How to recognise Motor Neurone Disease

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Outline

- What is MND
- Clinical symptoms and signs of MND
- Established variants of MND
- Epidemiology
- What should you be looking for and red flags
- What should put you off a diagnosis of MND
- Cognition in MND
- Conditions that can mimic MND

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www.goZeroG.com

What is Motor Neurone Disease (MND)

- Progressive degeneration of motor neurones +/- FTD
- No sensory involvement

- Commonly termed Amyotrophic Lateral Sclerosis (Mixed UMN and LMN)
- Progressive Muscular Atrophy (LMN)
- Primary Lateral Sclerosis (UMN)

- Predominant limb (about two thirds of patients)
- Predominant bulbar (about one third of patients)
- Respiratory rarely
- A combination of these

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What does it present with?

- Highly variable
- Body region of onset and speed and progression unpredictable
- Not all patients will have all symptoms

- Often isolated unexplained symptoms
  - Focal wasting or weakness in a limb (painless)
  - Dysphagia
  - Dysarthria
  - Breathlessness (more often later in disease progression, rarely at onset)

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Variants

	Survival
• ALS most common (almost 90%)	
• UMN and LMN with limb wasting and spasticity	
• Bulbar usually affected to varying degree	2 – 5 years
• Progressive Bulbar Palsy	
• Small proportion of patients with only bulbar involvement early	
• Rapid progression of speech and swallowing difficulty	6 months – 4 years
• Progresses to affect limbs usually within months	
• PMA (5-10%)	> 4 years but can be
• LMN only, limb onset wasting and weakness, “flail arm”	10 years +
• PLS (2%)	
• UMN only, spastic paraparesis, balance problems	10 years +

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How common is it?

- Incidence 1-2 per 100,000 per year
- Prevalence 7 per 100,000 of UK population

- That's just over 4500 current patients living in UK with MND

- Can affect adults of any age but incidence highest age 55-79 yrs
- Slightly more common in men (ratio 3:2)

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Specifics, what should you be looking for?

- Focal Muscle Wasting
  - Asymmetry, First dorsal interossei, tibialis anterior,
  - Fasciculations, cramps, falls, foot drops
- Spastic dysarthria
  - Slow moving tongue, difficult articulation/slurring, quiet speech
  - TONGUE FASCICULATIONS
- Dysphagia
  - Excessive salivation, choking sensations
- Breathlessness
  - Exertional, orthopnoea, daytime somnolence, morning headaches, weak cough/sniff
- Cognitive changes
  - Unusual behaviour, apathy, poor motivation, Emotional lability, difficulty
  - with complex tasks, FTD

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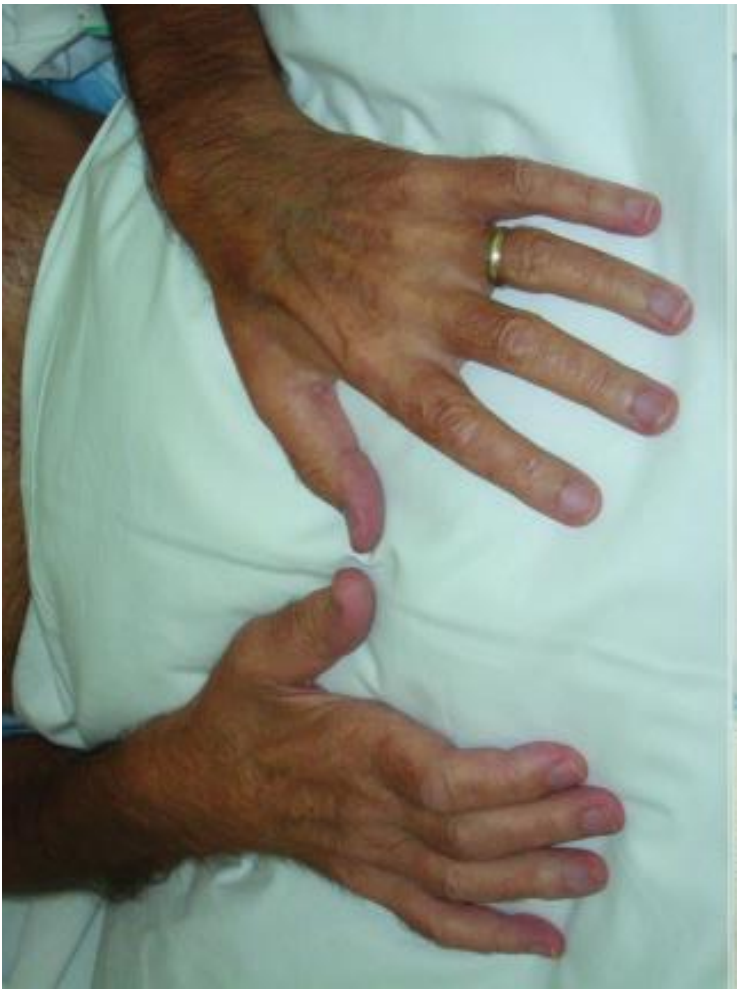
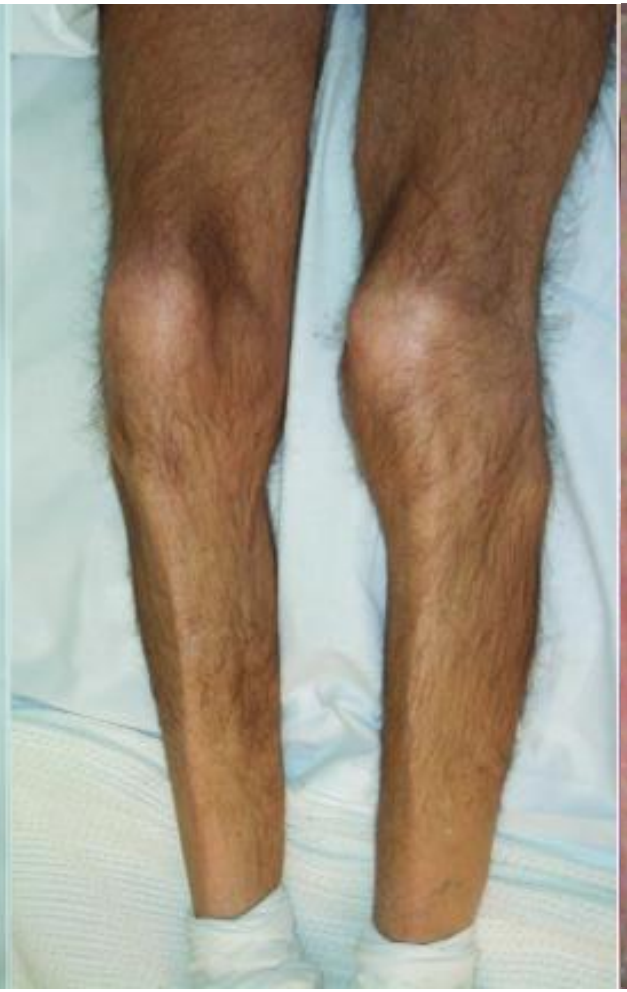
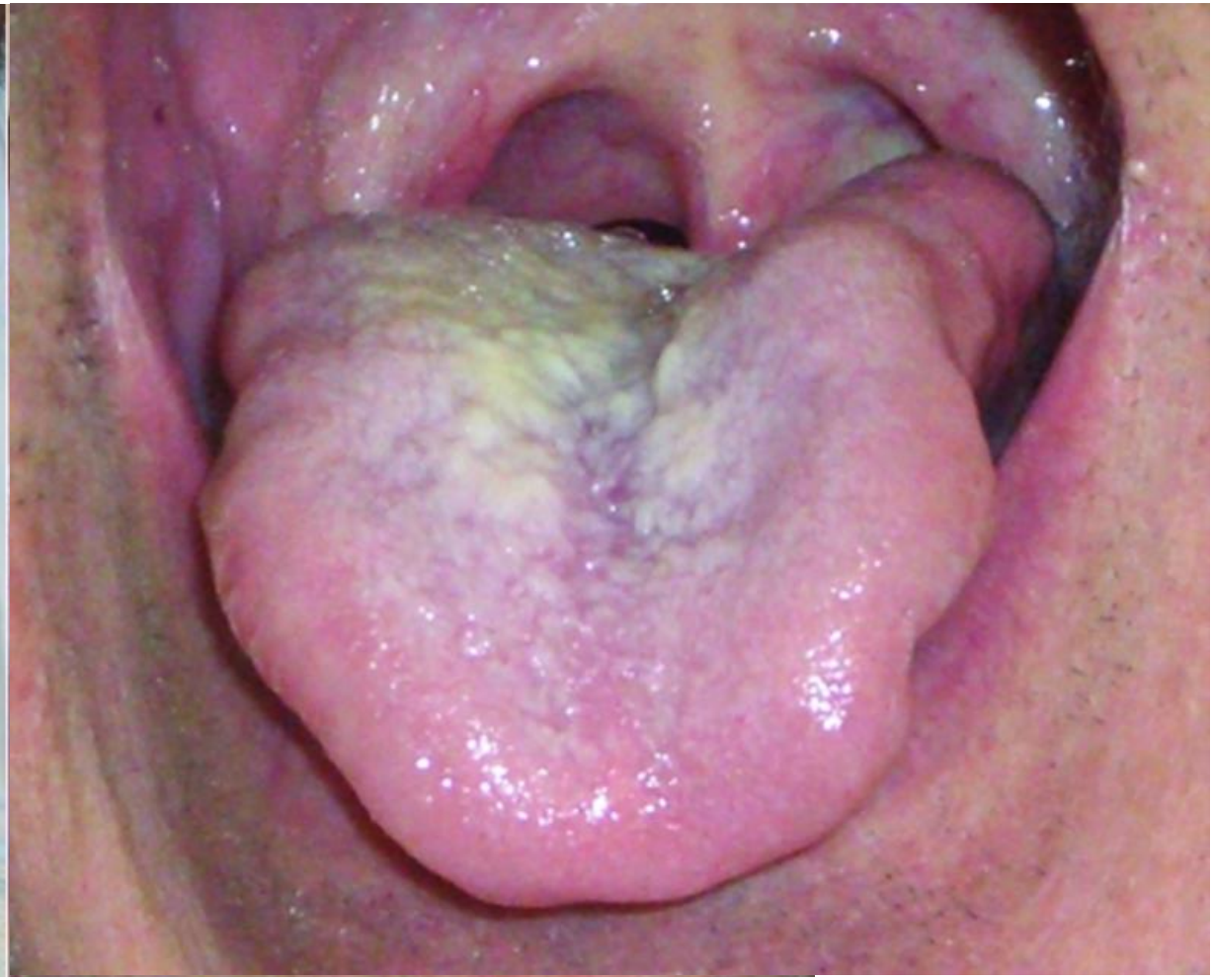

What should put you off a dx of MND?

- Sensory symptoms (paraesthesia, numbness)
  - Pain can be present once atrophy has occurred or with spasticity, cramps
- Bladder and/or bowel involvement (may be involved in late stages)
- Lack of progression or improvement
- Diplopia and or ptosis (much more suggestive of neuromuscular junction disorders eg. Myasthenia Gravis)

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Cognition

- MND patients may experience cognitive changes, ranging from mild effects to much more noticeable and significant impairments.

- Some will develop frontotemporal dementia (MND-FTD).

- Rather than presenting with MND initially some patients will present with behavioural changes, emotional lability (not related to dementia) or FTD, and then go on to display symptoms of MND.

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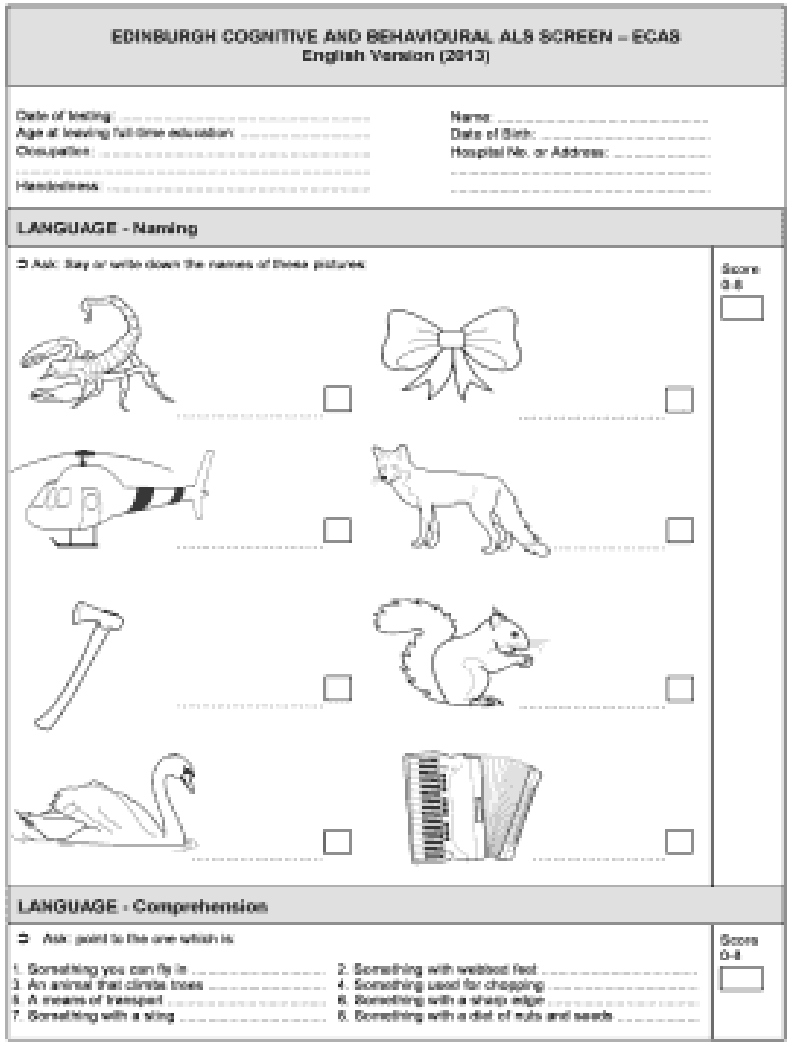
Edinburgh Cognitive and Behavioural ALS Screen

- ECAS is a multidomain assessment originally designed for people with MND but is also useful in other neurodegenerative disorders.
- It makes cognitive assessment fast and accessible and can be undertaken by a health care professional in the clinic or in a patient's home.
- https://ecas.psy.ed.ac.uk/



EDINBURGH COGNITIVE AND BEHAVIOURAL ALS SCREEN – ECAS
English Version (2013)

Date of testing: ..... Name: .....
Age at leaving full-time education: ..... Date of Birth: .....
Occupation: ..... Hospital No. or Address: .....
....
Handedness: ....

LANGUAGE - Naming

⊃ Ask: Say or write down the names of these pictures

Score 0-8

LANGUAGE - Comprehension

⊃ Ask: point to the one which is

1. Something you can fly in ..... 2. Something with webbed feet .....
3. An animal that climbs trees ..... 4. Something used for chopping .....
5. A means of transport ..... 6. Something with a sharp edge .....
7. Something with a sling ..... 8. Something with a diet of nuts and seeds .....

Score 0-8

Genetics

- Inherited/familial MND only accounts for 5-10% of all cases
- Genetic testing can be undertaken in those with a family history but requires appropriate genetic counselling prior to testing
  - 45% of inherited cases caused by C9orf72 gene
  - Less than 20% caused by SOD1
  - TDP43 and FUS cause 5%

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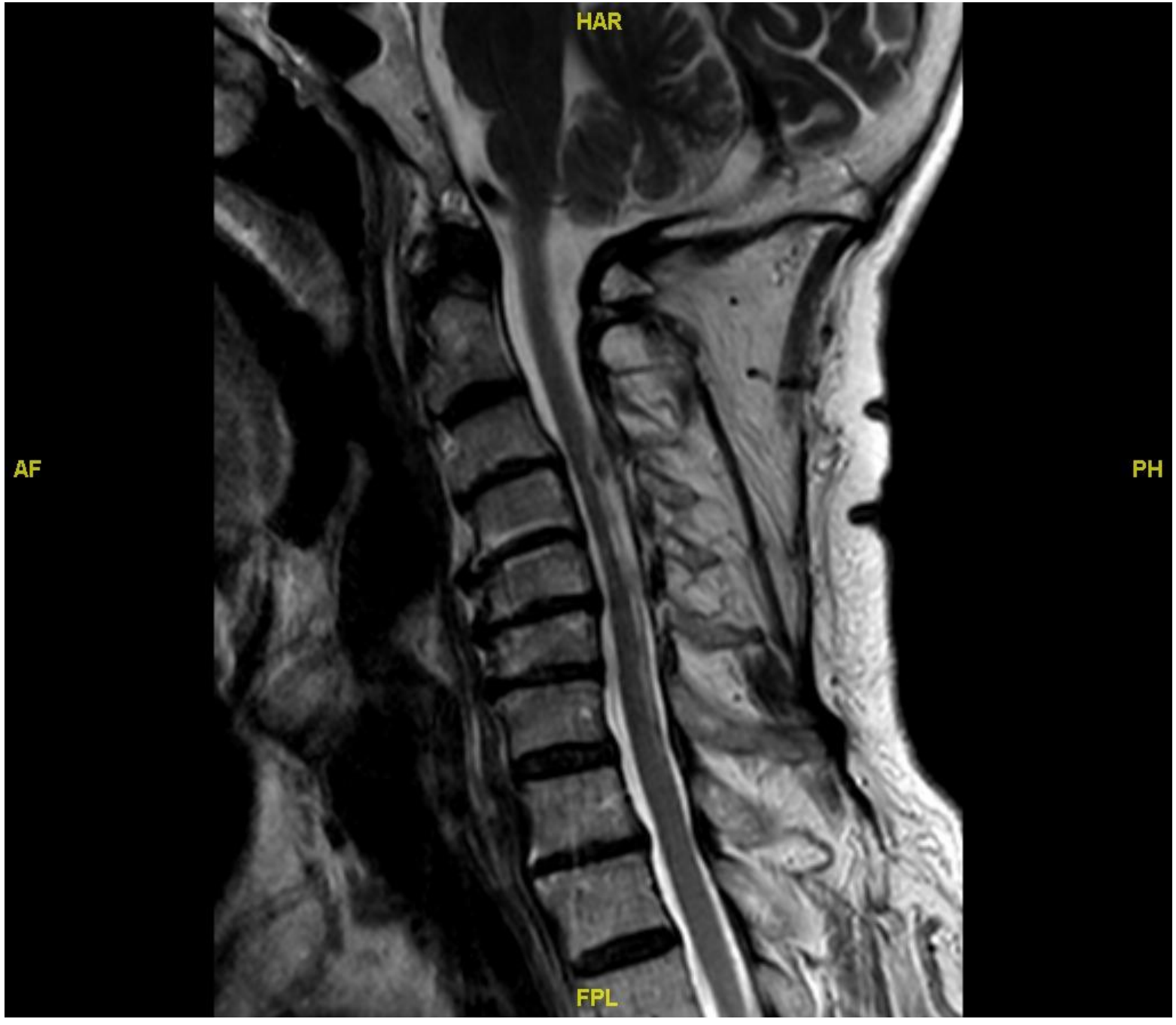
Conditions that can mimic MND

- Cramp fasciculation syndrome
  - Particularly calves, after exercise, no weakness or wasting
- Multifocal motor neuropathy with conduction block (MMN)
  - Typically younger age, distal upper limb weakness with minimal wasting
- Cervical myelopathy with polyradiculopathy/neuropathy
  - Degenerative OA in cervical spine, sensory symptoms, pain
- Inclusion body myositis
  - Distal upper limb weakness, proximal lower limb weakness particularly hip flexors, dysphagia


HAR
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Summary points

- Relatively rare neurodegenerative disease (Incidence 1-2 per 100,000)
- Typically seen in sixth to seventh decades of life
- Genetic cases are very rare (5-10%) and can present earlier in life
- Onset is insidious and can be predominantly limb, bulbar or rarely respiratory in its site of origin
- Red flags – painless focal wasting (1DI/TA), dysphagia, dysarthria, unexplained breathlessness
- Cognitive symptoms and FTD can be a presenting feature

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Thank you

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