The carrier of this card has Barth syndrome, a life threatening genetic disorder. The cardinal characteristics include:

- <u>Neutropenia</u>: weakness in the immune system due to a reduction of neutrophils, necessary in fighting bacterial infections.
- <u>Cardiomyopathy & Arrhythmia</u>: Heart muscle weakness and abnormal heartbeats sometimes causing death
- <u>Muscle weakness and general fatigue</u>: All muscles, including the heart, have a cellular deficiency which limits their ability to produce energy, causing extreme fatigue during activities requiring strength or stamina, from walking to writing to growing.

Barth syndrome is an X-linked recessive genetic condition that is transferred from mother to son. A mother who is a carrier of Barth syndrome shows no signs or symptoms of the disorder.

Barth syndrome is a metabolic disorder. The enzyme that converts cardiolipin into its mature form does not work properly; therefore the level of cardiolipin is reduced. Cardiolipin is found in regions of cells called mitochondria. Mitochondria are essential to the proper functioning of



muscle cells including those of heart and skeletal muscle.

Barth Syndrome Foundation of Canada

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