Reply to: 'Advances in imaging of brain abnormalities in neuromuscular disease'

Josef Finsterer

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Correspondence to:
Josef Finsterer
Neurological Department, Krankenanstalt Rudolfstiftung, Messerli Institute, Postfach 20, 1180 Vienna, Europe, Austria
fifigs1@yahoo.de

I read the article by Angelini *et al*. about cerebral involvement on imaging in neuromuscular disorders (NMDs) with interest.[1] They found widespread cortical and subcortical white matter lesions (WMLs) and gray matter lesions (GMLs) in muscular dystrophies, WMLs, gyration abnormalities, and ventricular widening in congenital muscular dystrophies, cortical atrophy in LGMD2I, DMD, and BMD, and global atrophy in LGMD2I.[1] The study raises the following comments and concerns.

Cerebral involvement in NMDs may not only be found in dystrophinopathies, dystroglycanopathies, myotonic dystrophies, facio-scapulo-humeral dystrophy, limb–girdle muscular dystrophy, congenital myotonia, congenital myopathies, congenital muscular dystrophies, glycogenosis, and oculopharyngeal muscular dystrophy, but also in mitochondrial disorders (MIDs), beta-oxidation defects, Barth syndrome, envelopathies, and myofibrillar myopathies, which were not discussed in this review. It is also not discussed why these disorders were excluded.

Another shortcoming of the review is that only a portion of the cerebral abnormalities found on imaging in NMDs have been discussed. Missing are stroke-like lesions (SLLs), calcifications, pituitary adenoma, laminar cortical necrosis, the toenail sign, hyperperfusion on angiography, reduced oxygen extraction, optic atrophy, ischemic stroke, and aneurysms.[2]

Particularly included should be SLLs, which are the morphological equivalent of stroke-like episodes (SLEs), and pituitary adenoma as they are accessible to treatment. SLLs/SLEs may respond to antiepileptic drugs (AEDs), nitric oxide (NO) precursors, antioxidants, steroids, and the ketogenic diet.[3] AEDs should be given even in the absence of seizures or seizure activity on encephalography (EEG) as it may trigger spreading of a SLL or may trigger the development of SLL in general. Optic atrophy has been described in congenital muscular dystrophies,[4] MIDs, myotonic dystrophy,[5] or muscle eye brain disease.[6]

Techniques not discussed in this review include MR-spectroscopy (MRS) and oxygen extraction functional (OEF)-MRI. MRS provides useful information about metabolic changes, which may indicate altered regional brain metabolism or function. Of particular importance in this respect is whether there is focal, regional, or global cerebral lactate elevation, indicating disturbed oxidative metabolism, or epileptiform discharges.[7] Other metabolites that may be useful to demonstrate disturbed cerebral metabolism include N-acetylaspartate, choline, glutamate, and creatine.

Overall, this interesting review has a number of shortcomings, such as selection of certain but not all CNS abnormalities occurring in NMDs and considering not all techniques available to document CNS involvement in NMDs. in addition, the review could benefit from widening the spectrum of NMDs with CNS involvement or at least from explaining why certain NMDs were not included. Since cerebral involvement in NMD may remain subclinical it is recommended to screen all NMDs for cerebral disease.

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ORCID iD
Josef Finsterer https://orcid.org/0000-0003-2839-7305

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