Diagnostic track in amyotrophic lateral sclerosis: the Brazilian experience

O processo diagnóstico na esclerose lateral amiotrófica: a experiência brasileira

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Diagnostic delay in amyotrophic lateral sclerosis (ALS) is a relevant problem that has been investigated in different countries for a long time. This delay has impact on the number of unnecessary investigations, social and personal disease economic burden, anxiety levels related to uncertain medical decisions, and increased risk of needless surgeries.[1] A wrong-surgery tends to further delay the diagnosis and to promote faster disease progression, a circumstance confirmed by neurologists dedicated to ALS and supported by scientific data.[2] Additionally, a diagnosis delay decreases the chances of clinical trial participation. Shortening diagnostic delay has been the rational for establishing the Revised-El Escorial Criteria,[3] the Awaji proposal (which is mainly focused on the neurophysiological features of ALS),[4] and the Gold Coast Criteria.[5] This demonstrates the importance of this subject, and the strong involvement of different institutions and experts on improving this constraint.

More recently, the median or mean time (variable in different sources) between the first symptoms (related to muscle weakness) and diagnosis is between 10 and 15 months, in general.[6] Consistently, bulbar-onset disease[7,8] and a faster observation by a neurologist has been associated with a shorter diagnostic delay.[1,9,10] Misdiagnosis is quite common in ALS, although quite variable in different centers.[6] Unquestionably, an initial wrong diagnosis delays ALS diagnosis, and increases the risk of inadequate medical interventions. It is uncertain if the modifications of the diagnostic criteria had a positive impact on decreasing delay of diagnosis in ALS.

The medical practice is different between countries, which depends on the health system organization, customary medical referrals routines, and other cultural influences. In Europe, medical care is first provided by general medical practitioners, who are available without costs, but in other countries this option is not so accessible. Moreover, it is reasonable to consider that the patient's salary income can influence the diagnostic track.[10] For this reason, investigating the reality in each country is recommended, to endorse the appropriate interventions for each setting.

In this issue, Borthetti et al. retrospectively investigated the diagnostic track and misdiagnosis in a population of 173 ALS patients followed in Ribeirão Preto, Brazil.[11] They concluded that misdiagnosis was common in their population (70%), and that 7% of the patients underwent to unnecessary surgeries. In particular, limb-onset patients were more frequently observed by orthopedic surgeons, who tended to diagnose root or spinal cord lesions, and bulbar-onset patients were more commonly observed by ENT specialists. This suggests that these specialists could benefit from more information about clinical manifestations of ALS. From our experience, information about initial respiratory symptoms in ALS should be shared with pulmonologists and cardiologists, as 4% of ALS patients present with respiratory-onset and are followed by those specialists.[12]

Electromyography investigation has a critical role in the diagnosis of ALS, and is an essential step to reduce misdiagnosis and shorten diagnostic delay.[10] This point was not approached in this study, but further research is welcomed.

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Finally, it should be said that ALS is a difficult clinical situation. Sometimes, neurologists decide to defer diagnosis since it is uncomfortable to communicate this dreadful disease. We need more information, and this study is important for our current knowledge.

Conflict of Interest
The author has no conflict of interests to declare.

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