptoms, such as genital warts. A sample of the wart is removed and sent to a lab. In women, **HPV testing involves:**

- **Pap test.** Pap tests, which check the cervix for irregular cells, are recommended every three years for women between ages 25 and 65.
- **HPV test.** Women between ages 25 and 65 should have an HPV test alone or an HPV test along with a Pap test every five years if previous test results were within the standard range. Testing may take place more often for those who are at high risk of cervical cancer or those who have irregular results on their Pap or HPV tests.

HPV is also linked to cancer of the vulva, vagina, penis, anus, and mouth and throat. Vaccines can protect both men and women from some types of HPV. But they're most effective when given between ages 9 and 26

TAT : 2-4 days

Details: Bion do all STI test by real time PCR with high Accuracy and highly sensitive KITs and Methods.

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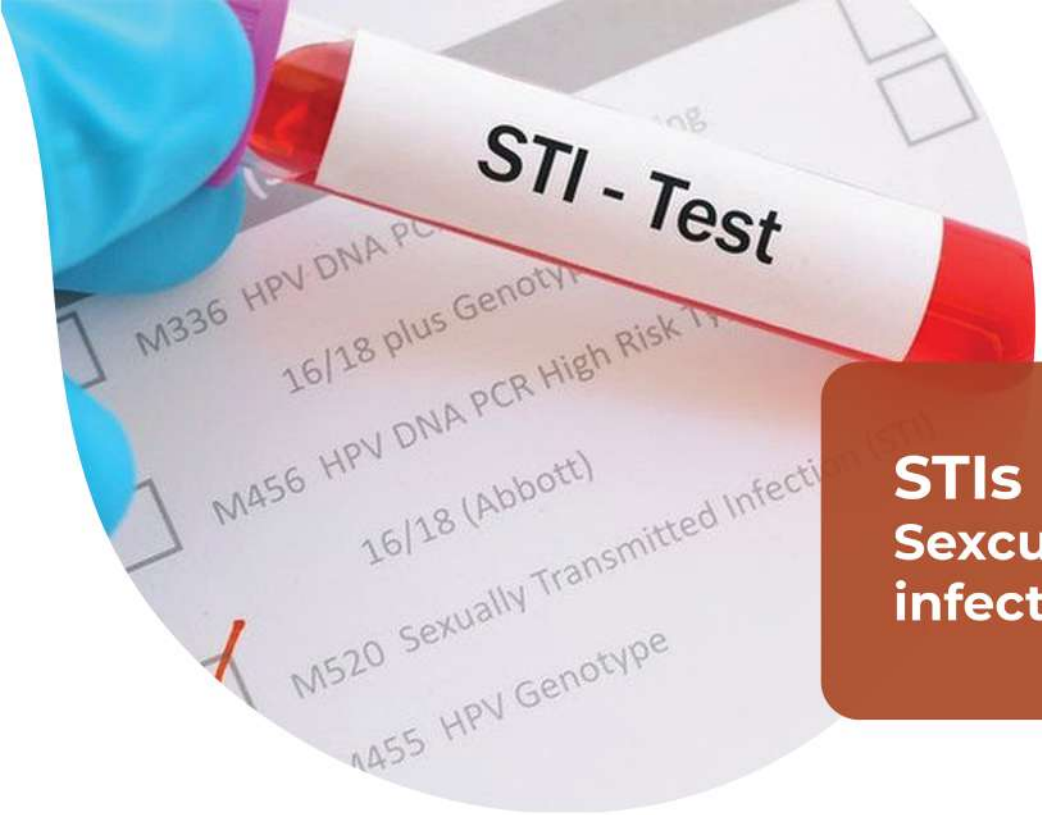

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STIs
Sexcually transmitted infections

Test	Method	Specimen	Comment
STI	Multiplex real-time PCR for detection of pathogen genes by TaqMan® technology	Specific sampling up to the symptoms, such as: . Vesicle fluid . Epithelial cell scrap . And ...	12 Pathogens: . Chlamydia trachomatis . Mycoplasma genitalium . Neisseria gonorrhoeae . Trichomonas vaginalis . Ureaplasma urealyticum . Ureaplasma parvum . Mycoplasma hominis . Treponema pallidum . Candida albicans . Gardnerella vaginalis . HSV-1 / HSV-2 **Report Time: 48 hours**

MOH NO: 167/2023

All infectious diagnosis tests are performed in the shortest possible time with TaqMan® technology and using ROCHE LC480 Realtime-PCR machine.

The BION Advantage

Processing time: **In a short time after sample receipt**

Highest confidentiality and quality standards

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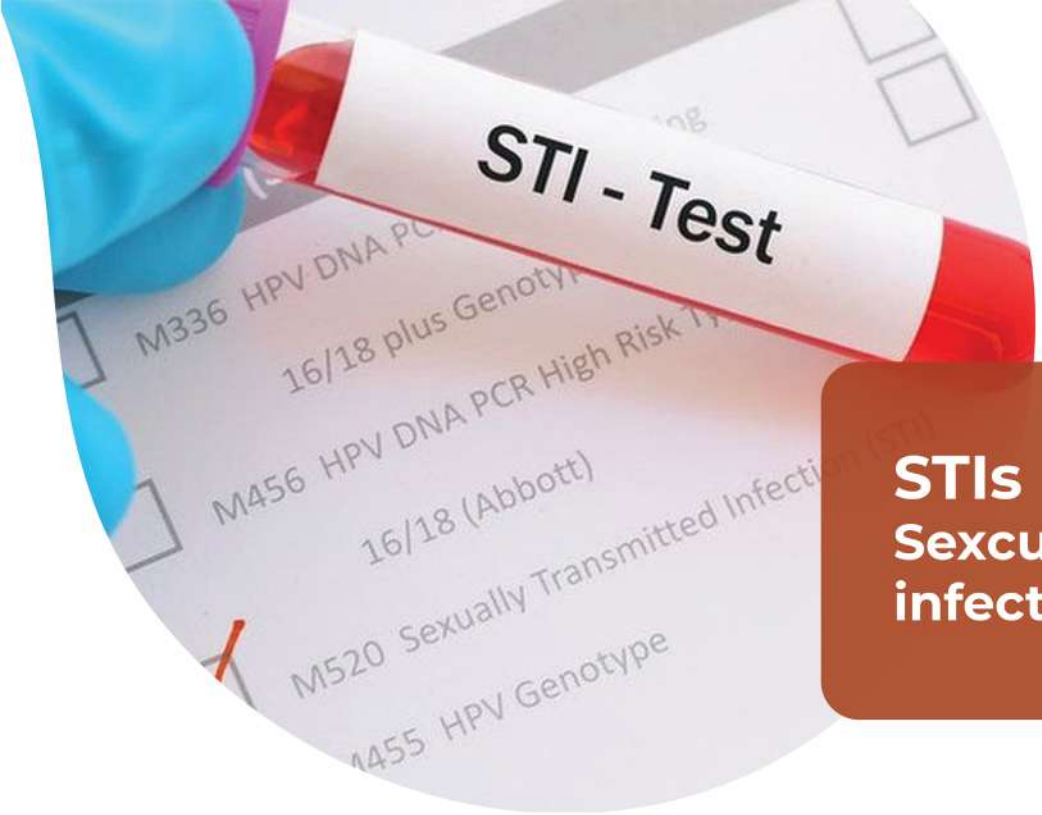

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STIs
Sexcually transmitted infections

Test	Method	Specimen	Comment
HPV	Genotyping Real-time PCR for detection of types by TaqMan® technology	Specific sampling up to the symptoms, such as: . Vaginal Swab . Epithelial cell scrap . Wart biopsy . Urine . And ...	High-Risks genotypes: . 31, 33, 35, 39, ... especially16 & 18 **Report Time: 48 hours**
HIV	Real-time PCR for detection of pathogen genes by TaqMan® technology	. Plasma . Serum	. Qualitative . Quantitative (Viral load) **Report Time: 24 hours**
HSV	Real-time PCR for detection of pathgen genes by TaqMan® technology	Specific sampling up to the symptoms, such as: . Vesicle fluid . Epithelial cell scrap . And ...	. Qualitative . HSV1 & HSV2 **Report Time: 24 hours**

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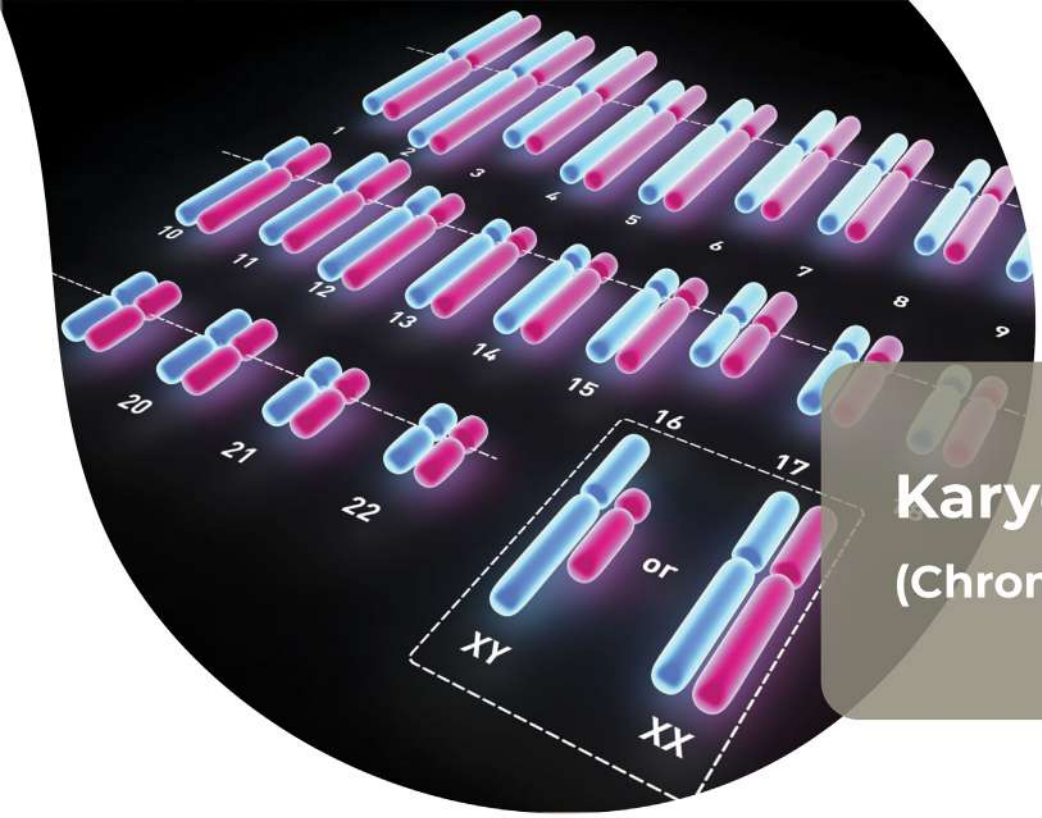

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Karyotyping test
(Chromosome analysis)

Karyotyping test
(Chromosome analysis)

Genetic karyotyping—also known as chromosome analysis—is testing that can reveal certain genetic abnormalities. It can be used to confirm or diagnose a genetic disorder or disease. Or, the testing may reveal that a couple is at risk for having a child with a genetic or chromosomal disorder.

It may be recommended genetic karyotyping if:

- You've been unable to conceive for more than a year.
- You've experienced two or more consecutive miscarriages.
- You've experienced a stillborn birth.
- The male partner has no sperm in his semen or an extremely low sperm count. (Also known as azoospermia or severe oligozoospermia.)
- The female partner has been diagnosed with primary ovarian dysfunction. (Also known as POI, primary ovarian insufficiency, or POF, premature ovarian failure.)

Genetic karyotyping may be required before receiving assisted reproductive technology, including IUI or IVF. This is especially true for those considering IVF with ICSI, which increases the risk of passing on male infertility and some genetic disorders.

TAT: 10-14 days

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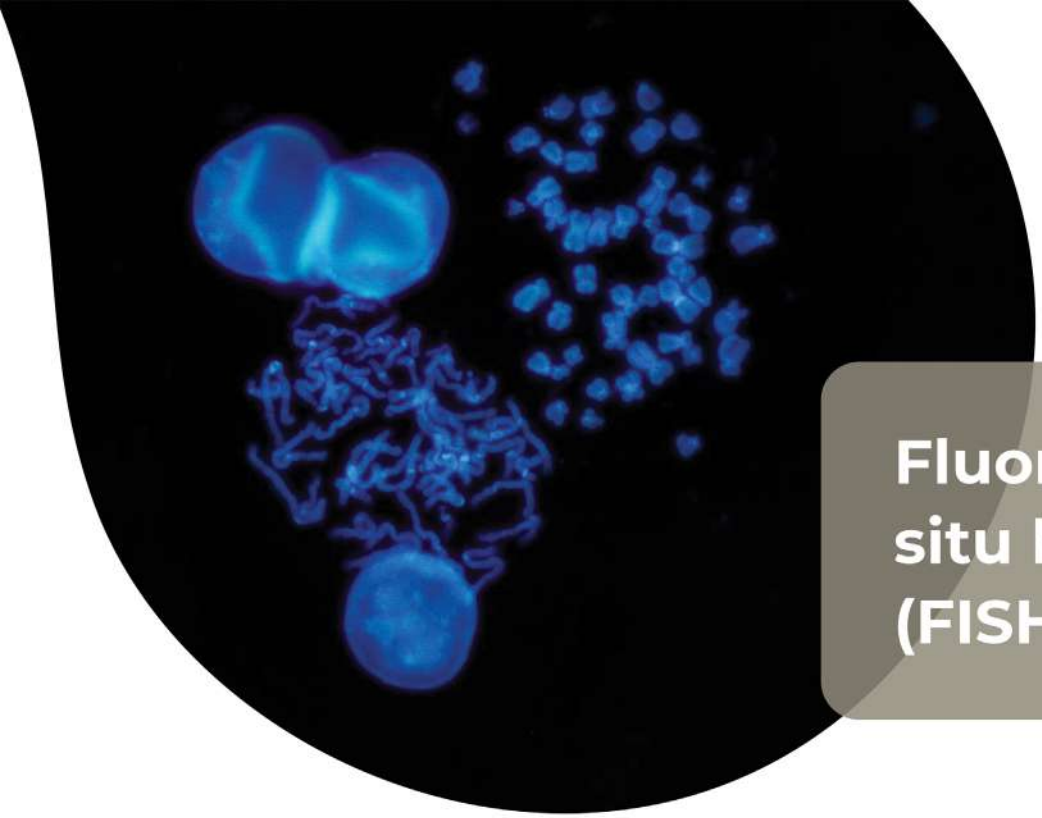

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Fluorescence in situ hybridization (FISH)

Fluorescence in situ hybridization (FISH)

Fluorescence in situ hybridization (FISH) is one of several techniques used to search your cells' DNA, looking for the presence or absence of specific genes or portions of genes.

FISH and other in situ hybridization procedures are used to diagnose a variety of chromosomal abnormalities—changes in the genetic material, changes in chromosomes, including the following: 3

Deletion: part of a chromosome is gone
Translocation: part of one chromosome breaks off and sticks onto another chromosome
Inversion: part of a chromosome breaks off and reinserts back in, but in reverse order
Duplication: part of a chromosome is present in too many copies within the cell

FISH is most commonly used in breast cancer, sarcoma, lymphoma, multiple myeloma, myelodysplastic syndrome (MDS) and some leukemias.
FISH testing is done on breast cancer tissue removed during biopsy to see if the cells have extra copies of the HER2 gene.

TAT: 2-4 days

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Chromosomal Microarray Analysis

Chromosomal Microarray Analysis

Cytogenic variations are known to cause a broad range of developmental disorders, primarily neurodevelopmental and congenital anomalies.

Chromosomal microarray analysis (CMA) is recommended for analyzing cytogenic variations in patients suffering for unexplained developmental delays, intellectual disabilities, autism spectrum disorders, and/or multiple congenital malformations.

Microarray-based solution enables the genome-wide detection of known novel structural aberrations, copy number variations (CNVs), chromosomal imbalances, regions exhibiting loss/absence of heterozygosity (LOH), uniparental isodisomy (UPD), and mosaicism.

MOH NO: 167/2023

What is CGH array Recommended?

As a first-step analysis for cases of unexplained developmental delays, intellectual disabilities, autism spectrum disorders, and/or multiple congenital malformations
For deletion/duplication analysis of extremely large genes where gross deletions involving large segments of genes, flanking intergenic regions, or neighboring genes are frequently reported

TAT: 21 days

The BION Advantage

- Processing time: **In a short time after sample receipt**
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OTHER TESTS

1. Karyotyping High resolution
2. FISH (Fluorescence in Situ Hybridization) according to your request
3. NIPT (Ch 13, Ch 18, Ch 21, X,and Y) with Illumina CE-IVD
4. Familial Mediterranean Fever (FMF) Gene, Sanger sequencing (Golden standard method)
5. Beta Thalassemia (Beta Globin) Gene, Sanger sequencing (Golden standard method)
6. Alpha Thalassemia (Alpha Globin) Gene, Sanger sequencing (Golden standard method)and MLPA
7. Hemochromatosis, HFE gene hotspot mutations (C282Y, H63D) - Sanger sequencing (Golden standard method)
8. CF, CFTR gene (Common mutations) - Sanger sequencing (Golden standard method)
9. Sickle Cell Mutation - Sanger sequencing (Golden standard method)
10. SMA 1 and 2, MLPA
11. DMD , MLPA
12. JAK2 (V617F/G1849T), Sanger sequencing (Golden standard method)
13. Deafness, GJB2 gene (ex1,2), Sanger sequencing (Golden standard method)
14. Glucose-6-phosphate dehydrogenase deficiency (FAVISM), G6pd gene,), Sanger sequencing (Golden standard method)
15. MTHFR , (Real time PCR)
16. PAI-1 , (Real time PCR)
17. FV , (Real time PCR)
18. FII , (Real time PCR)
19. Wolf-Hirschhorn syndrome (4p16.3)
20. Cri-du-Chat syndrome (5p15)
21. Sotos syndrome (5q35.3)
22. Williams-Beuren syndrome (7q11.23)
23. Williams-Beuren duplication syndrome (7q11.23)
24. DiGeorge syndrome-2 (10p13-p14)
25. Prader-Willi syndrome (15q11.2)
26. Angelman syndrome (15q11.2)
27. Rubinstein-Taybi syndrome (16p13.3)
28. Miller-Dieker syndrome (17p13.3)
29. Lissencephaly-1 (17p13.3)
30. NF1 microdeletion syndrome (17p11.2)\
31. DiGeorge syndrome (22q11.21)
32. 22q11.2 microduplication syndrome
33. Distal 22q11.2 deletion syndrome
34. Phelan-McDermid syndrome (22q13)
35. Rett syndrome (Xq28)
36. MECP2 duplication syndrome (Xq28)

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Gen QA Genomics Quality Assessment
Member of Gen QA