

List of Gaucher Tests

Blood Tests	Purpose	Frequency of Tests
Enzyme level Of β -glucocerebrosidase	Everyone with Gaucher will have a reduced level of this enzyme compared to unaffected individuals. Low levels are diagnostic	Only at diagnosis
DNA test (genotyping)	To identify the specific mutation for Gauchers disease	Only at diagnosis
Full blood count including haemoglobin → platelets → white blood cells → Iron studies→ Serum Ferritin → Immunoglobulins →	<ul style="list-style-type: none"> - if low can result in anaemia and fatigue - reduction in the level of platelets can lead to bruising and bleeding - if lower than normal can increase susceptibility to infection - to exclude other causes of anaemia - often elevated in Gaucher and reduces in response to therapy - Gaucher can affect the immune system causing elevation of these proteins which again often reduce in response to treatment 	At first visit and at each follow up visit to assess response to therapy or monitoring
Biochemistry, liver function tests, vitamin D, calcium levels, serum ACE, CRP, bilirubin & acid phosphatase	The liver often becomes enlarged in Gaucher due to storage material accumulating in certain liver cells. Liver enzymes are often abnormal and should improve in response to treatment. If vitamin d or calcium levels are low then supplements may be advised to maintain good bone strength	At the baseline visit and at each follow up

<p>Lipid profile : cholesterol, (high and low density) and triglycerides</p>	<p>In Gaucher the cholesterol level in untreated patients is often below normal and may increase when treatment is initiated</p>	<p>At the first visit and each follow up</p>
<p>Clotting screen and clotting factors</p>	<p>Quite often the clotting time in affected individuals is prolonged causing a tendency to bleed for longer than normal, for instance following phlebotomy or minor trauma. Gaucher occurs more frequently among the Ashkenazi Jewish population, factor X1 deficiency is also more common among the Ashkenazim and this will be screened for</p>	<p>At first visit and all subsequent follow up visits</p>
<p>Chitotriosidase</p>	<p>A biomarker which is useful in the diagnosis and monitoring of Gaucher disease. It is greatly elevated in Gaucher and is a good indicator of severity. The levels decline in response to treatment, it is therefore a very useful marker of disease activity and can be a useful guide to the dose and frequency of treatment required.</p>	<p>At first visit and at every follow up assessment</p>
<p>Chitotriosidase genotyping</p>	<p>A significant percentage of the normal population have a mutation of the gene encoded to produce this enzyme, if this is the case in a person with Gaucher it may not be possible to use these levels to monitor treatment. There are new biomarkers now available which may eventually replace this test</p>	<p>At first visit and every follow up</p>

	IMAGING	
MRI Scan	<p>MRI scans are a very useful tool in staging the disease. The images provide very clear pictures of the bones, bone marrow, skeletal abnormalities and the volume of the liver and spleen. Volume of liver and spleen can be calculated at baseline and compared with scans at follow up at 12 – 18 months to measure treatment effect</p>	At first visit and at 12- 18 months follow up
DEXA SCAN	<p>Measure of bone mineral density (BMD), this scan will measure how strong the bones are. It provides a score comparing Gaucher with BMD of the normal population according to age and gender. If the bone density score is poor (osteopenia) there may be an increased risk of bone fracture. The addition of a bisphosphonate therapy in addition to ERT or SRT may improve the BMD. This treatment can be taken orally or intravenously</p>	Baseline and every 12 – 18 months
ULTRASOUND	<p>Gallstones are often associated with Gaucher and can be identified by ultrasound scan, plain X-Ray or MRI</p>	At baseline or if symptoms of abdominal pain occur
LUNG FUNCTION TESTS	<p>The lungs can be affected by the glycolipid storage although this is a less common complication of Gaucher and is more likely to occur in patients who have had their spleen removed</p>	At baseline and follow up if breathing difficulties occur

EYE MOVEMENT TESTS	A highly specialized test to exclude neurological involvement, may be indicated with certain genotypes or those with neurological symptoms to identify the more severe form of Gaucher which does affect the central nervous system	Only available in special centres
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