Our Heritage and Our Health:

Genetic Conditions Frequently Affecting The Jewish Community

DID YOU KNOW?

Gaucher disease type 1 is the most common genetic disorder among the Ashkenazi Jewish Community?

Carrier Frequency	Incidence	Severity	Onset
~ 1:15	~ 1: 850	Extremely variable, from asymptomatic or mild symptoms to severe debilitation	From birth to the 7th- 8th decade of life

Signs & Symptoms:

Anemia and fatigue:
Reduction of circulating red blood cells

Easy bruising and bleeding:
Reduction in platelets makes it harder for blood to clot

Infections:
Decrease in the number of available white blood cells may lead to more frequent illnesses

Bone pain:
Loss of blood supply and oxygen is known to cause pain and may lead to bone death (osteonecrosis)

Enlarged liver and spleen
May result in decreased appetite and/or expanding of the abdomen

Osteopenia and osteoporosis:
Loss of calcium and mineral content weaken the bones

Other possible bone involvement:
Spontaneous fractures, joint pain, arthritis, and joint damage

Testing for Gaucher Disease

Enzyme test for glucocerebrosidase activity is the gold standard for a definitive diagnosis of Gaucher disease

Testing Details

Test:

- Enzyme analysis for glucocerebrosidase can be performed by various labs

- DNA testing can identify disease-causing variants. There is a residual risk of disease even with a negative DNA test result

Results:

Result time may vary from lab to lab

Specimen requirements vary from lab to lab. May include whole blood, dried blood spot, saliva, and buccal swab.

The ability to share knowledge about genetic diseases is a way to impart information that may help empower future generations.

- Gaucher disease type 1 can be effectively managed.

- Treatment options for Gaucher disease type 1 are available, including oral therapies.

- Be sure to discuss testing options, disease management, and treatment options with your doctor.

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For more information about Gaucher Disease Type 1, contact a CareConnect Personalized Support Services™ Patient Education Liaison at 1-800-745-4447, option 3 or by visiting https://www.careconnectpss.com