

D-dimer

Why Get Tested?

To help diagnose or exclude thrombotic (blood clot producing) or [bleeding diseases](#) and conditions.

When To Get Tested?

When you have symptoms of a disease or condition that causes [acute](#) and/or [chronic](#) inappropriate blood clot formation such as: DVT (Deep Vein Thrombosis), PE (Pulmonary Embolism), or [DIC \(Disseminated Intravascular Coagulation\)](#), and to monitor the progress and treatment of DIC and other thrombotic conditions.

Sample Required?

A blood sample taken from a vein in your arm.

Test Preparation Needed?

None

DHEAS

Why Get Tested?

DHEAS is measured to evaluate adrenal function and certain adrenal tumours, and to investigate the cause of [virilisation](#) (showing features of male hormones) or excess facial and body hair ([hirsutism](#)) in girls and women or early (precocious) puberty in boys.

When To Get Tested?

The measurement of DHEAS is used very infrequently in the UK as other tests can be more helpful. A doctor may measure DHEAS in those with signs or symptoms that may be due to the presence or too much male hormone.

It is sometimes used in the investigation of [Cushing's syndrome](#).

Sample Required?

A blood sample taken from a vein in your arm

Test Preparation Needed?

No test preparation is needed

Digoxin

Why Get Tested?

To determine if the amount of digoxin in your blood is at an appropriate level or to detect potentially toxic levels

When To Get Tested?

Soon after the start of digoxin therapy and at regular intervals to ensure that drug levels are within the desired range and are not low or at toxic concentrations

Sample Required?

A blood sample collected from a vein in your arm

Test Preparation Needed?

No special preparation is needed, but timing of the sample for testing is important. When you have your blood taken, tell the healthcare professional when you took your last dose of digoxin. You may want to write down the exact time at which you took your dose and when the blood was taken. This information will be useful if your doctor has any questions about your results.

Direct Antiglobulin Test

Why Get Tested?

To help diagnose the cause of [haemolytic anaemia](#) as caused by [autoimmune disease](#) or induced by drugs; to investigate a transfusion reaction; to diagnose [haemolytic disease of the foetus and neonate](#)

When To Get Tested?

When your doctor wants to find out the cause of your haemolytic anaemia; when you have had a blood transfusion recently and are experiencing symptoms of a transfusion reaction; or when a newborn shows signs of haemolytic disease of the foetus and neonate (HDFN).

Sample Required?

The test is performed on a sample of blood obtained from a vein in the arm using a needle. This is a process which may be referred to as 'venepuncture'.

Test Preparation Needed?

None

Direct LDL Cholesterol

Why Get Tested?

To help determine your risk of developing [heart disease](#) and to monitor lipid-lowering lifestyle changes and drug therapies; to accurately determine your [low-density lipoprotein cholesterol \(LDL-C\)](#) concentration when you have raised [triglyceride](#) concentrations

When To Get Tested?

As a follow-up to a [lipid profile](#) if your [triglycerides](#) are significantly elevated; at regular intervals to monitor efforts to lower LDL concentrations

Sample Required?

A blood sample taken from a vein in your arm

Test Preparation Needed?

No test preparation is needed. Your doctor might recommend that you fast (water only) for 14 hours prior to the test so that other related substances can be measured in the same sample.

Drugs of Abuse

Why Get Tested?

To detect or exclude the presence of commonly abused and/or illegal drugs. This may be carried out for a number of reasons including screening for pre-employment purposes or to comply with a drug rehabilitation programme.

When To Get Tested?

- If you apply for a job where drug screens are carried out as a routine. People with drug and alcohol problems have worse records for accidents and absenteeism; for this reason some employers screen job applicants prior to appointment.
- If you have admitted having a drug problem and are enrolled in a detoxification or drug rehabilitation scheme where testing is part of the programme.
- If you believe you may have taken a drug accidentally or been given a drug without consent (e.g. drink spiking).
- If you are admitted to hospital in an emergency and doctors think that your treatment could be improved if drug abuse could be proved or excluded.
- If you take part in a sport at a professional level.
- If you apply for an insurance policy – some companies perform limited drug screening on applicants.

- For legal reasons (e.g. child custody cases).

Sample Required?

A [random urine sample](#) is usually collected for detection of drugs of abuse although they can be detected in blood, sweat, saliva, breast milk and hair samples.

Test Preparation Needed?

Some prescription and over-the-counter drugs may give a positive screening result; before you are tested, indicate any medications that you have taken and/or for which you have prescriptions.

eGFR - estimated Creatinine Clearance

Why Get Tested?

In the UK, the National Institute for Health and Care Excellence (NICE) guidance (CG182) states: "Whenever a request for serum creatinine measurement is made, clinical laboratories should report an estimate of GFR (eGFR)"

When To Get Tested?

As a practical test to look for evidence of kidney dysfunction. To monitor changes in kidney function in long term/chronic medical conditions.

Sample Required?

eGFR is an estimate of actual glomerular filtration rate and may be calculated using your age, weight, height, gender, ethnicity, [serum creatinine](#) and/or serum [cystatin C](#) (requires a blood sample from a vein in your arm)

Test Preparation Needed?

NICE guidelines advise people not to eat any meat in the 12 hours before having a blood test for eGFR creatinine. Recent evidence also suggests that fish should not be eaten before having a blood test.

Electrolytes and Anion Gap

Why Get Tested?

To detect a problem with the body's electrolyte balance.

When To Get Tested?

As part of routine blood testing, or when your doctor suspects that you have an imbalance of one of the electrolytes (usually [sodium](#) or [potassium](#)), or if your doctor suspects an [acid-base imbalance](#). Electrolytes may also be checked if you are prescribed certain drugs, particularly [diuretics](#) or ACE inhibitors.

Sample Required?

A blood sample taken from a vein in the arm or in some cases, a urine sample

Test Preparation Needed?

Emergency and Overdose Drug Testing

Why Get Tested?

To detect, measure, and occasionally to monitor drugs that may have been taken in overdose or are causing acute overdose symptoms; results from emergency and overdose drug testing are used mainly to help treatment. If results are needed for legal proceedings, then special legal (forensic) procedures must be followed for sample collection, storage, and testing.

When To Get Tested?

If a drug overdose is suspected, or when a person has symptoms such confusion, difficulty breathing, feeling sick, agitation, fits, changes in heart rhythm, or increased temperature that the Accident and Emergency (A&E) doctor thinks may be drug-related; at intervals to monitor a drug overdose

Sample Required?

A blood sample taken from a vein in your arm, a urine sample, or sometimes a breath sample; rarely, saliva or another body fluid

Test Preparation Needed?

None

ENA Panel

Why Get Tested?

To help diagnose and distinguish between [autoimmune disorders](#) as well as to monitor autoimmune disease progression

When To Get Tested?

When your [antinuclear antibody \(ANA\) test](#) is positive; when you have symptoms that suggest an autoimmune disorder; when monitoring the activity of an autoimmune disorder

Sample Required?

A blood sample taken from a vein in your arm

Test Preparation Needed?

None

Epstein-Barr Virus Antibodies

Why Get Tested?

The usual reason to test for EBV is to help diagnose glandular fever (also known as Infectious Mononucleosis). Some people may also be tested for EBV if they are having treatment that weakens the immune system, such as immunosuppressive medication or a transplant.

When To Get Tested?

If you have symptoms of glandular fever (fevers, tiredness, swollen lymph nodes, sore throat)

Sample Required?

A blood sample taken from a vein in your arm

Test Preparation Needed?

No test preparation is needed

Erythrocyte Sedimentation Rate (ESR)

Why Get Tested?

To detect and monitor the activity of [inflammation](#) as an aid in the diagnosis of the underlying cause

When To Get Tested?

When your doctor thinks that you might have a condition that causes inflammation and to help diagnose and follow the course of this, especially [temporal arteritis](#) or [polymyalgia rheumatica](#)

Sample Required?

A blood sample taken from a vein in the arm

Test Preparation Needed?

None

Erythropoietin

Why Get Tested?

Either to help tell the difference between [polycythaemia vera](#) and secondary polycythaemia or to help tell the difference between different types of [anaemia](#). It also shows whether the amount of erythropoietin being produced is appropriate for the level of anaemia present

When To Get Tested?

If a patient has an elevated red blood cell count or an anaemia that the doctor suspects may be caused by decreased red blood cell production

Sample Required?

A blood sample taken from a vein in your arm

Test Preparation Needed?

None

Ethanol

Why Get Tested?

To find out if a person has drunk alcohol and to measure the amount of alcohol present in the body

When To Get Tested?

When a patient has symptoms that suggest drunkenness or alcohol toxicity, or when a person is suspected of breaking drinking-related laws or as part of a drug testing panel for pre-employment or other purposes.

Sample Required?

Ethanol (Ethyl alcohol) can be measured in a blood or urine sample, in breath or (rarely) in saliva. Blood, urine, and saliva samples must be sent to a laboratory for analysis. A breath sample is analysed immediately on site using a breath analyser ("breathalyser").

Test Preparation Needed?

None

Factor V Leiden Mutation and PT 20210 Mutation

Why Get Tested?

To determine whether you have an inherited gene mutation that increases your risk of developing a venous [thromboembolism](#) (blood clot)

When To Get Tested?

If you have a strong family history of thrombosis associated with these mutations; if you have unexplained thrombosis that requires further investigation

Sample Required?

A blood sample taken from a vein in your arm

Test Preparation Needed?

No test preparation is needed.

Faecal Immunochemical Test

Why Get Tested?

To screen for bleeding from the gut/intestine, which may be an indicator of [bowel cancer](#)

When To Get Tested?

The test is used to screen for bowel cancer especially as part of the [National Bowel Cancer Screening Programme](#). Outside of the Screening Programme the test is also widely available to GPs following the publication of the NICE Guidelines on the recognition of suspected cancers (NG12). Patients with symptoms that are

suspicious of gut cancer will usually be asked to complete a FIT test before being referred for other investigations such as colonoscopy.

Sample Required?

One sample of faeces collected into a special container.

You collect a sample yourself, in the privacy of your own bathroom, using a special kit. The actual collection method varies between manufacturers. Read all the instructions very carefully before starting to collect your sample.

Test Preparation Needed?

For the FIT test, there are no dietary, drug, or dental procedure restrictions. However, avoid collecting samples during a menstrual period. The test uses antibodies to detect only human blood from the lower digestive tract (colon). For all tests, follow the instructions that are provided by the doctor or included in the test kit.

Ferritin Test

Why Get Tested?

To help assess the levels of iron stored in your body

When To Get Tested?

When your doctor suspects that you have either too little or too much iron in your system

Sample Required?

A blood sample taken from a vein in your arm

Test Preparation Needed?

You may be instructed to fast for 12 hours before the test; in this case, only water is permitted.

Fibrinogen

Why Get Tested?

To determine whether your fibrinogen level is adequate to allow normal blood clotting, to help diagnose [disseminated intravascular coagulation \(DIC\)](#), to help determine whether you have an inherited fibrinogen deficiency or abnormality.

Sometimes as a non-specific marker of inflammation in the blood. Very occasionally to help evaluate your risk of developing [cardiovascular disease](#)

When To Get Tested?

When you have unexplained or prolonged bleeding, an abnormal [Prothrombin Time \(PT\)](#) or [activated Partial Thromboplastin Time \(aPTT\)](#) test, or have a relative with a hereditary fibrinogen deficiency or abnormality. To evaluate whether certain abnormalities in a [Full Blood Count](#) are due to inflammation or are self-generated. When your doctor wants additional information to help evaluate your risk of developing [heart disease](#)

Sample Required?

A blood sample taken from a vein in your arm, or sometimes, via a finger prick (mainly performed in children)

Test Preparation Needed?

None

First Trimester (Combined) Screen for Down's Syndrome and other fetal anomalies

Why Get Tested?

To assess the risk of a foetus having a chromosomal abnormality, such as [Down's syndrome](#) (trisomy 21).

When To Get Tested?

Usually between 11 and 14 weeks of [pregnancy](#)

Sample Required?

A blood sample taken from a vein in your arm. Measurement of nuchal translucency (NT) requires a special ultrasound examination to be performed.

Test Preparation Needed?

You may be requested to have a full bladder when having the nuchal translucency ultrasound performed. Having a full bladder when you attend for your scan usually makes it easier to see the baby clearly and measure the nuchal translucency.

Fructosamine

Why Get Tested?

To help monitor your [diabetes](#) over time, especially if it is not possible to monitor using the [HbA1c test](#); to help determine the effectiveness of changes to your diabetic treatment plan

When To Get Tested?

When you are diabetic and your doctor wants to evaluate your average blood glucose level over the last 2-3 weeks

Sample Required?

A blood sample taken from a vein in your arm

Test Preparation Needed?

No test preparation is needed.

FSH Test

Why Get Tested?

To evaluate the function of your [pituitary gland](#), which regulates the [hormones](#) that control your reproductive system

When To Get Tested?

If you are having difficulty getting pregnant or are having irregular menstrual periods (in order to identify or rule out ovarian failure/menopause in women and sperm production failure in men); if your doctor thinks that you have symptoms of a [pituitary](#), ovarian, testicular or [hypothalamic](#) disorder; when your doctor suspects that a child has delayed or earlier than expected sexual maturation (or delayed or early growth).

Sample Required?

A blood sample taken from a vein in your arm

Test Preparation Needed?

No special preparation is required for the test, but the blood sample should be taken at the start of a woman's cycle (days 1 to 4 if the cycles are regular)

FT3

Why Get Tested?

To help diagnose [hyperthyroidism](#) and to monitor it's treatment

When To Get Tested?

If you get an abnormal thyroid stimulating hormone ([TSH](#)) or free thyroxine ([FT4](#)) result

Sample Required?

A blood sample taken from a vein in the arm

Test Preparation Needed?

None needed; however, certain medications can interfere with the FT3 test, so tell your doctor about any drugs that you are taking

FT4

Why Get Tested?

To diagnose [hypothyroidism](#) or [hyperthyroidism](#) in adults and to monitor response to treatment

When To Get Tested?

When you have symptoms of an underactive (hypothyroidism) or overactive (hyperthyroidism) thyroid gland

Sample Required?

A blood sample taken from a vein in the arm

Test Preparation Needed?

No test preparation is necessary

Full Blood Count (FBC)

Why Get Tested?

Commonly requested as part of a general screen in a patient who is unwell to screen for a variety of disorders, such as [anaemia](#) and infection, inflammation nutritional status and bleeding

When To Get Tested?

As determined by your doctor; there are many illnesses which will affect the full blood count (FBC) and the result may help to make a diagnosis

Sample Required?

A blood sample taken from a vein in the arm or a finger-prick or heel-prick (newborns)

Test Preparation Needed?

None

Fungal Tests

Why Get Tested?

To detect a [fungal](#) infection, to determine which specific fungus or fungi are present, and sometimes to isolate and grow the fungi for subsequent [susceptibility testing](#)

When To Get Tested?

When the doctor suspects that you have a skin, lung, or [systemic](#) fungal infection; sometimes after treatment to monitor its effectiveness

Sample Required?

The sample collected depends upon the suspected location(s) of the infection. For superficial infections, the sample may include scrapings of the skin, clipped or shaved nail or hair, vaginal swabs, or a urine sample. For deeper tissue, organ or [systemic](#) infections, the sample may involve the collection of blood from a vein, [sputum](#) from the lungs, and/or the collection of a tissue [biopsy](#). If [meningitis](#) is suspected, a sample of [cerebrospinal](#) fluid may be collected.

G6PD

Why Get Tested?

To see whether you have an inherited deficiency of Glucose-6-Phosphate Dehydrogenase (G6PD), necessary to keep red blood cells healthy

When To Get Tested?

If a child has long term [jaundice](#) as a newborn and no other cause can be identified; when you have had one or more bouts of haemolysis (break-up of red blood cells, which may cause jaundice, dark urine or anaemia), particularly if the haemolysis follows "[oxidative](#)" stress caused by some medicines, foods, or infections

Sample Required?

A blood sample taken from a vein in your arm

Test Preparation Needed?

None required

Gamma-Glutamyl transferase (GGT) Test

Why Get Tested?

To screen for [liver disease](#) or alcohol abuse; and to help your doctor tell whether a raised concentration of [alkaline phosphatase \(ALP\)](#) in the bloodstream is due to liver or bone disease

When To Get Tested?

If your doctor thinks that you have symptoms of a liver disorder/disease

Sample Required?

A blood sample taken from a vein in your arm

Test Preparation Needed?

None required

Gastrin

Why Get Tested?

To detect an overproduction of gastrin, to help diagnose [Zollinger-Ellison syndrome](#) (ZE syndrome), to help diagnose multiple endocrine neoplasia type 1 (MEN-1) and to monitor for recurrence of a gastrin-producing tumour (gastrinoma)

When To Get Tested?

When you have [peptic ulcers](#) and/or diarrhoea and abdominal pain that your doctor suspects is caused by excess gastrin; periodically to monitor for a gastrinoma recurrence

Sample Required?

A blood sample taken from a vein in your arm.

Test Preparation Needed?

You should fast for 12 hours and avoid alcohol for 24 hours before the test. Your doctor may also ask you to stop taking certain stomach medications for several days before the test.

Genetic Tests for Targeted Cancer Therapy

What are genetic tests for targeted cancer therapy?

Genetic tests for targeted cancer therapy detect [mutations](#) (changes) in the [DNA](#) of cancer cells. Knowing whether the cancer has a particular mutation can help guide the type of treatment that a person receives. The presence or absence of certain mutations can predict who may benefit from certain drugs and who is not likely to respond.

Cancer is the uncontrolled growth of abnormal cells. Multiple factors may contribute to this uncontrolled growth. One such factor is the malfunctioning of [proteins](#) involved in controlling cell growth and maturation. The proteins usually malfunction as a result of a mutation in the DNA of the [gene](#) that codes for that protein. Some mutations may result in a defective protein that cannot stop cell growth while other mutations may produce a protein with altered function that stimulates cell growth. The net result is unchecked growth and division of these abnormal cells (cancer).

Medical researchers have long studied these changes in genes in order to better understand cancer and to develop drugs to fight it. Their goal has been to create drugs that disrupt a specific step in cancer growth, while doing minimal damage to normal cells. These are called targeted drugs or targeted therapy. What researchers

have noted is that specific types of cancer are frequently associated with specific genetic mutations. Not every cancer will have them, but a significant percentage will, and cancers with these mutations usually have a more predictable response to certain drug treatments compared to cancers without these mutations.

These findings have led to two important developments:

- Cancer drugs that inhibit or target very specific proteins associated with certain cancers (two examples are [tyrosine kinase inhibitors](#) and epidermal growth factor receptor (EGFR) antibodies.)
- Genetic tests to detect the presence of mutations in cancer tissue that tell a healthcare practitioner whether the person being tested is likely to benefit from a specific therapy

Medical researchers continue to explore the genetics of cancer and to look for opportunities to develop new therapies. Additionally, some cancers eventually stop responding to certain therapies and develop resistance to that therapy. Genetic research may offer insights into how resistance to therapy occurs.

Why is this testing important?

Standard treatment for cancer usually involves surgery, chemotherapy, radiation therapy, or some combination of these. Treatment with chemotherapy drugs and radiation aims to slow the growth of cancer, keep it from spreading, and kill any cancerous cells that have spread to other parts of the body (metastasized).

Chemotherapy works by attacking cells that are actively growing and dividing. Radiation therapy kills cancer cells by damaging their genes and preventing them from growing and dividing. Both types of therapy can affect all cells that are growing and dividing, including normal cells. This often leads to harmful side effects, and these treatments require careful adjustment to maximize the killing of cancer cells while minimizing the damage to healthy tissue.

Targeted therapy is a newer type of cancer treatment that offers healthcare practitioners and their patients the opportunity to use a drug that has a greater effect on cancerous tissue, reducing many of the side effects associated with standard therapy. It is based on the fact that the genetic makeup of the cancer cells is different than the normal cells around them. Targeted therapy aims to disrupt specific steps or processes that are somewhat unique to the growth of cancer cells. Testing the cancer cells taken from patients in a biopsy prior to initiating drug therapy may be necessary to determine the cancer's likely response to certain drugs.

Targeted cancer drugs are expensive, and they generally only work in patients whose cancer has the genetic makeup that they have been developed to work against. Genetic testing prior to beginning therapy is necessary to match the treatment up with the patients and cancers likely to benefit from them.

Examples of targeted cancer drugs for which tests are available include:

- Drugs that bind to receptors on the cell surface and block growth signalling to the cell
- Drugs that are small molecules that cross the cell membrane and block the signals for growth at the receptor's active site

Testing

How is genetic testing for targeted cancer therapy used?

These genetic tests are used to help guide treatment for certain cancers. They help to inform a healthcare practitioner as to whether certain targeted cancer drugs may or may not work.

Genes are the basic units of genetic material, the segments of **DNA** that usually code for the production of specific **proteins**. Alterations in DNA are called genetic variants (also **polymorphisms** or **mutations**) and occur throughout the population. Variants or mutations are largely inherited and affect all cells, but they can occur later in life, because of exposures to radiation, toxins, or for unknown reasons, and these mutations may result in cancer.

In a variety of cancers, there may be a mutation that leads to an increased amount of a particular protein present in the tumour tissue or to production of a protein that has altered activity. **Tumours** that have these mutations may tend to grow more aggressively, be more likely to spread (metastasize), and/or may be more resistant to standard chemotherapy. Sometimes, however, the changes in the protein also make the tumours candidates for therapy that targets the changed protein ("targeted therapy"). Genetic tests for cancer therapy detect the mutations that code for these proteins, thus identifying tumours that may be susceptible to targeted therapy.

Conversely, genetic tests may also identify tumours that will not respond to targeted therapy. Certain mutations, when present, make the cancer cells resistant to the drug and targeted therapy will not be used for treatment.

When are the tests requested?

Testing may be ordered as part of an initial workup of particular cancers or performed on those with certain cancers that are not responding to chemotherapy. It requires a sample of the tumour tissue, and if a sample is available from a previous **biopsy** used for diagnosis, it can be done on that sample.

The tests are usually performed only once. However, testing may be done more than once if a patient's tumour progresses while on therapy to see if the tumour has acquired mutations that are resistant to the therapy.

Tests

Each genetic test for a specific targeted cancer therapy identifies mutations in a single gene, and test results are specific to the gene and the targeted therapies * Gene names are typically abbreviated for ease of use because full names are often several words long.

Type of Cancer	Gene Tested*	Interpretation of Test Result
Breast cancer	Her2/neu	When present, likely response to trastuzumab
Chronic myelogenous leukaemia (CML)	ABL1	Nonresponsive to imatinib when mutation not present
	BCR-ABL	When present, can be measured periodically to monitor response to targeted drug
Bowel cancer	KRAS	When mutation present, likely resistance to anti-EGFR targeted drug
	BRAF	Poor prognosis when mutation present
Gastrointestinal stromal tumour (GIST)—rare tumours of the digestive tract	KIT	Depending on mutation present, better response to imatinib; increased dose of imatinib likely necessary; sunitinib, or possible resistance to imatinib
	PDGFRA	When mutation present, less likely to respond to imatinib
Melanoma	BRAF	Better response to vemurafenib when mutation present in melanoma
Myeloproliferative neoplasms (MPNs)	JAK2	When mutation present, may be measured to monitor responsiveness to treatment (e.g., Ruxolitinib)
Non-small cell lung cancer (NSCLC)	EGFR	Best response to tyrosine kinase inhibitors in those with certain mutations
	EML4-ALK	If ALK is present, may respond to ALK kinase inhibitors (e.g., crizotinib)
	ROS1	If ROS1 is present, ALK kinase inhibitors may be effective
	KRAS	Poorer prognosis when certain mutations present; no response to tyrosine kinase inhibitors, and poor response to platinum-based chemotherapy
	PDL1	Likely response to immunotherapy
Cancers of unknown origin—cancers detected in unusual body sites and thought to have spread (metastasized) from another location	Several genes evaluated together (genomic array or profile)	Helps determine the organ or body part of origin in order to help guide treatment

Usually, the cancer drugs and genetic tests listed in the table above have been developed concurrently and the tests are referred to as companion diagnostics. These are laboratory tests that are developed specifically to provide information that is essential for the safe and effective use of a corresponding therapeutic product. In many cases, results from these tests are needed for healthcare practitioners to be able to make decisions regarding treatment of their patients.

Cancers associated with a strong family history and those that occur at a young age may have different characteristics than those that develop sporadically in adults. For instance, paediatric cases of GIST are very different to adult cases and do not typically have KIT or PDGFRA mutations.

Only common mutations are tested. A negative test result does not rule out the possibility that a person has a less common mutation. To rule out the possibility that the mutation was not present in the sample tested, additional samples may be needed.

Some tests for specific gene mutations in certain types of cancer are available on a limited basis and/or not used routinely for medical purposes. These genetic tests may, however, be used in research settings and their utility in medical care may evolve as research progresses. Some examples include:

- Colon cancer: PIK3CA and NRAS
- Melanoma: KIT and NRAS
- Myeloproliferative neoplasms: PDGFRA

Glucose Tests

Why Get Tested?

To determine whether or not your blood glucose level is within normal ranges; to screen for, diagnose, and monitor [diabetes](#), and to monitor for the presence of [hypoglycaemia](#) (low blood glucose) and [hyperglycaemia](#) (high blood glucose)

When To Get Tested?

If you have symptoms suggesting [hypoglycaemia](#) or [hyperglycaemia](#), or if you are [pregnant](#). If you have [diabetes](#), you may be required to monitor glucose levels several times a day using a self-monitoring device.

Sample Required?

A blood sample taken from a vein in your arm or, for self-monitoring, a drop of blood from your finger. A few diabetic patients may use a continuous glucose monitor which is a small sensor wire inserted beneath the skin of the abdomen that measures blood glucose every five minutes.

Test Preparation Needed?

For screening purposes, fasting is generally recommended (nothing to eat or drink except water) for at least 8 hours (generally 8-10 hours fasting) before a blood glucose test. Those who have been diagnosed with diabetes and are monitoring their glucose levels are often tested both while fasting and after meals. For random and timed tests, follow the instructions given to you by your healthcare professional.

There is another test called an oral glucose tolerance test ([OGTT](#)). It requires that the person fasts (as described above) for the first blood sample and then drink a liquid containing a specified amount of glucose; a further blood sample is then taken after 2 hours. This test is commonly offered to pregnant patients for diagnosis of diabetes in [pregnancy](#) (gestational diabetes).

Gonorrhoea Test

Why Get Tested?

To screen for *Neisseria gonorrhoeae*, which causes the [sexually transmitted disease](#) (STD) gonorrhoea

When To Get Tested?

If you have symptoms of gonorrhoea or are [pregnant](#)

Sample Required?

A swab of secretion or discharge from the infected area.

Testing is available at genito-urinary medicine (GUM) clinics , sexual health centres, contraceptive centres and some GP surgeries.

Test Preparation Needed?

Please tell your doctor or healthcare professional about the use of antibiotics or, for women, douches or vaginal creams within 24 hours before testing vaginal samples, as they may affect test results. For a urine sample, you may be instructed to wait one to two hours after you last urinated before collecting a urine sample. Follow any instructions you are given.

Gram Stain

Why Get Tested?

To identify the cause of a [bacterial](#) infection so appropriate treatment can be given

When To Get Tested?

Seek advice from your doctor if you have an area of inflamed, red or a painful skin, a wound which does not seem to be healing or any other concerns which might lead you to believe you may have a bacterial infection

Sample Required?

A skin swab or fluid/pus (if present) from the site of infection

Test Preparation Needed?

None

Growth Hormone

Why Get Tested?

Growth hormone (GH) is produced by the pituitary, situated at the base of the brain, behind the bridge of your nose and has growth promoting properties. It is measured to check if there is under or overproduction. In addition it is used to examine the function of the pituitary and to monitor the effectiveness of treatment.

When To Get Tested?

The evaluation of GH status is based on clinical findings, medical history, imaging and biochemical tests. Slow growth in height and delayed development (in children), whilst decreased bone density and/or muscle strength, and increased lipids (in adults) could all be related to insufficient GH production. Symptoms suggestive of [gigantism](#) in children or [acromegaly](#) in adults may be a result of excess GH production. It is also measured as part of an evaluation of pituitary function.

Sample Required?

After an overnight fast, several blood samples are taken at timed intervals from veins in your arm, as part of a stimulation or suppression test. Pre-adolescents require priming prior to performing a stimulation test. A sample is usually taken for measurement of insulin-like growth factor-1 ([IGF-1](#)) on the baseline sample. When monitoring treatment for GH excess a single sample of blood may be drawn following a fast.

Test Preparation Needed?

In healthy adults GH is released in bursts throughout the day, it rises sharply 3-4 hours after a meal and within 60 minutes after the onset of sleep making random GH results in general uninterpretable. GH may be measured after stimulation or suppression testing. Fasting levels are used to monitor treatment for GH excess.

What is being tested?

GH is needed for a child's normal growth and development. It promotes growth of the long bones from birth through puberty. Children with insufficient GH production grow more slowly and are small in size for their age; one of the first symptoms of growth hormone deficiency (GHD). It should be noted that short stature can also be related to familial traits or other genetic disorders. Constitutional delay (i.e. temporary delay in growth of no obvious cause) is the most common cause of short stature in childhood.

An excess of GH is most often due to a [benign](#) GH-secreting pituitary tumour i.e. a tumour that has not spread to other tissues, although larger tumours can have other effects e.g. headaches and impaired vision Gigantism is a disorder resulting from long-term secretion of too much GH, which increases the growth of muscle, bones

and connective tissue in childhood or adolescence before the end of puberty. This results in a child becoming excessively tall (e.g. over 2.1 meters). Children with excessive GH production may also have thickening of their facial features, general weakness, delayed puberty, and headaches.

In adults GH plays a role in regulating bone density, muscle mass, and lipid metabolism. Deficiencies can lead to decreased bone densities, lower muscle mass, and altered lipid concentrations. Excess GH in adults can lead to [acromegaly](#), with bone and skin thickening. Symptoms such as sweating, fatigue, headaches and joint pain can be subtle at first. Increased GH concentrations can lead to enlargement of the hands, feet, facial bones and internal organs and carpal tunnel syndrome (trapped nerves). If untreated, acromegaly in adults and [gigantism](#) in children can lead to complications such as type 2 diabetes, increased cardiovascular disease risk, high blood pressure, arthritis, and in general, a decreased life span.

GH stimulates the secretion of the true growth factors, most notably IGF-1. The concentration of which represents the secretion of GH in the previous few days. This is measured prior to stimulation or suppression tests used to diagnose GH abnormalities and to monitor treatment.

How is the sample collected for testing?

A GH suppression or stimulation tests is performed after fasting for 10 to 12 hours when a blood sample is taken from a vein in the arm. Under medical supervision, a standard glucose solution is given to the patient to drink (for a suppression test), or an intravenous (IV) injection of a solution of insulin, glucagon, arginine, clonidine or GH releasing hormone (GHRH, for a stimulation test) is given through a vein in your arm. Blood samples are then taken from your veins at timed intervals. GH is measured on each sample collected to look at the change in levels over time. Sometimes it is necessary to perform a second test.

During treatment for growth hormone excess a sample of blood may be taken, following a fast, to monitor growth hormone production.

Is any test preparation needed to ensure the quality of the sample?

Samples collected for stimulation or suppression tests are collected after fasting. Pre-adolescents are usually primed with sex steroids prior to a stimulation test.

Haemoglobin

Why Get Tested?

If you have [anaemia](#) (too few red blood cells) or polycythaemia (too many red blood cells), to assess its severity, and to monitor response to treatment

When To Get Tested?

As part of a [full blood count \(FBC\)](#), which may be requested for a variety of reasons

Sample Required?

A blood sample collected from a vein in your arm or by a finger-prick (children and adults) or heel-prick (newborns)

Test Preparation Needed?

Ideally you should be reasonably hydrated when having a haemoglobin test or the result may be inaccurately high.

Haemoglobinopathy Evaluation

Why Get Tested?

To investigate [haemoglobinopathy](#) as the cause of signs and symptoms; to screen for a haemoglobin disorder

When To Get Tested?

As follow up to abnormal results on a [full blood count](#) (FBC) and/or [blood film](#); when you have symptoms of [haemolytic anaemia](#) such as weakness and fatigue and your doctor suspects that you have an abnormal form of haemoglobin (haemoglobinopathy); when you have a family history of haemoglobinopathy; as part of [newborn screening](#)

Sample Required?

A blood sample taken from a vein in your arm

Test Preparation Needed?

None

Haptoglobin

Why Get Tested?

To help detect and evaluate [haemolytic anaemia](#)

When To Get Tested?

When you have signs of [jaundice](#), or of anaemia such as weakness, paleness, or breathlessness that the doctor suspects may be due to red blood cell destruction (haemolytic anaemia).

Sample Required?

A blood sample taken from a vein in your arm.

Test Preparation Needed?

No test preparation is needed.

HbA1c Test

Why Get Tested?

To monitor average blood glucose levels in someone with [diabetes](#) and to help treatment decisions. It can also be used to make a diagnosis of type 2 diabetes and to identify prediabetes.

When To Get Tested?

When first diagnosed with diabetes and then at least twice a year

Sample Required?

A blood sample taken from a vein in the arm

HDL Cholesterol Test

Why Get Tested?

To screen for risk of developing [cardiovascular disease](#) (heart disease, stroke and related diseases); to monitor treatment

When To Get Tested?

Aged 40 as part of a routine cardiovascular health check, or if you are already thought to be at risk of cardiovascular disease for another reason (including already having suffered from cardiovascular disease).

Sample Required?

Testing for HDL cholesterol requires a blood sample. Most often, the blood sample is collected by venepuncture (using a needle to collect blood from a vein in the arm). Occasionally a fingerprick test can be used, although this is not commonly available in GP practices or hospitals in the UK.

Test Preparation Needed?

No fasting is needed for an HDL-cholesterol test, or the full lipid profile. On the other hand, there may be circumstances when fasting is still required, so you should follow the instructions given by your health care team.

Heavy Metals

Why Get Tested?

To screen for, detect, and monitor excessive exposure to specific heavy metals

When To Get Tested?

Periodically when you work with heavy metals, or when your doctor suspects that you may have been exposed. If you have a metal-on-metal hip prosthesis, depending upon the size and type of implant, you may need annual blood monitoring of metal ions (e.g. chromium and cobalt). Consult your doctor for further advice.

Sample Required?

A blood sample taken from a vein in your arm or a [24-hour urine sample](#); rarely, a hair sample, tissue sample, or other body fluid sample

Helicobacter Pylori Test

Why Get Tested?

To diagnose an infection with *Helicobacter pylori* that can cause peptic ulcers

When To Get Tested?

If you have gastrointestinal pain or symptoms of an [ulcer](#)

Sample Required?

A stool sample, blood sample from a vein, [tissue biopsy](#) of the stomach lining, or urea breath test

Test Preparation Needed?

No special preparation is needed for the blood test. For the urea breath test and if submitting a stool or having a biopsy you may be instructed to refrain from certain medications. If undergoing endoscopy, you may be instructed to fast after midnight the night prior to the procedure. Please follow any instructions given to you by a healthcare professional.

Heparin Anti-Xa

Why Get Tested?

To monitor low molecular weight heparin (LMWH) therapy and sometimes to monitor unfractionated heparin (UFH) therapy

When To Get Tested?

When you are being treated with LMWH or UFH and your doctor wants to monitor the amount of heparin in your blood

Sample Required?

A blood sample taken from a vein in your arm

Test Preparation Needed?

No test preparation is needed, although the timing of the test is important

What is being tested?

This test measures the effect of low molecular weight heparin (LMWH) or unfractionated heparin (UFH) in the blood by measuring anti-Xa activity. Heparin is an [anticoagulant](#), a drug that inhibits blood clotting. Heparin molecules vary in size and activity. UFH includes a broad range of sizes, while LMWH consists of a narrower range of smaller heparin molecules. There are several types of LMWH available and each one is slightly different. UFH is usually given [intravenously \(I.V.\)](#) and LMWH is usually given by a [subcutaneous](#) injection to people who have inappropriate blood clots (thrombi) and/or are at an increased risk of developing them.

Blood clotting is a normal response to blood vessel or tissue injury. It is a process that involves a sequential activation of proteins that regulate blood clot development. A variety of [acute](#) and [chronic](#) risk factors, including surgery, pregnancy and some oral contraceptives, serious illnesses and immobility are associated with inappropriate blood clot (thrombus) formation in veins – especially in the legs. These clots can obstruct blood flow and cause pain and swelling in the affected area. Pieces of the blood clot can break off and travel to the lungs - causing pulmonary embolism.. Heparin can also inhibit blood clot formation in diseased arteries, which sometimes cause heart attacks or strokes.

Heparin, through its action on a protein called Antithrombin, interferes with the clotting process by accelerating the inhibition of coagulation factors, particularly factors Xa and IIa (thrombin). UFH, which affects both Xa and IIa, is more variable in its inhibitory activity, and must be closely monitored using a variety of tests including the APTT, ACT or less commonly by anti-Xa. Complications may include bleeding, and sometimes a serious complication called “HIT” (Heparin Induced

Thrombocytopenia) causing a low platelet count and thrombosis at the same time. UFH is usually given in a hospital setting. High doses of UFH are given during surgery requiring cardiopulmonary bypass.

LMWH has more anti-Xa action than anti-IIa activity and the response to it is more predictable. It may be given in either an outpatient or hospital setting. Routine monitoring of LMWH is seldom required but when it is monitored, the anti-Xa test is used.

How is the sample collected for testing?

A blood sample is obtained by inserting a needle into a vein in the arm, most commonly taken three to four hours after your heparin injection to check the 'peak' level.

Is any test preparation needed to ensure the quality of the sample?

No test preparation is needed other than correct timing.

Heparin-induced Thrombocytopenia Antibody

Why Get Tested?

To detect antibodies against the anticoagulant heparin, to help diagnose immune-mediated heparin-induced thrombocytopenia (HIT II). There is also a non-immune mediated HIT (type I) that occurs when heparin binds directly to platelets, causing activation; it is more common than type II but is transient and a milder form.

When To Get Tested?

When you are receiving heparin therapy and your [platelet count](#) significantly decreases ([thrombocytopenia](#)), especially when you also have new blood clots ([thrombosis](#))

Sample Required?

A blood sample taken from a vein in your arm

What is being tested?

This test detects and measures [antibodies](#) that are produced by some people when they are treated with heparin. Heparin is a common [anticoagulant](#) that is given [intravenously](#) or through subcutaneous injections to prevent the formation of blood clots (thrombosis) or as an initial treatment for those who have a blood clot, to prevent the clot from enlarging. It is often given during some operations, such as cardiopulmonary bypass, when the risk for developing blood clots is high. Small

amounts of heparin are frequently used to flush out [catheters](#) and intravenous lines to keep clots from forming in them.

When a person is given heparin, the drug can combine with a substance found in platelets called platelet factor 4 (PF4) and form a complex. In some people, the body's [immune system](#) recognises the heparin-PF4 complex as "foreign" and produces an antibody directed against it. This antibody can activate platelets and lead to a drop in the number of platelets, a condition known as heparin-induced thrombocytopenia (HIT). It may also lead to the development of new thrombosis or worsening thrombosis.

Platelets are cells that are an important part of the blood clotting system. When a blood vessel is injured and leaks blood, platelets are activated and clump together at the site of the injury, and work with [coagulation factors](#) to promote clot formation and stop the bleeding.

Not everyone on heparin produces HIT antibodies, and not everyone with HIT antibodies develops a low platelet count, but about 1% to 5% of those with the antibodies do. In HIT, the antibodies bind to the heparin-PF4 complexes, which then attach to the surface of platelets. This activates the platelets, which in turn, triggers the release of more PF4. This starts a cycle that can cause a rapid and significant drop (e.g., 50% or more) in the number of platelets in the blood. Usually, a decrease in platelets results in a higher risk of bleeding, but in HIT, the activation of platelets by HIT antibodies can paradoxically lead to new and progressive blood clot formation in the veins and arteries. This occurs in about 30% to 50% of those who have the HIT antibody and thrombocytopenia.

This condition, associated with the presence of HIT antibody, low platelet count, and excessive clotting, is formally called immune-mediated heparin-induced thrombocytopenia or HIT type II. It typically develops about 5-10 days after a person starts heparin therapy but may also develop rapidly, within 1-2 days, if a person has been treated with heparin in the last 3 months and starts treatment again.

How is the sample collected for testing?

A blood sample is obtained by inserting a needle into a vein in the arm.

Is any test preparation needed to ensure the quality of the sample?

No test preparation is needed.

Hepatitis A Virus Antibodies

Why Get Tested?

To diagnose an infection with hepatitis A [virus](#), or to find out the need for or the response to hepatitis A vaccination

When To Get Tested?

If you have symptoms of an infection with or have been exposed to the hepatitis A virus; to detect previous infection or vaccination

Sample Required?

A blood sample taken from a vein in your arm

Hepatitis B Virus Antibodies

Why Get Tested?

To detect, diagnose and follow the course of an infection with hepatitis B virus (HBV) or to determine if the [vaccine](#) against hepatitis B has produced the desired level of [immunity](#)

When To Get Tested?

If you have symptoms of a hepatitis B infection or are likely to have been exposed to the [virus](#); if you have chronic liver disease (possibly due to some other cause), if you have received the vaccine, if you were born to a mother who was HBV positive or if you are being treated for HBV

Sample Required?

A blood sample taken from a vein in your arm

Hepatitis C Virus Antibodies

Why Get Tested?

To screen for and diagnose a [hepatitis C virus](#) infection and to monitor treatment of the infection

When To Get Tested?

If you may have been exposed to/have risk factors for the hepatitis C virus, such as through contact with infected blood, sexual relations with an infected person, IV drug use or you have symptoms associated with [liver disease](#)

Sample Required?

A blood sample taken from a vein in your arm

What is being tested?

Hepatitis C is a [virus](#) that can infect and damage the liver. In most cases, it is contracted through exposure to blood (usually from sharing contaminated needles while injecting drugs or, before 1992, through a blood transfusion), through sex with an infected person, via healthcare occupational exposure and it can also be passed from mother to baby.

Many people who are infected with Hepatitis C are not aware they are as acute infection produces few to mild non-specific symptoms. However Hepatitis C can also exist as a chronic (longstanding) infection and you can show no signs of this for a number of years (even decades) but it can then cause significant liver damage. About 65-75% of those infected can develop [chronic liver disease](#) with 20-30% of these developing [cirrhosis](#) over many years.

Hepatitis C antibody is produced by the body in response to exposure to the hepatitis C virus (HCV). The most common test for HCV looks for these [antibodies](#) in your blood. Some first line tests are also looking for the hepatitis C antigen, which the virus itself produces, as well as your antibody response. Other tests detect the presence of and actual amount of virus present or determine the specific subtype of virus.

HER-2 testing

Why Get Tested?

To determine whether a cancer, usually a [breast cancer](#), is positive for HER-2 gene amplification or protein expression, which helps to guide treatment and determine outcome. HER-2 evaluation is also used in the assessment of some other cancer types including gastric and oesophageal cancer.

When To Get Tested?

If you have been diagnosed with a cancer which may be responsive to treatment with HER-2 targetted therapy and your doctor wants to determine whether the HER-2 [gene](#) is amplified in the tumour.

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. While HER-2 levels can be assessed in a blood sample, this is not usually done as treatment decisions rely on testing of the cancer cells in biopsy material.

Herpes Testing

Why Get Tested?

To screen for or diagnose [infection](#) with herpes simplex virus

When To Get Tested?

If you have symptoms of an infection with herpes simplex virus

Sample Required?

Your doctor will take a swab or scraping from a blister or sore in the mouth or genital area.

Histamine

Why get tested?

To help confirm a diagnosis of [anaphylaxis](#) or symptomatic [mastocytosis](#). However, due to the need for blood samples to be processed extremely quickly after the blood is taken, this test is rarely performed.

When To Get Tested?

When you have symptoms such as flushing, nausea, throat swelling or low blood pressure that may be due to a life-threatening allergic reaction; sometimes when your healthcare practitioner suspects that you have mastocytosis.

Sample required?

A blood sample drawn from a vein in your arm or a [24-hour urine](#) collection. The blood sample must be spun and frozen within 5 minutes of collection.

Test preparation needed?

None for anaphylaxis, but timing of the sample very soon after the beginning of symptoms is important. If testing is done for other conditions, you may be instructed to refrain from taking [antihistamine](#) and other medications. This should be discussed with your healthcare professional.

HIV 1 Viral Load

Why Get Tested?

To monitor the status of [HIV 1](#) disease in conjunction with other lab tests and physical disease progression and to guide therapy. Your viral load levels are usually used as an indicator of how well your immune system is dealing with HIV. If you are on anti-HIV treatments, it can be an indicator of how well the treatments are working.

When To Get Tested?

When first diagnosed with HIV 1, frequently at the start of therapy, and every 3-12 months during lifelong therapy thereafter, or as your doctor recommends. Patients with viral loads consistently <50 copies/mL are likely to be tested less often than patients whose virus level is less well controlled.

Rarely your doctor may also ask for the test;

- If you have had an indeterminate HIV 1/2 antibody/antigen result
- You are at risk and have symptoms of acute HIV, irrespective of the HIV 1/2 antibody/antigen result

Sample Required?

A blood sample taken from a vein in your arm

Test Preparation Needed?

No test preparation is needed.

HIV Antibody and HIV Antigen (p24)

Why Get Tested?

To determine if you are infected with [human immunodeficiency virus \(HIV\)](#)

When To Get Tested?

When you think you may have been exposed to the [virus](#); once a year if you are at risk of being exposed to the virus; when your doctor thinks your symptoms may be due to HIV; before becoming pregnant or during [pregnancy](#)

Sample Required?

A blood sample collected from a vein in your arm or from a fingerprick; some tests can also be performed on urine or saliva (spit)

What is being tested?

[Human immunodeficiency virus \(HIV\)](#) infects the cells of a person's immune system and is the cause of AIDS (acquired immunodeficiency syndrome).

When a person becomes infected with HIV, through exposure to the blood or body fluids of an infected individual, the virus begins to reproduce very rapidly. So, during the first few weeks of infection, the amount of virus (viral load) in the blood can be quite high.

The [immune system](#) responds by producing antibodies directed against the virus and these begin to be detected in the blood around 3-4 weeks after exposure to the virus. As the level of HIV antibody increases, the viral load in the blood decreases.

This early HIV infection may cause no symptoms or sometimes a flu-like or glandular fever-type illness. The only way to determine whether a person has been infected is through HIV testing. Modern HIV screening tests detect HIV [antigens](#) (parts of the virus itself, usually a protein called the p24 antigen) and/or [antibodies](#) produced in response to an HIV infection.

Two main test types are available for HIV screening:

- Combination HIV antibody and HIV antigen test— this is the recommended screening test for HIV and is available only as a blood test. By detecting both antibody and antigen, the combination test increases the likelihood that an infection is detected soon after exposure. These tests can detect HIV infections in most people by 2-6 weeks after exposure.
- HIV antibody testing— This test takes a little longer to become positive after an exposure but can be carried out on blood or oral fluid. HIV antibody tests can detect infections in most people 3-12 weeks after exposure.

HIV Resistance Testing

Why Get Tested?

If you have been diagnosed with [HIV](#), this test can be used to help doctors select the right drugs for treating the infection.

When To Get Tested?

Before starting HIV treatment (anti-retroviral therapy), or if your [HIV viral load](#) increases (or does not decrease) even though you are receiving anti-retroviral therapy

Sample Required? A blood sample taken from a vein in your arm

HLA Testing

Why Get Tested?

To identify which human leukocyte antigen (HLA) [genes](#) and [antigens](#) a person has inherited. Either to;

- 1) Match donors and recipients of organ and bone marrow transplants and to detect [antibodies](#) to HLA antigens that would cause transplants to be unsuccessful
- 2) Confirm the diagnosis of a disorder which only affects individuals of a certain HLA type
- 3) Predict the likelihood of an adverse drug reaction associated with a specific HLA type

When To Get Tested?

Most often, transplant recipients are tested when it is determined that they need an organ or bone marrow transplant, prior to seeking and selecting a suitable donor; potential donors are tested when they are being evaluated for compatibility with a specific recipient or are signing up with a national donor registry.

If someone is suspected of having a clinical disorder associated strongly with a given HLA type, testing may be undertaken as part of the diagnostic workup.

If a clinician plans to start a drug which has serious side effects, associated with a specific HLA type, testing would be done before the medicine is prescribed or rarely as part of investigating an unusual reaction.

Sample Required?

A blood sample drawn from a vein in your arm; sometimes, for HLA typing, a swab from the inside of the cheek (buccal swab) or a saliva sample.

Test Preparation Needed?

When providing a saliva sample you should not eat, drink, smoke or chew gum for 30 minutes prior to giving a sample.

HLA-B27

Why Get Tested?

To find out whether you have [human leucocyte antigen B27](#) (HLA-B27) on the surface of your cells; to help assess whether you might have an [autoimmune disorder](#) associated with the presence of HLA-B27

When To Get Tested?

When you have symptoms of chronic inflammation, pain, and stiffness in certain areas of your body, such as your back, neck, and chest, or the interior portion of your eyes [uveitis](#), especially if you are male and the symptoms began between late teens and your early 30s

Sample Required?

A blood sample taken from a vein in your arm

Test Preparation Needed?

None