# Panel of investigations for Motor Neurone Disease

## Suspected MND-Essential Tests

| Blood test | Imaging | Neurophysiology |
|---|---|---|
| • FBC<br>• Plasma viscosity and ESR<br>• U&Es<br>• Creatine kinase<br>• Calcium and phosphate<br>• Protein and LFTs<br>• B12 and Folate<br>• Blood sugar<br>• Serum protein electrophoresis<br>• Thyroid function<br>• Vitamin D<br>• ANA and ANCA<br>• HIV and Syphilis screen | Magnetic resonance imaging (MRI)<br>• Head<br>• Cervical spine<br>• Thoracic<br>• Lumbosacral<br><br>Chest x-ray<br><br>ECG<br><br>Pulmonary function test | Electromyography<br>Nerve conduction studies |

## Suspected MND-Additional Tests for Atypical Presentations

| Blood test | Imaging | CSF analysis | DNA analysis |
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| • White cell enzymes (Serum hexosaminidase A & B)<br>• Borrelia titres<br>• Anti-GM1 ganglioside Ab's<br>• Anti-MAG antibodies<br>• Blood and urine lead<br>• Tumour markers<br>• Very long chain fatty acids(*adrenomyeloneuropathy*)<br>• Acid alpha-1,4 glucosidase (acid maltase) *[Pompe's disease]*<br>• Paraneoplastic antibodies(cerebellar antibodies screen) | Single Photon Emission Computed Tomography (SPECT) (Mill's syndrome)<br><br>Base of skull imaging (CT+/-MRI)<br><br>CT chest/abdomen/pelvis | Protein<br>Cells<br>Glucose<br>Oligoclonal bands<br>Cytology | ***Gene*** / ***Disease***<br>SOD1 gene mutations — *Familial autosomal dominant MND*<br>Androgen receptor mutation — *Kennedy's syndrome*<br>Spastin and other HSP-related genes — *Progressive UMN syndromes*<br>Neurofilament light chain gene mutations — *Charcot-Marie-Tooth type 2E*<br>SMN gene mutations — *Late onset SMA (Spinal muscular atrophy)*<br>Dynactin gene mutations — *Familial LMN disorder*<br>Hexosaminidase gene mutations — *Juvenile or early onset MND* |

## Confirmed MND- For patients taking Riluzole

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|---|---|
| **Blood test**<br>• FBC<br>• U&E<br>• LFT | Monthly for the first 3 months, followed by 3 monthly for the following 9 months and annually thereafter.<br>Stop Riluzole if the ALT is 5 times the normal limit |

For MNDA Leeds website and ordercom panel of investigation for LTHT