

Title: Ataxia with Vitamin E Deficiency *GeneReview* Histopathologic Findings

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Note: The following information is provided by the authors listed above and has not been reviewed by *GeneReviews* staff.

**Histopathologic findings.** [Larnaout et al 1997, Yokota et al 2000, El Euch-Fayache et al 2014, Ulatowski et al 2014]

- Spinal sensory demyelination with neuronal atrophy and axonal spheroids
- Dying back-type degeneration of the posterior columns
- Neuronal lipofuscin accumulation in the third cortical layer of the cerebral cortex, thalamus, lateral geniculate body, spinal horns, and posterior root ganglia
- Fiber type grouping of the peroneus brevis muscle
- Mild loss of Purkinje cells

## References

El Euch-Fayache G, Bouhlal Y, Amouri R, Feki M, Hentati F. Molecular, clinical and peripheral neuropathy study of Tunisian patient with ataxia with vitamin E deficiency. *Brain*. 2014;137:402-10.

Larnaout A, Belal S, Zouari M, Fki M, Ben Hamida C, Goebel HH, Ben Hamida M, Hentati F. Friedreich's ataxia with isolated vitamin E deficiency: a neuropathological study of a Tunisian patient. *Acta Neuropathol (Berl)*. 1997;93:633-7.

Ulatowski L, Parker R, Warriar G, Sultana R, Butterfield DA, Manor D. Vitamin E is essential for Purkinje neuron integrity. *Neuroscience*. 2014;260:120-9.

Yokota T, Uchihara T, Kumagai J, Shiojiri T, Pang JJ, Arita M, Arai H, Hayashi M, Kiyosawa M, Okeda R, Mizusawa H. Postmortem study of ataxia with retinitis pigmentosa by mutation of the alpha-tocopherol transfer protein gene. *J Neurol Neurosurg Psychiatry*. 2000;68:521-5.