

**Gaucher disease (GD) is a rare genetic disorder (1 in 75,000 births World-wide), an inborn error of metabolism due to a deficiency of the lysosomal enzyme acid  $\beta$ -glucosidase (glucocerebrosidase). Gaucher's Disease is a Prescribed Minimum Benefit condition under Diagnostic Treatment Pair (DTP) code 901K.**

## Gaucher disease

This enzyme deficiency results in accumulation of glycosphingolipid-laden macrophages (Gaucher cells) throughout the liver, spleen, bone marrow, skeleton and occasionally the lung. There are 3 types of GD. In types 2 and 3 pathology also occurs within the brain.

- *Type 1 GD* - constitutes 94% of all cases and is usually considered to be non-neuronopathic. The onset of the disease may occur at any age in type 1 GD.
- *Type 2 GD* - the onset is in infancy (so-called neuronopathic infantile, cerebral, or perinatal lethal GD) and, accounts for 1% of all GD. It is characterised by a short life expectancy of 2-3 years or less due to severe neurological consequences related to the disease.
- *Type 3 GD* - presents in early childhood and accounts for approximately 5% of all patients. The range of neurological involvement in this group is quite broad.

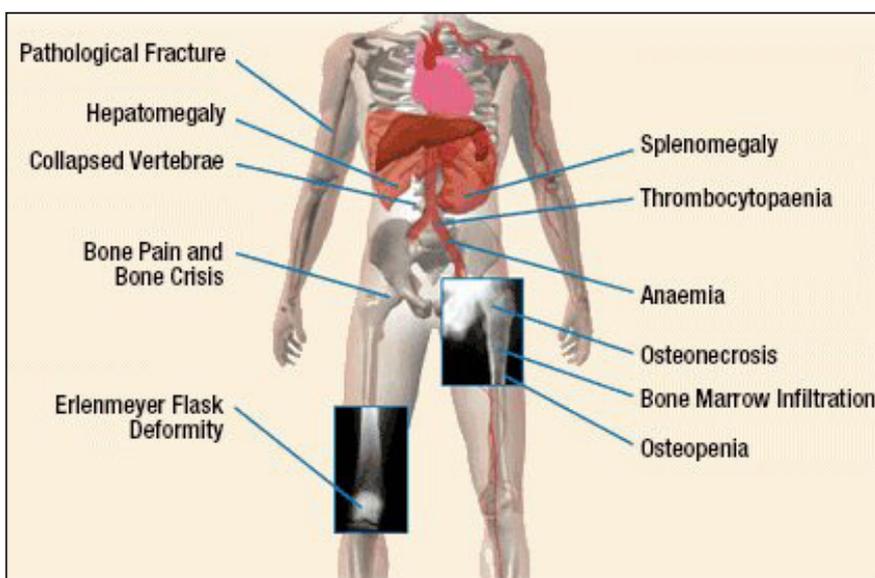
Gaucher disease has been demonstrated to occur in all ethnic groups in South Africa. Some population groups such as the Ashkenazi Jews have a higher incidence.

## Signs and symptoms

The signs and symptoms of Gaucher disease are a result of the progressive accumulation of Gaucher cells in the body. It includes an enlarged liver and spleen, anaemia, easy bruising and bleeding caused by low blood platelets (thrombocytopenia), diseases of the lungs, excessive fatigue, bone abnormalities such as bone pain, fractures, and arthritis. The diagram above depicts signs and symptoms of Gaucher disease.

## Diagnosis

The diagnosis of Gaucher disease is based on history, clinical evaluation, laboratory investigations and diagnostic imaging. The diagnosis is preferably confirmed by enzyme analysis together with DNA gene mutational analysis.



<http://www.health-pictures.com/images2/gaucher's%20disease.jpg>

## Diagnostic and follow-up imaging

- Magnetic resonance imaging (MRI) may be done to assess structural bone abnormalities.
- Dual energy X-ray absorptiometry (DEXA) is required at baseline and as needed for identification of patients at risk of fractures, and prior to and at follow-up of patients requiring antiresorptive or anabolic bone therapy.
- Plain radiology is done when clinically indicated for acute bone crisis or diagnosis of a fracture and chest X-ray (CXR) for suspected pulmonary (lung) involvement.
- Ultrasound is necessary for organ measurement (liver and spleen size when volumetric MRI is not available), gall stones, portal hypertension (high blood pressure) or chronic liver disease and renal (kidney) involvement. Heel ultrasound if indicated is needed to assess bone involvement if evaluation at other sites are not possible by DEXA scans.

## Treatment

- Enzyme replacement therapy (ERT) is the treatment of choice for types 1 and 3 Gaucher disease.
- Supportive therapy is indicated for those patients who decline ERT, usually elderly patients and require symptomatic supportive intervention with blood products, bisphosphonate therapy, and/or analgesia.
- Mobility aids such as crutches and wheelchairs to aid

- mobility for everyday living.
- Monitoring is important for patients identified with Gaucher disease mutations, who may be asymptomatic and do not require treatment at present, but must be monitored regularly (6-monthly) for disease progression according to the goals of treatment and indications to start therapy.
- Bone marrow transplantation may still be considered under certain circumstances for type 3 patients when a matched, unaffected sibling donor has been identified.
- Other therapies such as substrate reduction therapy is currently used in some patients with mild disease.
- Genetic counselling is an important component of supportive care for any family and best provided by a healthcare professional well versed in these aspects of care. Parents of affected individuals, individuals themselves when they reach an age of understanding, siblings of carriers or affected individuals, and spouses/potential spouses should be included. Genetic counselling aims to enable the patients and their families to understand the medical facts, role of inheritance, strategies to prevent recurrence and to make the best possible adaptation to the disorder.

### Other treatment considerations

Vitamin D, calcium; specific pain medication; seizure/neurological management; and pulmonary hypertension (high blood pressure) management.

### Surgical treatment

Orthopaedic surgical intervention is commonly required to restore function and correct deformity such as in cases of subchondral bone collapse due to avascular necrosis where joint replacement may be required.

Gallstone disease is also common in Gaucher disease.

### Prescribed Minimum Benefits

Gaucher's Disease is a Prescribed Minimum Benefit condition under Diagnostic Treatment Pair (DTP) code 901K. The DTP refers to life-threatening congenital abnormalities of carbohydrate, lipid, protein and amino acid metabolism. The treatment component of this DTP is specified as Medical management.

The interpretation of the PMB's should follow the predominant public hospital practice.

In terms of PMB surgical treatment, the appropriate DTP code will apply. For example, avascular necrosis will qualify for PMB benefits if there is a hip fracture under DTP code 178H. The treatment component specified for this DTP is reduction; hip replacement.

In relation to gallstones, calculus of bile duct with cholecystitis is a PMB diagnosis under DTP code 910G. The treatment component specified for this DTP is Medical management; cholecystectomy; other open or closed surgery.

### PMB treatment and care cover includes

- Pathology, radiology and other investigative and monitoring services
- Acute and chronic medication
- Prosthesis, appliances, devices – subject to managed care protocols
- Allied and supplementary health services such as physiotherapy, occupational therapy and speech therapy

Due to the rare nature of the condition combined with the scarcity of the specialist expertise required to manage this disease, it comes as no surprise that the expertise often reside in State Central Hospitals, depending on the region of the country. For this reason the Council for Medical Schemes has accepted arrangements where patients are managed in State Central Hospitals as well as private service providers. These arrangements are subject to terms negotiated on a case-by-case basis and are acceptable on merit. The member, therefore, might end up with a combination of private and public healthcare services where necessary.

**Enzyme Replacement Therapy:** Whilst specialist care maybe provided in the State, the scheme is responsible for funding ERT. It should be noted that Central hospitals may not have sufficient ERT for insured and non-insured patients. Therefore, schemes are to fund ERT dispensed at private pharmacies.

Out-hospital auxiliary services, PMB cover does not restrict a setting. As the state has overburdened auxiliary services, CMS does not encourage medical schemes to refer members for auxiliary services to the State. The medical scheme should fund treatment according to the PMB regulation. This service is not unlimited and is subject to continuous improvement with skills transfer to the caregiver.

*The Physiotherapist, Occupational Therapist, Speech Therapist should ensure transfer of skills to the caregiver. The providers should ensure that the scheme is provided with progress reports to enable the scheme to apply managed care principles in allocating benefits for the requested services. The providers should note that even if a condition is included in the PMB regulations, unlimited sessions cannot be approved.*

### References

- Bhengu, L., Davidson, A., du Toit, P., Gerntholtz, T., Govendrageloo, K., Heitner, R., Henderson, B., Mubaiwa, L and Varughese, S. 2012. South African guidelines for the management of Gaucher disease, 2011. South African Medical Journal 102(8):697-702.
- Rosenbloom, BE and Weinreb, NJ. 2013. Gaucher Disease: A comprehensive review. Critical Reviews in Oncogenesis, 18(3), 163-175.

Gaucher Disease | Signs And Symptoms of Gaucher Disease . [ONLINE] Available at: <http://www.gauchercare.com/en/patient/about/signsandsymptoms.aspx>. [Accessed 5 May 2015]

## PMBs

Prescribed minimum benefits (PMBs) are defined by law. They are the minimum level of diagnosis, treatment, and care that your medical scheme must cover – and it must pay for your PMB condition/s from its risk pool and in full. There are medical interventions available over and above those prescribed for PMB conditions but your scheme may choose not to pay for them. A designated service provider (DSP) is a healthcare provider (e.g. doctor, pharmacist, hospital) that is your medical scheme's first choice when you need treatment or care for a PMB condition. You can use a non-DSP voluntarily or involuntarily but be aware that when you choose to use a non-DSP, you may have to pay a portion of the bill as a co-payment. PMBs include 270 serious health conditions, any emergency condition, and 25 chronic diseases; they can be found on our website by accessing the link provided ([www.medicalschemes.com/medical\\_schemes\\_pmb/index.htm](http://www.medicalschemes.com/medical_schemes_pmb/index.htm)).

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