A Mitochondrial-Specific ATP-Binding Transporter Gene (ABC7) Is An Iron Transporter In An Inherited Ataxia-Anemia Syndrome

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Technology description

Summary

The gene responsible for the rare genetic disease, X-linked siderblastic anemia and ataxia (XLSA/A) has been identified and linked to a mutation of the ATP-binding transporter gene (ABC7). Two sequence changes which correspond to amino acid changes at positions 50 and 396 were detected. This gene may prove useful as a diagnostic for XLSA/A carriers or as a means to rule out XLSA/A from other siderblastic anemias. ABC7, an iron transporter, may prove to be a valuable tool for studying the function and regulation of muscle cells and the loss of motor function associated with many diseases with faulty iron metabolism, i.e. neuromuscular disease, cardiac disorders and neurological disorders.

Institution

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