RAPID RECOGNITION OF NEUROMUSCULAR WEAKNESS

THE PRIMARY CARE DIAGNOSTIC PATHWAY

CRITICAL WARNINGS & RED FLAGS (EMERGENCY)

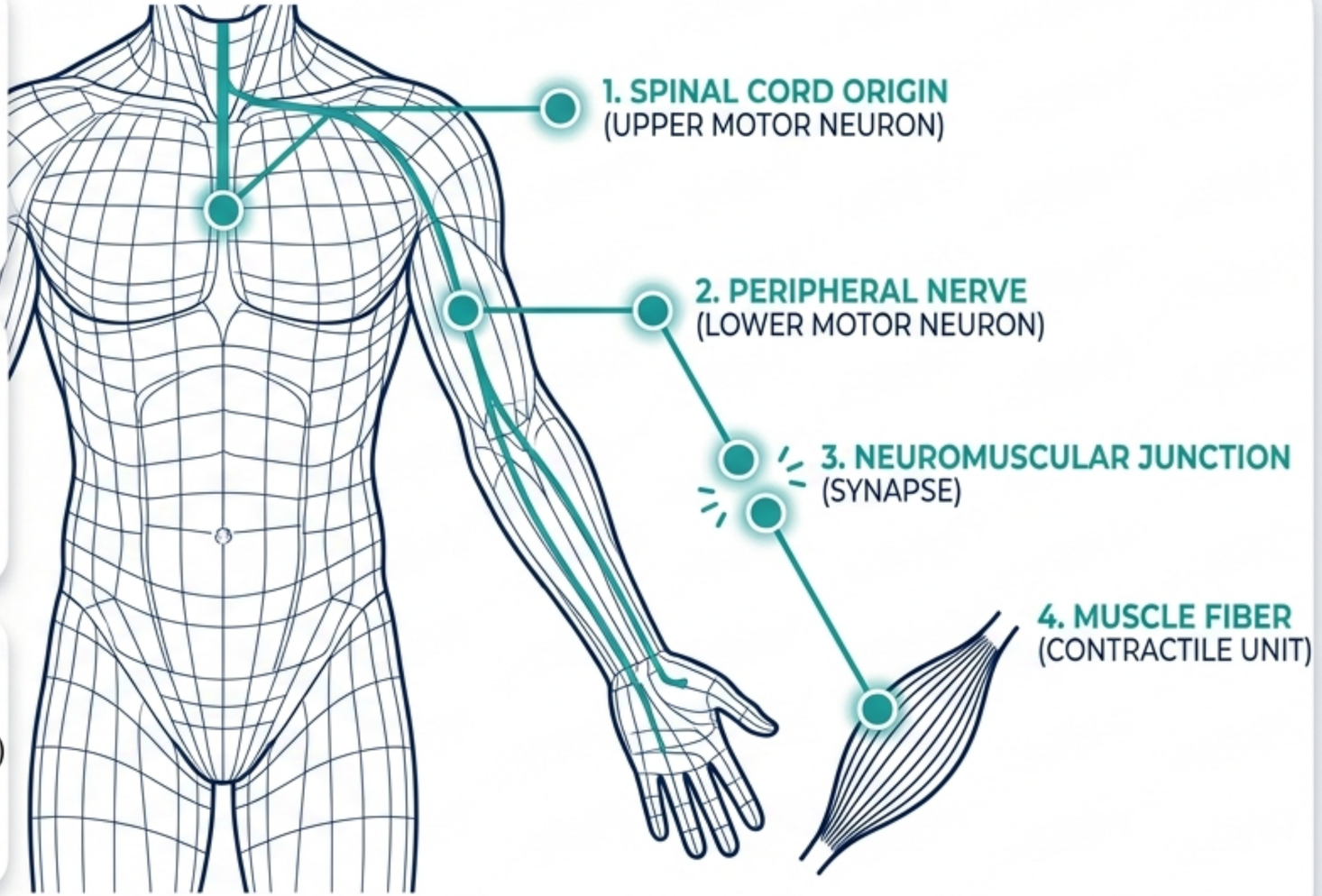
- **Acute Respiratory Failure**
- **Rapid Onset, Progressive Bulbar Symptoms** (Swallowing Difficulty, Dysarthria)
- **Signs of Brainstem or Cranial Nerve Involvement**
- **Autonomic Instability**
- **Rapidly Ascending Weakness** (e.g., Guillain-Barré Syndrome)

DIAGNOSTIC WARNINGS & TRANSITIONAL STATES

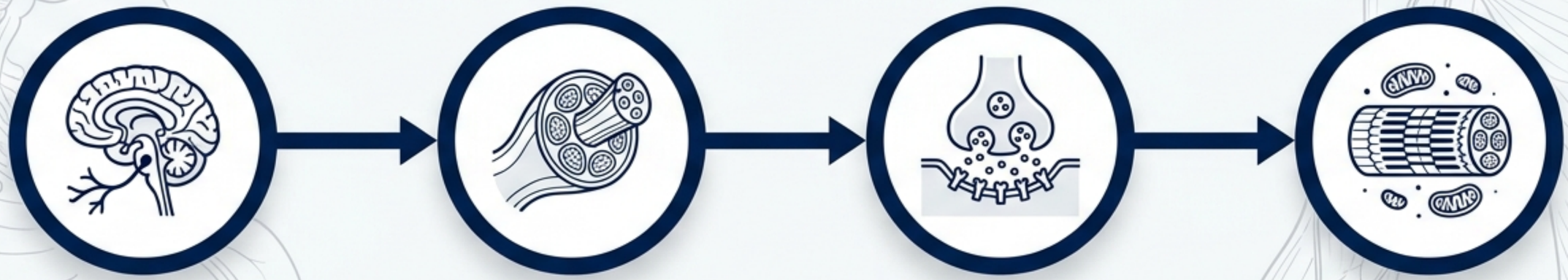
- **Fluctuating Weakness** (e.g., Myasthenia Gravis)
- **Subacute, Symmetric Proximal Weakness** (e.g., Myopathies)
- **Fatiguability** or **Post-Exertional Weakness**
- **Asymmetric Weakness** or **Sensory Deficits**
- **Delayed Reflexes** or Absent DTRs

STANDARD TREATMENT PATHWAY & NORMAL FINDINGS

- **Normal Sensation and Reflexes** (Early Stage)
- **Initial Neurologic Exam within Normal Limits** (Except for Isolated Weakness)
- **Baseline Blood Work** (CPK, Electrolytes, TSH, ESR/CRP) WNL
- **Patient Education & Close Monitoring**
- **Referral to Neurology** for Advanced Testing (EMG/NCS)



The Neuromuscular Axis: Mapping the Pathway



Node 1:
Anterior Horn Cell
(Brain/Cord)

Node 2:
Peripheral Nerve

Node 3:
Neuromuscular Junction

Node 4:
Muscle Fiber

Australian Epidemiology Snapshots

Myasthenia Gravis

150–250

per million.
Peaks: F 20-30, M 60-80

Guillain–Barré Syndrome

1–2

per 100,000 person-years

Inflammatory Myopathies

5–10

per million per year

Myotonic Dystrophy Type 1

1 in 8,000

Australians

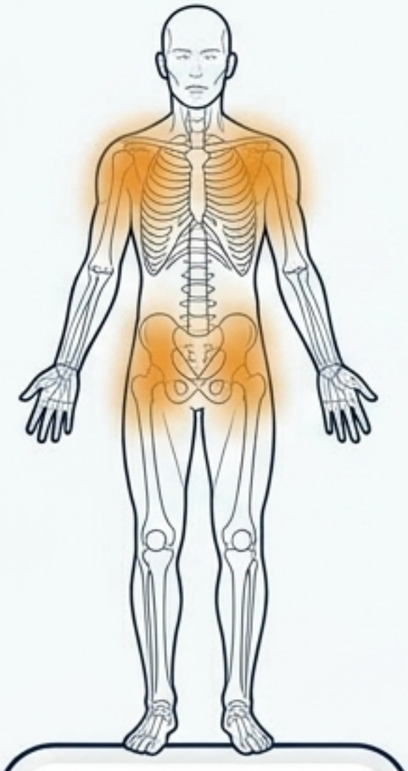
Motor Neuron Disease

2–3

per 100,000 per year.
Median survival 2–3 years

Critical Warning: Rapid Progression

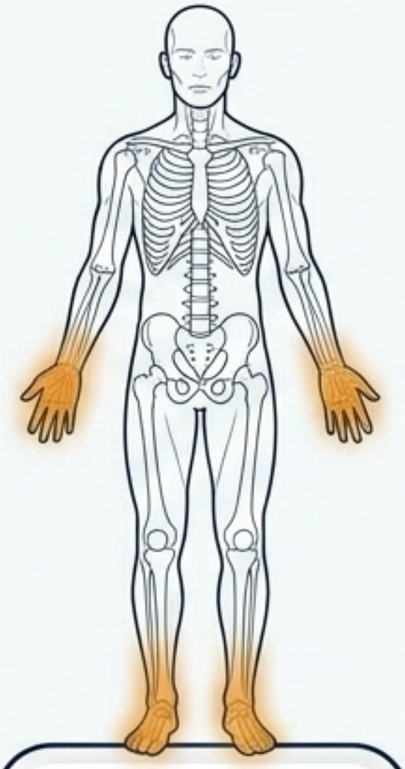
Pattern Recognition: The Anatomical Heatmaps



Proximal

Activities: Rising from chair, overhead tasks.

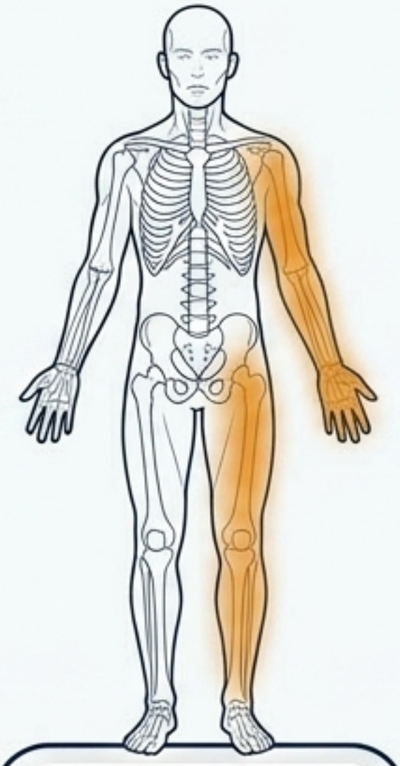
Localization: Muscle or NMJ.



Distal

Activities: Foot drop, grip, buttons.

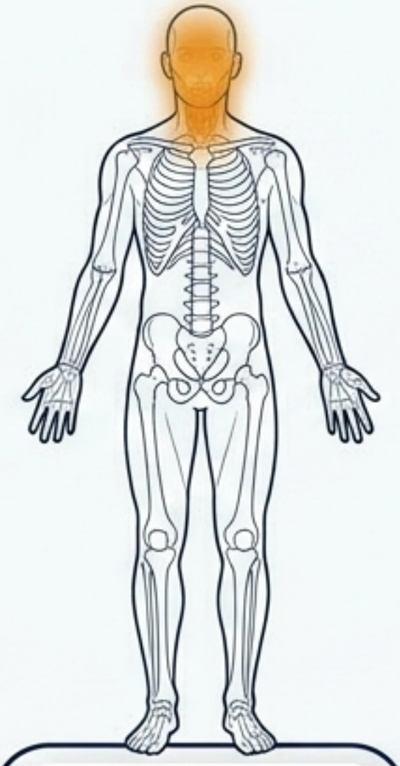
Localization: Peripheral nerve or Anterior Horn.



Focal / Asymmetric

Activities: Unilateral dragging/weakness.

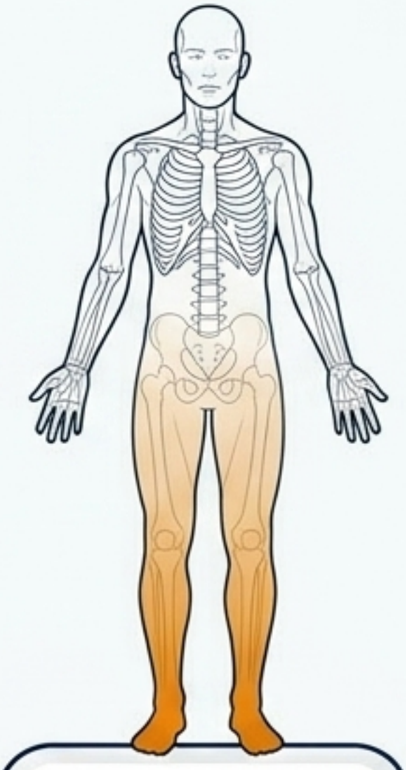
Localization: Nerve root, plexus.



Bulbar

Activities: Dysphagia, nasal speech.

Localization: Brainstem, NMJ, Bulbar muscles.



Ascending

Activities: Progressive bilateral weakness over days.

Localization: Demyelinating peripheral nerve.

Bedside Screening: Reflexes & Fatigability

The Reflex Decoder

Areflexia / Hyporeflexia

RED FLAG: Guillain-Barré, severe neuropathy, or advanced myopathy.

↓

Normal Reflexes in Weak Muscles

Consider NMJ disorder (Myasthenia) or early myopathy.

↓

Hyperreflexia with Weakness

Upper Motor Neuron pathology (Cord compression, MND). Not a primary muscle/NMJ issue.

The Fatigability Dashboard (NMJ/Myasthenia)

Sustained Upgaze

Metric: 60 seconds.

Positive: Ptosis or diplopia.

Counting Test

Metric: Count aloud 1 to 50.

Positive: Progressive nasal speech or hypophonia.

Arm Raise

Metric: Abducted 90° for 2 minutes.

Positive: Proximal arm drop.

Ice-Pack Test

Metric: Apply for 2 minutes.

Positive: Ptosis improves by ≥2 mm.

The "Big Three" Neurological Emergencies

Guillain-Barré Syndrome (GBS)	Myasthenic Crisis	Spinal Cord Compression
Pathology Acute immune demyelination. Often post-infection (Campylobacter).	**Pathology** NMJ exacerbation. Triggered by infection or medication changes.	**Pathology** Structural Upper Motor Neuron lesion. Metastatic, abscess, or disc.
Defining Triad Ascending weakness + Areflexia + Monophasic course.	**Defining Triad** Bulbar failure + Respiratory weakness + Limb fatigability.	**Defining Triad** Bilateral leg weakness + Sensory level + Sphincter dysfunction.
Acute Action IVIg or Plasma Exchange.	**Acute Action** Intubate early + IVIg/PLEX. Stop pyridostigmine temporarily.	**Acute Action** Dexamethasone 10mg IV stat + Emergent MRI <24h.

Guillain-Barré Syndrome (GBS)

Clinical Profile

- **Brighton Criteria (Modified):** Bilateral limb weakness (progressive over days-4wks) + Generalised Areflexia.
- Associated with *Campylobacter jejuni* infection.
- **Miller Fisher variant:** Ophthalmoplegia + Ataxia + Areflexia (anti-GQ1b).
- CSF shows Albuminocytological dissociation (elevated protein, normal cells).

Intervention Options

Treatment A (First Line)

Intravenous Immunoglobulin (IVIg)

0.4 g/kg/day IV for 5 days. PBS Authority Required.

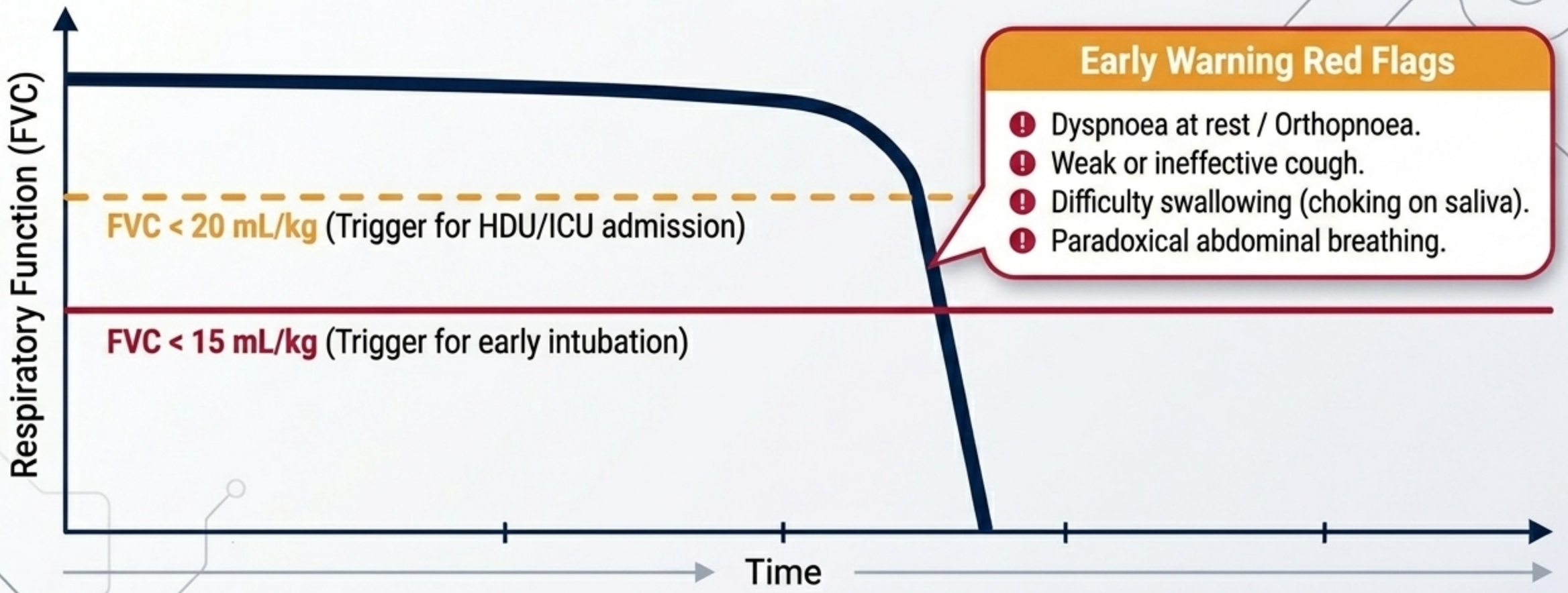
Treatment B (Alternative)

Plasma Exchange (Plasmapheresis)

5 exchanges over 8-10 days. Hospital-funded.

WARNING:
DO NOT COMBINE treatments. Corticosteroids are NOT effective in GBS.

The Respiratory Cliff: Recognizing Impending Failure



Early Warning Red Flags

- Dyspnoea at rest / Orthopnoea.
- Weak or ineffective cough.
- Difficulty swallowing (choking on saliva).
- Paradoxical abdominal breathing.

Key Takeaway: In GBS and Myasthenia, respiratory deterioration occurs rapidly. Admit patients with ascending weakness and areflexia immediately, even if breathing appears normal at presentation.

Spinal Cord Compression: The 24-Hour Protocol

Critical Window: < 24 Hours

Step 1: Recognise

Bilateral leg weakness + Sensory level (band-like) + Sphincter dysfunction (urinary retention).

Flaccid (shock) -> Spastic.

Step 2: Steroidise

Dexamethasone 10 mg IV stat immediately.

Do not wait for imaging to administer.

Step 3: Image

Emergent MRI whole spine (with contrast if malignancy/abscess suspected).

Step 4: Refer

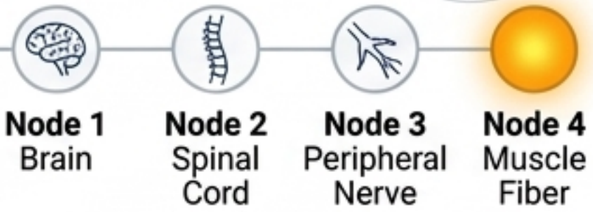
Neurosurgery / Spinal Surgery or Radiation Oncology.

Legend: ● High/Present | ◑ Variable/Moderate | ○ Absent/Low

The Slow Burn: Inflammatory vs. Hereditary Myopathy

	Inflammatory Myopathy	Hereditary Myopathy
Onset Speed	Subacute (weeks/months)	Insidious (months/years/childhood)
Family History	○	● Dominant/Recessive/X-Linked
CK Level Elevation	● 5-50x ULN	◑ Variable; marked in DMD, normal in others
Skin Changes	◑ DM only	○
Myotonia	○	◑ Myotonic dystrophy only
Steroid Response	● High, except IBM	○

Inflammatory Myopathies & The IBM Diagnostic Trap



Polymyositis (PM) & Dermatomyositis (DM)

Presentation: Proximal weakness, subacute onset. DM features skin changes (heliotrope rash, Gottron's papules).

The Malignancy Screen (DM only): Highly associated (20-25%). Requires CT chest/abdo/pelvis, pelvic ultrasound (ovarian screen), PET-CT.

Look for anti-TIF1-γ / anti-NXP2 antibodies.

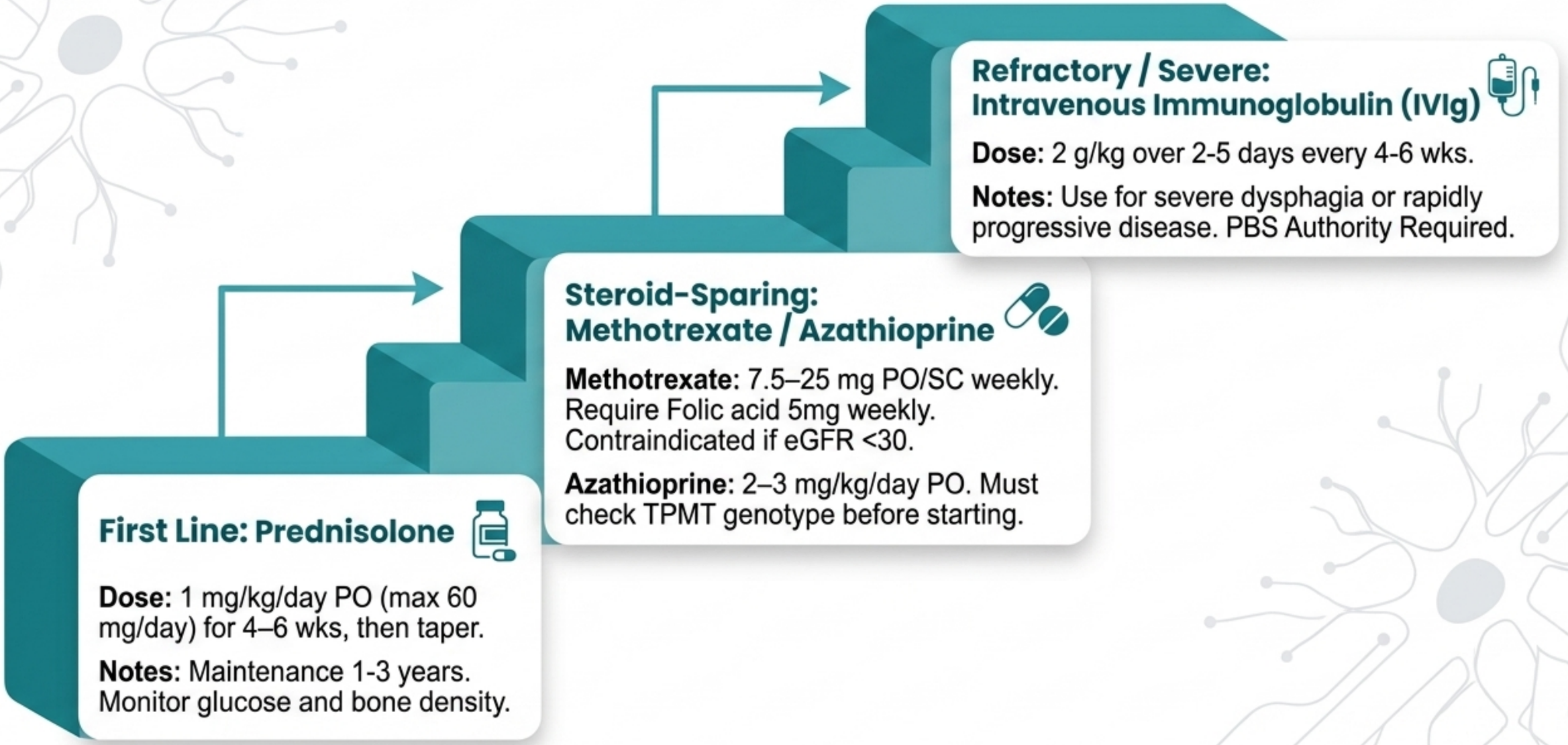
The Inclusion Body Myositis (IBM) Trap

Profile: Most common acquired myopathy in >50 years.

Unique Pattern: Uniquely involves finger flexors (opening jars) and quadriceps (falls). CK often normal or mild. Anti-cN1A positive.

THE TRAP: POOR or NO response to immunosuppression. Do not empirically treat older adults for PM without a biopsy. **Prolonged steroids cause severe harm.**

The Immunosuppression Ladder (PM / DM)



Hereditary Myopathies: Defining Clinical Profiles

Duchenne (DMD)

- **Inheritance:** X-linked recessive (DMD gene).
- **Profile:** Boys 2-5 yrs. Gowers' sign, calf pseudohypertrophy. **CK >10,000 U/L.**
- **Action:** Corticosteroids start from age 4-6.

Facioscapulohumeral (FSHD)

- **Inheritance:** Autosomal dominant (D4Z4 / SMCHD1).
- **Profile:** Asymmetric facial weakness (inability to whistle), prominent scapular winging, foot drop.
- **Labs:** Normal or mild CK.

Myotonic Dystrophy (DM1)

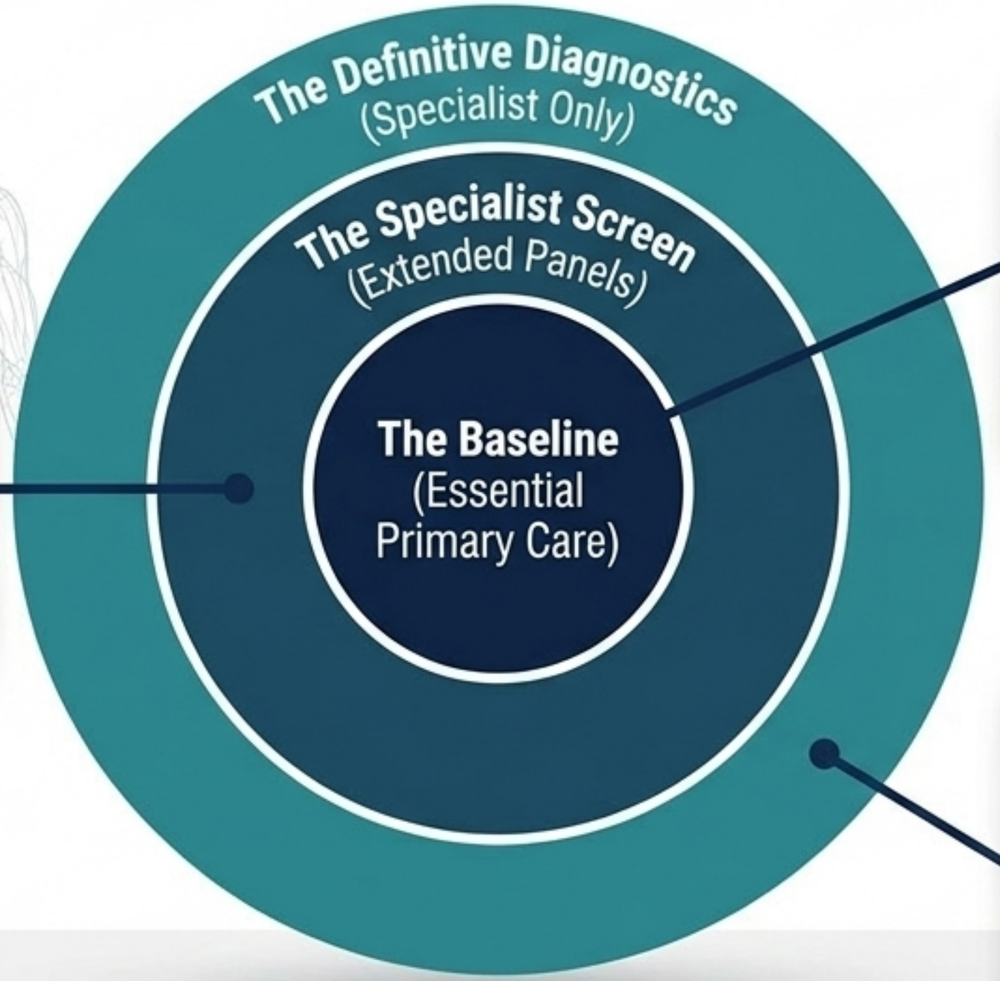
- **Inheritance:** Autosomal dominant (DMPK CTG expansion).
- **Profile:** Distal weakness, grip myotonia, 'hatchet face', cataracts, cardiac conduction defects.

Warning: Avoid suxamethonium in anaesthesia.

Limb-Girdle (LGMD)

- **Inheritance:** AD or AR (Multiple genes).
- **Profile:** Proximal shoulder and hip weakness. Variable age of onset.
- **Action:** Requires comprehensive next-gen sequencing panel.

The Diagnostic Radar: Tiered Investigations



- **Tests:** Autoimmune & Myositis specific (Anti-Jo-1, Mi-2, SRP), AChR antibodies (Positive in 85% generalized MG), SPEP, Vit D, B12/Folate, LFTs.

- **Tests:** Serum CK (The ultimate filter), **TSH/fT4** (Reversible myopathy), Electrolytes (K, Ca, **Electrolytes** (K, Ca, Mg), **HbA1c** (Diabetic neuropathy), **FBC/CRP**/ESR.

- **Tests:** NCS/EMG (differentiates demyelinating vs axonal/myopathic), Muscle Biopsy (Gold standard for IBM/PM), Muscle/Spine MRI, Genetic Panels.

Contextual Care: Special Populations (I)

Pregnancy

- **Pyridostigmine** (Mestinon) is safe. **Methotrexate** and **Azathioprine** are teratogenic ⚠
- Monitor GBS closely due to naturally reduced FVC in pregnancy.
- DM1 requires mandatory **pre-conception genetic counseling**.

Paediatrics

- Refer DMD early; deflazacort/prednisolone protocol starts age 4-6 ⚠
- Juvenile MG requires specific paediatric **pyridostigmine dosing** (1 mg/kg/dose).

The Elderly

- Statin Myopathy is the leading cause of proximal myopathy; trial 3-month cessation.
- Review polypharmacy (fluoroquinolones, aminoglycosides, colchicine) ⚠
- Beware diagnostic traps: late-onset MG mistaken for stroke, and IBM misdiagnosed as PM ⚠

Contextual Care: Special Populations (II)

Renal Impairment

Uraemic myopathy is common in CKD 4-5. Methotrexate is **contraindicated** if eGFR <30. Reduce Azathioprine dose by 25-50%.
IVIg sucrose-formulations are **nephrotoxic** (prefer **glycine-stabilised**).

Hepatic Impairment

Diagnostic Trap: CK and AST/ALT both elevate in primary muscle disease. Do not attribute elevated transaminases to liver disease without checking CK and GGT simultaneously. **Avoid Methotrexate** in Child-Pugh B/C.

Immunocompromised

Critical Illness Polyneuropathy occurs post-ICU/sepsis, presenting with **flaccid weakness/areflexia**. Differentiate from GBS via NCS/EMG. **Watch for opportunistic** CMV/VZV polyradiculopathy.

Aboriginal & Torres Strait Islander Health Considerations

Clinical Burdens

- **Type 2 Diabetes** rates are 3-4x higher; ⚠ diabetic peripheral neuropathy is the primary cause of distal weakness. Routine monofilament screening at ACCHS is vital.
- Note high rates of **Vitamin D deficiency** contributing to proximal myopathy. ⚠

Geographic & Diagnostic Barriers

- **Remote locations** cause significant diagnostic delays.
- **Telehealth** and **Royal Flying Doctor Service** (RFDS) outreach are critical for accessing specialist NCS, EMG, and muscle biopsy services.

Culturally Safe Care

- Engage Aboriginal Health Workers and Practitioners (AHWPs).
- Communicate diagnoses clearly using visual aids.
- Acknowledge and respect the role of community and extended family in medical decision-making and genetic counselling.

PBS & Medication Access

- Actively utilize the **'Closing the Gap' (CTG) PBS Co-Payment Programme** to ensure equitable financial access to essential therapeutics, including immunosuppressants, pyridostigmine, and corticosteroids.

Synthesis: The Primary Care Diagnostic Algorithm