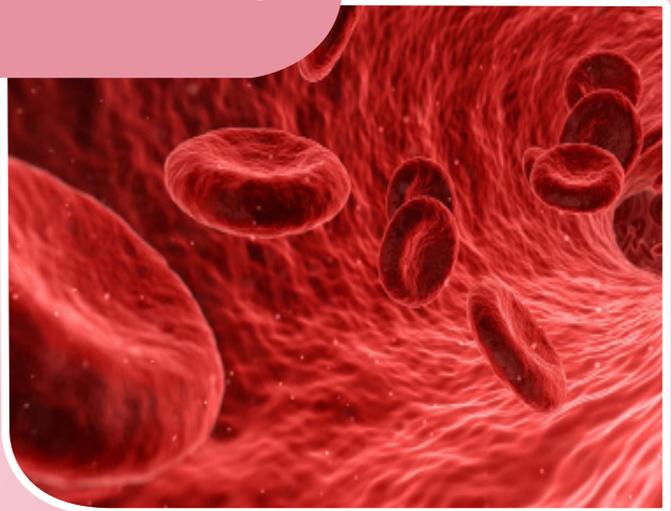


what do the tests mean?

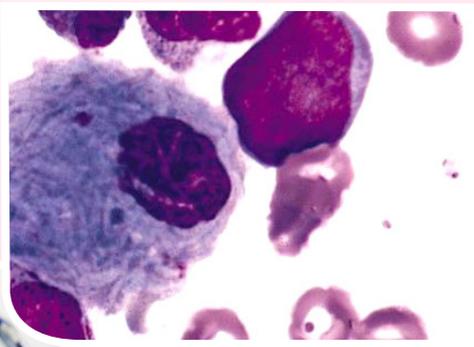
Your specialist team will arrange regular tests/investigations to assess your Gaucher disease and to check that the treatment is working effectively for you. The more your clinical team understand how Gaucher affects you the more they will be able to prevent/treat problems that may occur.



these tests commonly include:

diagnosis

| Test name | What is being assessed? | Why? | How? |
|--------------------|------------------------------------|---|--|
| White cell enzymes | Glucocerebrosidase (GBA) activity | Low levels indicate Gaucher disease | Blood test |
| Genetics (DNA) | GBA mutation analysis | It is thought that different alterations in the GBA gene may lead to different symptoms (<i>known as genotype:phenotype correlation</i>) | Blood test |
| Bone Marrow Biopsy | Blood cells within the bone marrow | This test is sometimes done before Gaucher is suspected, usually to look for blood cancers such as leukaemia. In Gaucher a characteristic cell can be seen. | A small sample of bone marrow is removed from your hip or spine through a fine needle after local anaesthetic. This takes about 30 minutes as an outpatient. |



blood tests



| Test | | Why? |
|---------------------------------|---|---|
| Full blood count (FBC) | Haemoglobin (Hb) | Low levels of Hb (anaemia) can cause fatigue |
| | Platelet count (plts) | Low levels of platelets can increase the risk of bruising/bleeding |
| | White cell count (wcc) | High levels of white cells might indicate a current infection Low levels may indicate a difficulty in fighting infection |
| Biochemistry | Liver profile (LFT) | This is a group of tests looking at how well the liver is working |
| | Renal profile (U&E) | This is a group of tests looking at how well the kidneys are working |
| | Bone profile | This is a group of tests looking at how well the bones are working |
| Immunoglobulins (IgA, IgM, IgG) | These are specific proteins (antibodies) created as part of the immune system | This is a group of tests looking at how well the immune system is working. Useful for early identification of myeloma |
| Biomarkers | Chitotriosidase (Chito) PARC/CCL18 | These are proteins found in the blood stream that are specifically increased in Gaucher disease. With treatment these levels decrease and are therefore good indicators of Gaucher activity |
| Anti-drug antibodies | Antibodies | This is a group of tests looking at how well the bones are working |

| Test | Usual Site | Why? | Special Notes | What will happen? |
|-------|---|--|---|--|
| X-ray | Chest Hips Sites of bone pain | X-rays are mainly used to look at bones and joints- to identify the cause of pain | Please tell the radiographer if there is any possibility you may be pregnant | You lie on a table or stand against a flat surface. The X-ray machine will be aimed at the part of the body being examined. The radiographer will operate the machine from behind a screen. You will not feel anything and it should last just a few minutes |
| MRI | Liver/Spleen Bones/Joints (BMB*) *BMB - a specific score assessing Gaucher involvement in the bones | The liver and spleen are assessed for size and shape/ texture Bones and joints are assessed for areas that might cause pain/ need joint replacement | MRI uses strong magnetic fields to create a detailed internal image. It may not be suitable for people with metal implants (including pacemakers) | You lie on a table that is moved into a round tube containing the magnet. The radiographer will operate the machine from another room and will talk to you through the intercom. The scanner will make loud intermittent tapping noises and you may be given earplugs to wear. You should not feel anything although it may be a little claustrophobic. It will last 30 - 60 mins depending upon the area being examined |
| DEXA | Hip, spine, wrist | DEXA assesses your bone density (or strength) of your bones | Please tell the radiographer if there is any possibility you may be pregnant | You lie on a table on your back. A large scanning arm will pass over you, You should not feel anything and it will last just a few minutes |

Other tests

At each visit to your specialist centre the clinical team will ask you lots of questions to see how you are. They may ask you to complete questionnaires about your health, undertake a physical examination (to feel your liver/spleen or assess how well your nerves are working for example) or look at your eye movements. You may be invited to participate in relevant research to help improve understanding of the disease and to create new improved treatments for the future.



This list is not exclusive and you may have other tests depending upon the symptoms you have. If you do not understand the tests your doctor has suggested for you do ask them to explain it to you.