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Neuroferritinopathy

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Jonathan B Weitzman
Email: jonathanweitzman@hotmail.com

In the Advance Online issue of Nature Genetics, Andrew Curtis and colleagues from the Institute of Human Genetics in Newcastle, UK, describe a new genetic disease that they have named 'neuroferritinopathy'. The neurological disease is characterized by adult-onset degeneration of the basal ganglia and extrapyramidal dysfunction. Affected individuals live within a 40km radius of the home of the earliest founder, a member of a local family from Cumbria, UK. Curtis *et al.* performed linkage analysis to map the disease gene to chromosome 19q13.3. The disease locus contains the gene encoding ferritin light polypeptide (FTL). In six local patients they found a mutation causing an adenine insertion at position 460, which changes the carboxy-terminal residues of the protein. They found that patients had low serum ferritin levels and ferritin-positive inclusions in the basal ganglia and the brain. Mutations in the iron-responsive element of the *FTL* gene have been described in hereditary hyperferritemia cataract syndrome and iron metabolism has been linked to neurodegenerative disease in mice.

References

1. *Nature Genetics* , [http://genetics.nature.com]

2. Institute of Human Genetics , [http://www.ncl.ac.uk/sbg/humgen.html]

3. Hyperferritemia cataract syndrome, [http://www.ncbi.nlm.nih.gov:80/entrez/dispomim.cgi?id=600886]

4. Targeted deletion of the gene encoding iron regulatory protein-2 causes misregulation of iron metabolism and neurodegenerative disease in mice.