Charcot-Marie-Tooth type 1B (CMT1B) is an autosomal dominant demyelinating neuropathy with a gene locus on chromosome 1. Recent studies revealed that mutations in the gene of P₀, the major structural protein of peripheral myelin, are responsible for CMT1B. Also, some patients with a more severe phenotype, formerly designated Dejerine-Sottas syndrome, showed (de novo) P₀ mutations.

Preliminary studies suggested that the neuropathic deficit and nerve conduction slowing was greater in CMT1B than in CMT1A patients. Only one study has reported pathological features in proven CMT1B (Thomas et al. Acta Neuropath 1994;87:91).

We present the morphological changes in sural nerve biopsies of seven patients of different families in which recent investigations showed P₀ mutations. Light and electron microscopic studies revealed two types of pathology. Next to a chronic process of demyelination and remyelination with onion bulbs in all, three patients showed abnormal myelin compaction of some myelin sheaths. Morphology in three patients was dominated by the abundant occurrence of focally folded myelin as was reported before by Thomas et al. (1994) in a CMT1B family. Relation with the genetic defect is discussed.