

Paris, January 10<sup>th</sup> 2019

Dear colleague neurologist,

EU has set up networks of expert centers: the European Reference Networks (ERNs), and the ERN for neuromuscular disorders is called EURO-NMD. These ERNs have been commissioned to offer cross border diagnostic expertise according to the Cross-border agreement directive within EU.

As the responsible clinician for your patients with neuromuscular disease you know how important the final and exact diagnosis is in order to provide the best possible management. This is particularly important as in some of these diseases heart involvement such as arrhythmia or cardiomyopathy also occurs, which should be treated by correct timing of preventive therapy. In other cases the correct diagnosis, even if genetic, can provide effective inexpensive treatment such as congenital myasthenias, riboflavin responsive MADD etc. On the other hand incorrect diagnosis may lead to harmful therapies as with long term steroids in dystrophies misdiagnosed with polymyositis. And patient associations rightly point out that the 'diagnostic odyssey' is also an emotional burden.

Because all neuromuscular diseases are either very rare or ultra-rare, finding the exact diagnosis can be very difficult or impossible without access to extended investigations such as up-to-date muscle pathology and molecular genetics. To meet these difficulties for patients with rare disorders if your neuromuscular patient does not have a final diagnosis you are allowed to make a referral for diagnostic investigations to the EURO-NMD according to the above-mentioned Cross-border agreement directive within EU. This directive secures that the expenses for examinations in another country will be covered by the national health authorities.

- In case your patient does not already have comprehensive medical records based on conventional examinations (muscle histopathology with cryotechnique, muscle imaging, molecular genetic DNA-tests) you may refer your patient to one of the centers within the EURO-NMD for such examinations, or directly to the EURO-NMD coordinator for advice how to proceed.
- In case your patient already has had very comprehensive examinations you may contact the EURO-NMD coordinator and get access to the EU-secured on-line platform CPMS for sharing the protected patient data with the experts within the EURO-NMD, and have their corresponding evaluation and conclusions on your case. This on-line procedure is free of charge but requires good medical record data to be analyzed by the experts.
- All contacts are available through the EURO-NMD home page: <https://ern-euro-nmd.eu/>

Sent by the EURO-NMD team and distributed by your national association of neurologists

On behalf of the ERN EURO-NMD

The coordinator

A handwritten signature in black ink, consisting of a large, stylized initial 'T' followed by a horizontal line and a vertical stroke.

Teresinha Evangelista