Authors' reply

Corrado Angelini and Elena Pinzan

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Correspondence to:
Corrado Angelini
Fondazione Ospedale San Camillo, IRCCS, Venezia, Italia
corrado.angelini@unipd.it

Elena Pinzan
Fondazione Ospedale San Camillo, IRCCS, Venezia, Italia

Dear Dr. Finsterer,

We thank you for your appreciation of our review of 'Advances in imaging of brain abnormalities in neuromuscular disease'.[1] It is challenging to answer your remarks point by point.

In response to the observation regarding the neuromuscular disease (NMD) included, we clearly explained in our work the inclusion criteria. We decided to report only neuroimaging results in NMDs selected from 1980 to May 2018 on the main databases (PubMed, MedLine, Google Scholar), including only freely available full texts written in English. Our inclusion criteria were established as follows: 'We choose to consider the papers on the following NMDs: dystrophinopathies, dystroglycanopathies, myotonic dystrophies, facioscapulohumeral dystrophy, limb-girdle muscular dystrophies, congenital myotonias, congenital myopathies, and related terms or abbreviations'.[1]

We decided to cover a precise time period and to focus on selected muscle disorders while excluding others. We know that an ideal review would cover all scientific contributions about neuroimaging in NMDs: that is, mitochondrial and neurometabolic disorders possibly describing stroke-like lesions, calcification, or pituitary adenoma, but owing to the large amount of literature published on these disorders and, considering the structure of our work we decided to provide limitations in the research process owing to editorial constraints. In fact, the field of neuromuscular disorders has recently expanded and to our knowledge, approximately more than 600 different NMDs have been described.[2] For this reason, it would have impossible to cover the whole spectrum in only one review. We are aware that mitochondrial disorders have relevant cerebral involvement as reported in a paper on MELAS syndrome.[3]

Regarding techniques we stated to include 'computed tomography' or 'CT', 'magnetic resonance imaging' or 'MRI', 'single photon emission computed tomography' or 'SPECT', 'positron emission tomography' or 'PET', ultrasound, 'transcranial sonography' or 'TCS'. We included only those papers containing information about brain involvement.[1]

We excluded expensive investigations such MR-spectroscopy, since most of the data are experimental and not used in clinical practice.

Regarding oxygen extraction, functional MRI was in part discussed, but again this is a rather time-consuming research-oriented investigation and not useful for clinicians. Our approach was directed toward the diagnosis of cerebral abnormalities and their significance in the clinical setting rather than to cover all technological advancements. On a clinical basis, the cost efficiency of the investigation used should be considered.

We agree that some central nervous system abnormalities such as pituitary adenoma, aneurisms, or AV malformations might be treated by neurosurgery and other signs such as seizure prevented by antiepileptic drugs. The objective of the review was not to discuss treatment in general but only those relevant to neuromuscular patient for improving quality of life.

The wide range of aspects of brain involvement in NMDs might be the aim of a subsequent review that could include the other types of neuromuscular diseases.

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Conflict of interest statement

The authors declare that there is no conflict of interest.

ORCID iD

Corrado Angelini https://orcid.org/0000-0002-9554-8794

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