Diagnosis

Patients with symptoms suggestive of ALS should be assessed as soon as possible by an experienced neurologist. Early diagnosis should be pursued, and investigations, including neurophysiology, performed with high priority. The patient should be informed of the diagnosis by a consultant with a good knowledge of the disease and the patient (Andersen and others 2012).

Andersen and others 2012

Our objective is to present guidelines for making the correct diagnosis as early as possible. As no single investigation is specific to ALS, and there is no sensitive and disease specific biomarker, diagnosis is based on symptoms, clinical examination findings and the results of electrodiagnostic, neuroimaging and laboratory studies.

Chio and others 2009

Several studies, but not all, have found that a longer delay from symptom onset to diagnosis carries a better prognosis. In general, the finding of a worse prognosis in patients with a short time delay between onset and diagnosis is likely to indicate a more aggressive disease, which leads the patient to seek medical attention more rapidly and is more readily diagnose

Vucic and others 2007

Currently, there is no pathognomonic test for the diagnosis of motor neurone disease

Clinicians rely on the presence of upper and lower motor neurone symptoms and signs to make the diagnosis of MND

Neurophysiological, neuroimaging and laboratory investigations are routinely applied in the diagnostic work-up of patients who may have MND to identify subclinical involvement and to exclude the presence of potentially treatable MND-mimic disorders

Further development of existing imaging and neurophysiological techniques may enable a more definitive early diagnosis of MND

Andersen and others 2007

The diagnosis should be pursued as early as possible. Patients of whom ALS is suspected should be referred with high priority to an experienced neurologist.

All suspected new cases should undergo prompt detailed clinical and paraclinical examinations.

In some cases, additional investigations may be needed.

Repetition of the investigations may be needed if the initial series of tests do not result in a diagnosis.

Review of the diagnosis is advisable if there is no evidence of progression or if the patient develops atypical features.