Reply to: Myopathic (not neuropathic) electrophysiologic abnormalities in dynamin 2-related centronuclear myopathy

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We are very grateful for the valuable comments on the electrophysiological data on DNM2-CNM. The author is absolutely right that fibrillations due to muscle fibre necrosis and reduced compound motor action potentials (muscular atrophy) can also be observed in some myopathies, most commonly in muscular dystrophies or inflammatory myopathies.

However, (segmental) necrosis and regeneration—classical findings in muscular dystrophies—are usually not observed in muscle biopsies of patients with CNM (Jeannet, 2004) or other congenital myopathies (CM). CM are histopathologically defined by characteristic structural abnormalities, such as an increased number of central nuclei in DNM2-CNM or core lesions in central or minicore disease, e.g. (Fardeau, 1994), but not by dystrophic changes. Consistently, these myopathies generally show no or only rarely pathological spontaneous activity on needle electron examination (Dumitru, 1995). Therefore, the muscle fibre necrosis, as claimed by the author, cannot be responsible for the observed fibrillations in DNM2-CNM. Thus, while the reduced CMAPs are consistent with features of both a myopathy and a motor axonopathy, the fibrillations and above all the reduced motor and sensory nerve conduction velocities (NCV) as well as the diminished sensory nerve action potentials (SNAP) in the lower leg nerves as seen in some DNM2-CNM patients (Fischer, 2006) argue against a ‘pure’ myopathy and for an additional neuropathic process in DNM2-CNM. Finally, in accordance to our observations there have been several earlier reports suggesting (additional) peripheral nerve involvement in CNM (Engel, 1968; Mouren, 1982; Sugie, 1982; Reske-Nielsen, 1987).

References