Title: Hypokalemic Periodic Paralysis *GeneReview* – Muscle Histology
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## HOKPP – Muscle Histology

The hypokalemic periodic paralysis myopathy is a vacuolar myopathy or a tubular agregate myopathy. In vacuolar myopathy related to hypokalemic periodic paralysis, vacuoles represent the predominant or the sole pathologic feature. A varying proportion of fibers display vacuoles that range from less than 10 µm in diameter to a width that nearly encompass the entire fiber. They are round or dome-shaped membrane limited spaces. They are membrane-limited. They are usually centrally placed and solitary, but sometimes several vacuoles in various situations can be seen in one fiber. Most vacuoles are empty but some contain varying amounts of granular material. In individuals with the myopathic form of HOKPP, muscle biopsy with adequate histochemical and histoenzymologic staining has been the mainstay of diagnosis. Light microscopy shows vacuoles [Pearson 1964, Olivarius & Christensen 1965, Martin et al 1984] or sometimes tubular aggregates [Faugere et al 1981]. Although the latter are less specific to HOKPP, they are the only lesions in some cases.

- ***CACNA1S*.** Vacuoles (or less frequently, nonspecific myopathic changes), but not tubular aggregates, were the histologic lesions seen in individuals with the c.1583G>A or c.3716G>A mutation.
- ***SCN4A*.** Tubular aggregates appear to be the primary histologic lesion in individuals with the c.2014C>G mutation [Sternberg et al 2001]; however, vacuoles may be the main lesion in those with different *SCN4A* mutations [Miller et al 2004].

## References

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