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DIAGNOSES AND REFERENCES**

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Case 2007-7

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Diagnosis: Autosomal recessive spastic ataxia of Charlevoix-Saguenay (ARSACS)

Comment: The MRI exhibited atrophy of the superior vermis, similar to alcoholic cerebellar degeneration. The molecular layer of the cerebellum was thinned, and there was depletion of retinal ganglion cells, with atrophy of the optic nerve. The corticospinal tracts and the dorsal columns in the cord were depleted also. Genetic testing of this patient revealed a defect of chromosome 13q11, where a 40 kD protein known as sarsin is coded, diagnostic of autosomal recessive spastic ataxia of Charlevoix-Saguenay (ARSACS). Sarsin is involved in protein folding. Charlevoix and Saguenay are two towns in Québec, Canada, where the gene frequency for this mutation is 1 in 22. Both of this patient's grandmothers were from Québec. The pathology of this disorder has some similarities with that of pellagra. Dr. Robitaille commented that there can be axotomy in the central white matter, producing a type of chromatolysis of neurons.

References:

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