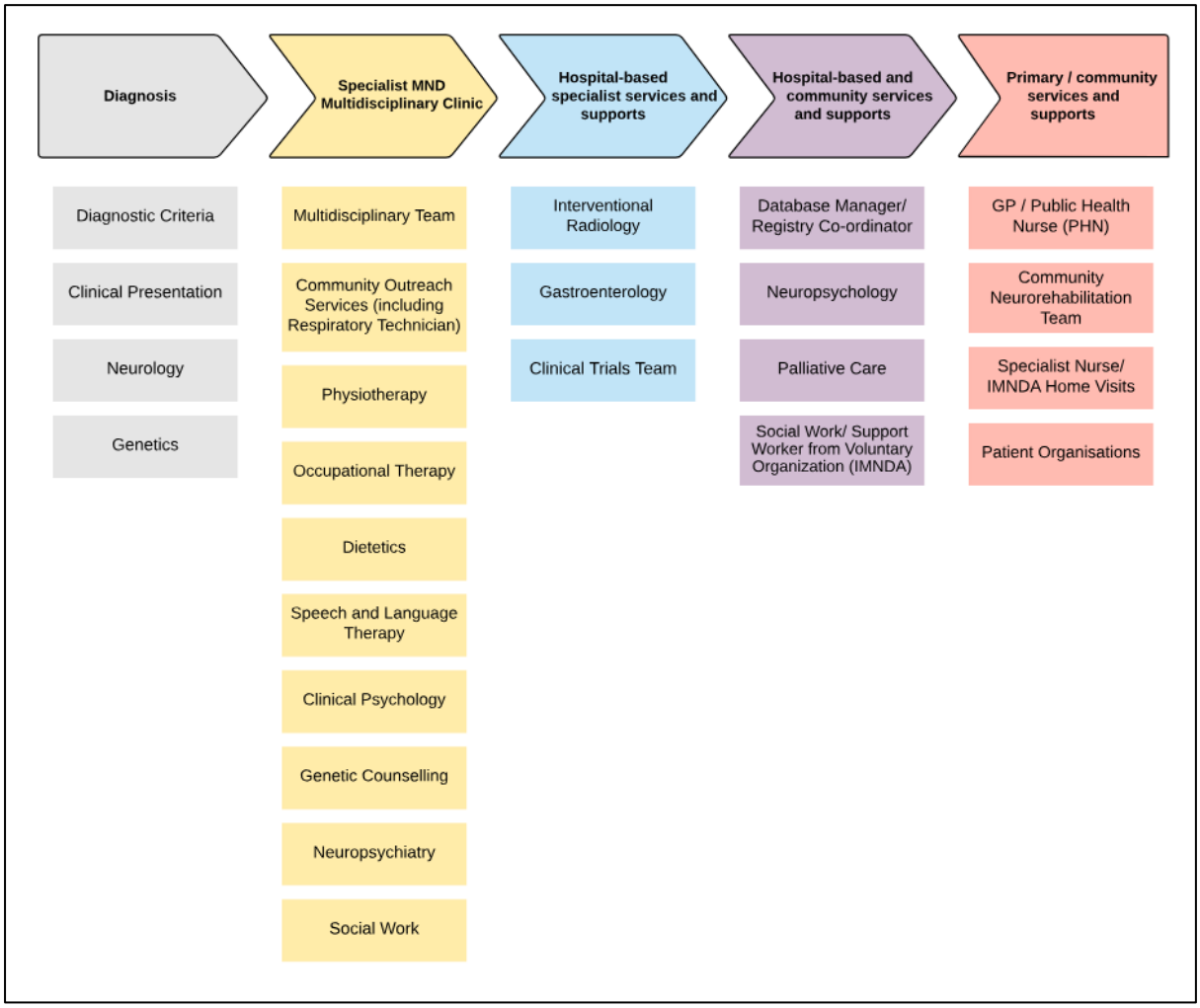
# Amyotrophic Lateral Sclerosis (ALS) Adult Care Pathway

HSE National Rare Diseases Office

# Amyotrophic Lateral Sclerosis (ALS) Adult Care Pathway




## Clinical Characteristics:

**Orphacode: 803**

**Disease Definition**: A neurodegenerative disease characterized by progressive muscular paralysis reflecting degeneration of motor neurons in the primary motor cortex, corticospinal tracts, brainstem and spinal cord

| Diagnosis | Diagnostic Criteria | Gold Coast Criteria (Shefner et al. 2020) |
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| Diagnosis | Clinical Presentation | Spinal onset (72% Irish data) - present with symptoms related to focal muscle weakness and wasting with stiffness, upper and lower limbs;<br>Bulbar onset (28%) - Dysphagia and dysarthria;<br>Minority (15%) present with cognitive or behavioural abnormalities fulfilling diagnostic criteria for frontotemporal dementia, fasciculations, cognitive and behavioural decline (50%), progressive paralysis and respiratory failure generally within 2-5 years |
| Diagnosis | Neurology | EMG and nerve conduction studies (neurogenic changes) - normal motor and sensory nerve conduction, absence of conduction block; blood tests (ESR, CRP, FBC, LFTs, Vitamin B12 and folate, thyroid function tests, serum protein electrophoresis, CK, electrolytes, glucose, ACE);<br>MRI/CT of brain and spinal cord;<br>Lumbar puncture if appropriate |
| Diagnosis | Genetics | 10-15% familial;<br>50% of familial cases in Ireland associated with C9orf72 repeat expansion;<br>Variant may also be present in non-familial cases;<br>Other known genes (*SOD1*, *TDP43* not prevalent in the Irish population);<br>FUS variants in young population<br>Ataxin 2 expansion Modifies risk<br>UNC13A modifies risk<br>Genomic sequencing of known genes |
| Specialist MND Multidisciplinary Clinic | Multidisciplinary Team | OPD review 6-8 week intervals;<br>Muscle cramps - quinine sulphate;<br>Spasticity - baclofen, tizanadine;<br>Emotional lability - amitriptyline, fluvoxamine or citalopram;<br>Depression – evidenced-based psychological interventions, amitriptyline, SSRIs or mirtazapine;<br>Anxiety – evidenced-based psychological interventions & supportive counselling (Palliative services and supports);<br>Respiratory management using non-invasive ventilation;<br>Prevent Deep Vein Thrombosis;<br>Condition modifying therapy: Riluzole;<br>Clinical trials where eligible;<br>Education, home services and supports, patient organisations, home visits; |

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| | | Treatment decision-making, advance care directives, end of life decision-making and care |
| Specialist MND Multidisciplinary Clinic | Community Outreach Services (including Respiratory technician) | Monitor for respiratory condition, respiratory function assessment each clinic visit (Forced Vital capacity, Sniff Nasal Inspiratory Pressure, Peak cough flow)<br>Home monitoring using hand-held spirometry (COVID19 restrictions).<br>Non-invasive positive pressure ventilation (initiated at home by MDT – outreach specialist nurses).<br>Sialorrhoea - suctioning, amitriptyline, oral or transdermal hyoscine, sublingual atropine drops;<br>Refractory sialorrhoea - botulinum toxin injections into parotid and/ or submandibular gland, salivary gland irradiation;<br>Bronchial secretions - humidification, nebulisers, mucolytics (if sufficient cough flow) - guaifenesin, N-acetyl cysteine, beta-receptor antagonist and/ or anticholinergic bronchodilator;<br>Mechanical insufflator-exsufflator;<br>Treat respiratory tract infections;<br>Manage respiratory failure |
| Specialist MND Multidisciplinary Clinic | Physiotherapy | Respiratory:<br>Respiratory insufficiency – decreased lung volume, decreased cough strength, decreased expiratory muscle strength:<br>Monitor for symptoms of respiratory insufficiency<br>Lung volume recruitment training – breath stacking, assisted breath stacking<br>Chest clearance techniques – mechanical in-exsufflator (cough assist), assisted cough<br>Breathing control with postural correction<br>Oxygen therapy<br>Non-invasive Ventilation<br><br>Neuromusculoskeletal:<br>Gait abnormalities, Progressive muscle weakness and paralysis (end-stage), Spasticity - risk of contractures, postural changes with joint immobility, high falls risk, progressive muscle weakness and atrophy with resultant joint instability (shoulders, hips, ankles) and decreased grip strength:<br>Preventative, compensatory and restorative strategies and interventions<br>Prescribe, monitor and provide guidance on activity and exercise – gentle low impact aerobic exercise<br>Postural monitoring and management program<br>Prescription of assistive devices or orthotics<br>Pain management – passive stretching, joint mobilization and range of motion exercises; education on proper joint support and protection |

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| | | Patient, family and caregiver education and training<br>Moving and handling risk assessment<br>Falls prevention strategies – balance training<br>Physiotherapy is tailored to the individuals needs and goals to improve quality of life (supportive treatment)<br>Avoid overwork damage and excessive fatigue, consider isometric exercise<br>Patients should be offered regular monitoring and review |
| Specialist MND Multidisciplinary Clinic | Occupational Therapy | Assessment/review of daily living skills, such as functional transfers, functional mobility, feeding, self-care. Assessment/review of Instrumental activities (activities) of daily living skills, such as accessing the community and domestic skills<br>Completion of standardised assessments.<br>Signposting anticipatory needs.<br>Interventions may include postural management and specialist seating including powered mobility.<br>Environmental adaptations in the home and school including housing adaptation support and recommendations.<br>Promoting participation in school environment within the classroom / yard<br>Specialist equipment for feeding and aids for independence<br>Specialised equipment such as, hoists, shower chairs, slings, Upper limb assessments / splinting / casting<br>Preparation for transition to adult services<br>Accessing leisure activities within community<br>Pain/fatigue management<br>Energy conservation techniques<br>Sensory processing<br>Stress management<br>Sleep hygiene<br>Assessment for assistive technology to access educational curriculum<br>Additional input with Children / adults who present with coexisting condition such as ADHD or Autism and those with executive functioning difficulties |
| Specialist MND Multidisciplinary Clinic | Dietetics | Nutritional assessment and regular screening for risk of malnutrition, to ensure adequate nutritional and hydration status<br>Work closely with Speech and Language Therapist to manage dysphagia<br>Provide dietary education on texture modified diets<br>Invitation of early nutrition support (as indicated)<br>Involved/support in the decision-making processes around gastrostomy feeding<br>Pre and post gastrostomy tube feeding education, monitoring and ongoing support |

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| Specialist MND Multidisciplinary Clinic | Speech and Language Therapy | Assessment, support and intervention for speech, language and communication needs (SLCN) and/or feeding, eating, drinking & swallowing (FEDS) skills based on individual needs / priorities<br>Dysphagia management<br>Voice banking<br>Alternative and augmentative communication aids<br>Patient advocacy<br>Carer / staff / patient education, training and awareness |
| Specialist MND Multidisciplinary Clinic | Clinical Psychology | Assessment for cognitive and behavioural condition<br>Psychological interventions for mental health, behavioural and cognitive difficulties in patients or psychological presentations in family caregivers<br><br>Specialist Rare Disease Centres of Expertise will aim to provide specialised psychology support and liaison psychiatry (when available). An outreach shared care model can be established (when requested) at community level (with support as needed from the specialist centre) |
| Specialist MND Multidisciplinary Clinic | Genetic Counselling | Counselling pre- and post-testing of symptomatic individuals. Testing of adult (over 18 years) pre-symptomatic family members as appropriate in line with EFNS taskforce guidelines (2012)<br>Discussion of all reproductive options with at-risk adults of child-bearing age |
| Specialist MND Multidisciplinary Clinic | Neuropsychiatry | Pharmacologic management of psychosis/ anxiety/ depression/ behavioural change |
| Specialist MND Multidisciplinary Clinic | Social Work | Psychosocial support: Assess social and family supports, safeguarding<br><br>Link with community supports as required e.g., GP, Public Health Nurse, Primary Care SW, Local authority SW, Mental Health SW, Disability SW, TUSLA<br><br>Offer 1-1 counselling or GP referral to Counselling in primary care (CIPC) www.hse.ie/eng/services/list/4/mental-health-services/counsellingpc/<br><br>Financial support (as required): Patient advocacy, support applications for Medical Card, Disability |

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| | | Allowance, Supplementary Welfare allowance, Exceptional Needs payment, Long-term illness card, direct to Citizens' information www.citizensinformation.ie/<br><br>Housing and / or mobility issues: Advocacy and support<br><br>Home Care Packages: if issues with activities of daily living - arrange application for inpatients, liaise with Public Health Nurse to arrange for out-patients<br><br>Respite Care: liaise with public health nurse or community disability services to arrange<br><br>Employment issues:<br>Link to Intreo public employment<br>www.gov.ie/en/campaigns/fb84c0-intreo/<br>EmployAbility services<br>www.gov.ie/en/service/8578c4-access-the-employability-service/ |
| Hospital-based specialist services and supports | Interventional Radiology | Radiologically Inserted Gastrostomy (RIG) |
| Hospital-based specialist services and supports | Gastroenterology | Percutaneous Endoscopic Gastrostomy (PEG) |
| Hospital-based and community services and supports | Database Manager/Registry Co-ordinator | Population-based registry<br>Create and maintain database of patients attending service<br>Record patient biographical, clinical and research data<br>Audit and quality improvement<br>Ensure minimum data set standards for ERN registry |
| Hospital-based and community services and supports | Neuropsychology | Assessment for cognitive and behavioural condition<br>Psychological interventions for mental health, behavioural and cognitive difficulties in patients<br>Link with liaison psychiatry |
| Hospital-based and community services and supports | Palliative care | Treatment decision-making, advance care directives, end of life decision-making and care |
| Hospital-based and community services and supports | Social Work/ Support Worker from Voluntary Organization (IMNDA) | Safeguarding<br>Psychosocial support<br>Financial advice & support<br>Housing and mobility issues - advocacy & support<br>Home Care Packages<br>Respite Care<br>Employment issues |

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| Primary / community services and supports | GP & Public Health Nurse (PHN) | Red flag education diagnostic tool red-flags-final-2.pdf (mndassociation.org)<br>Integration with specialist palliative care services<br>Management of inter-current conditions<br>Co-ordination of local services and supports<br>Refer/communication across services |
| Primary / community services and supports | Community Neurorehabilitation team | Neurorehabilitation services and supports |
| Primary / community services and supports | Beaumont Hospital Specialist Nurses/ IMNDA Home Visits Beaumont Clinical Professional Outreach | IMNDA nurse - support across the patient journey from diagnosis, hospital, MDT, community through to palliative care<br>Support for chronic condition, depression, anxiety<br>Symptom management, end of life care, advance care directives |
| Primary / community services and supports | Patient organisations | Advocacy, support and information:<br>Irish Motor Neuron Disease Association (www.imnda.ie)<br>Rare Diseases Ireland (www.rdi.ie ) |

**CLINICAL LEAD**:

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| Prof. Orla Hardiman, Beaumont University Hospital |

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