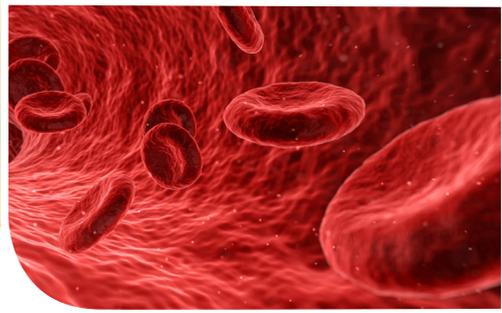


Gaucher disease & other conditions



Gauchers
ASSOCIATION



The most frequent problems encountered in Gaucher disease are low blood counts, enlarged abdominal organs and manifestations in the bone including pain, structural changes and fractures however there has been recent advances in describing and understanding some other associations with Gaucher, namely myeloma and Parkinson's disease.

These are conditions which occur within the non-Gaucher population as well but in which there seems to be a specific association with both Gaucher and one of the conditions occurring at the same time.

This understandably raises concerns amongst patients that not only do they have a rare inherited disorder but also have an increased chance of a blood cancer or neurological condition. Whilst it is true that in both cases the risk is elevated compared to the non-Gaucher population, the absolute risk for an individual patient remains low.

The value of the research has been highlighting these associations so that physicians can look out for early signs and manage them in Gaucher disease but also learn something about the more common forms of the conditions which might also help treatment.

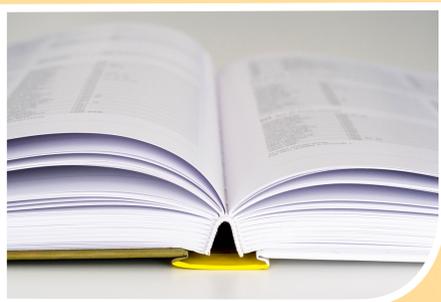
Myeloma is a type of blood cancer in which cells which normally make antibodies that fight infection multiply in of their own accord the bone marrow producing too much of irrelevant antibody proteins.

Myeloma often starts as an insidious low level condition with small amounts of the abnormal antibody protein (paraprotein) and minimal myeloma plasma cells in the bone marrow. At this stage there is no damage to organs and the condition is known as MGUS. It can continue like this for many years and there is a small chance of progressing for an individual patient each year.

This condition is called monoclonal gammopathy of undetermined significance (MGUS) and there has been found to be elevated incidence in Gaucher disease. MGUS does not require specific management but physicians should check the blood test for the antibody protein on a one to two yearly basis.

On occasion, MGUS will transform to myeloma. This condition is more serious and the paraprotein levels are generally higher. The abnormal cells and protein can cause damage to organs including the kidney and bones. There can be a high level of calcium with anaemia and higher risk of infection.

In Gaucher disease it can be difficult to work out whether the anaemia and bone disease are due to the Gaucher disease or the myeloma itself. Studies using the International Gaucher Disease Registry and also co-operative studies between European Groups have demonstrated that there is an increased risk of myeloma somewhere between 6 and 50 times the non-Gaucher population.



Treatment of myeloma has advanced remarkably over the last 10 years and include the combination of chemotherapy drugs and a newer agents which target specific parts of the cellular pathway. Much of this treatment is given as an out-patient and is not associated with the sort of side effects commonly attributed to chemotherapy.

Depending on the age of the patient treatment may include a high dose procedure where they also receive stem cells back from themselves as rescue-known as an autologous transplant. This is usually performed in hospital but now even this, in some hospitals, can be given in 'ambulatory care' where the patient stays in a hotel rather than in a hospital.

It is important to diagnose myeloma in the early phase of its development so the correct treatment can be given promptly with better outcomes. The most recent interesting finding in the area of myeloma and Gaucher disease has been an understanding that the antibodies in myeloma in Gaucher patients seem to be directed against fatty substances that accumulate as part of Gaucher disease and in mouse models of Gaucher disease treatment to prevent the build up of the fatty substrates prevents the development of myeloma and similar conditions.

Parkinson's disease is a neurological disorder in which patients have problems with movement including a tremor, stiffness and difficulty walking.

Other problems may be encountered with sense of smell for example and mood including depression. Gaucher disease has usually been classified with three types with Type 1 the most commonly occurring type not exhibiting neurological problems whereas Type 2 and 3 having neurological problems such as eye movement difficulties and epilepsy at some stage. However, it has been found that there is an increased incidence of Parkinson's disease in people who carry one of the normal Gaucher genes (ie a Gaucher carrier) and some patients with Gaucher disease also can develop Parkinson's disease.

A large multi-centre analysis looking at the risk of Parkinson's disease in Gaucher carriers suggested that the risk was 4-5 times more than the non-carrier population. Various mechanisms have been proposed for the development of Parkinson's disease including the effect of normal lipid accumulation on an association with a protein called synuclein which is involved in Parkinson's disease. These experiments do implicate the normal enzyme itself in parts of the brain which do not function properly in patients with Parkinson's disease. Current studies are investigating whether it is possible to use novel oral treatments to enhance the Gaucher enzyme and potentially help their Parkinson's disease.

Currently, clinical trials are monitoring both patients and their carrier relatives to investigate the emergence of subtle problems which may relate to Parkinson's disease.

If you have any questions or concerns relating to myeloma or Parkinson's or would like to participate in any of the studies then please contact your local clinician who can put you in touch with their relevant teams.

