Navigating through KD's Symptoms

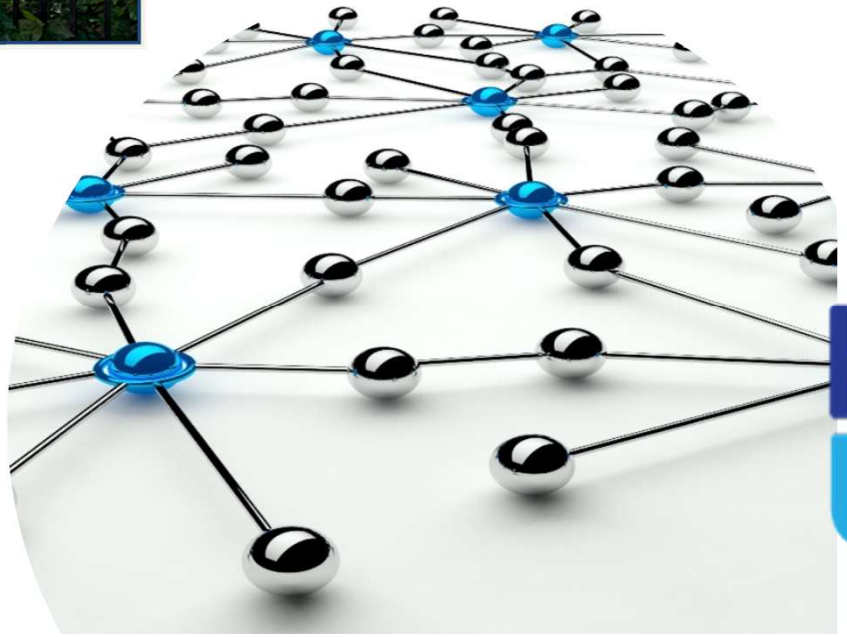
Differences and Similarities between KD and MND

Kennedy's Disease Conference and Patient Day
Sunday 10th July 2022 – The Army & Navy Club – London - UK

UCL

Kennedy's Disease
ASSOCIATION

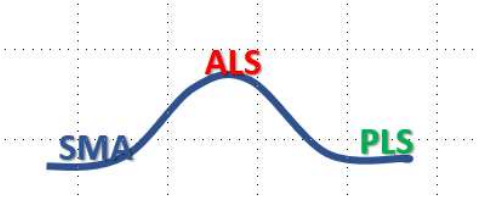
KDRA
Kennedy's Disease
Raising Awareness



mnda
motor neurone disease
association

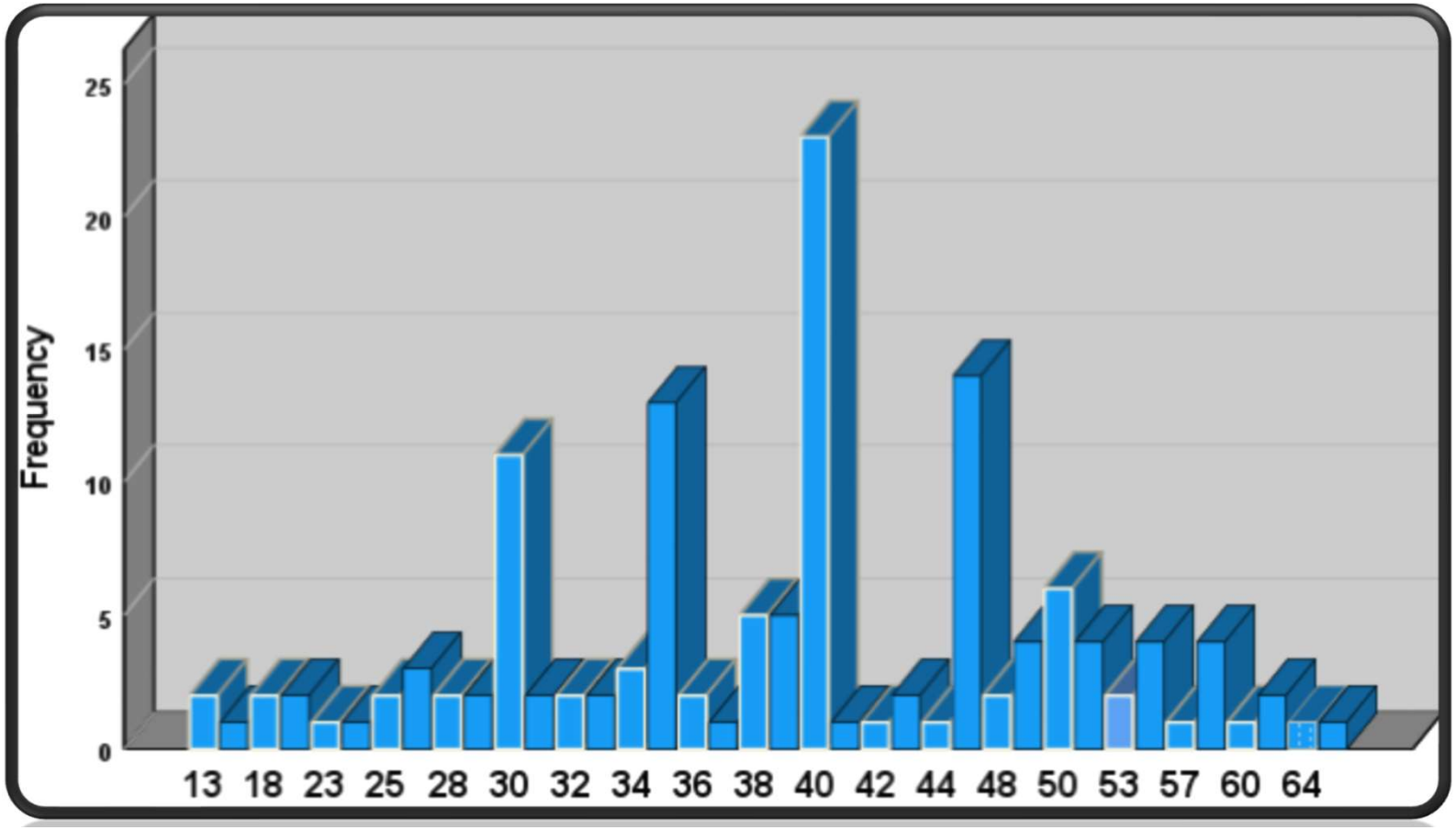
KD
UK

Kennedy's disease – Clinical features

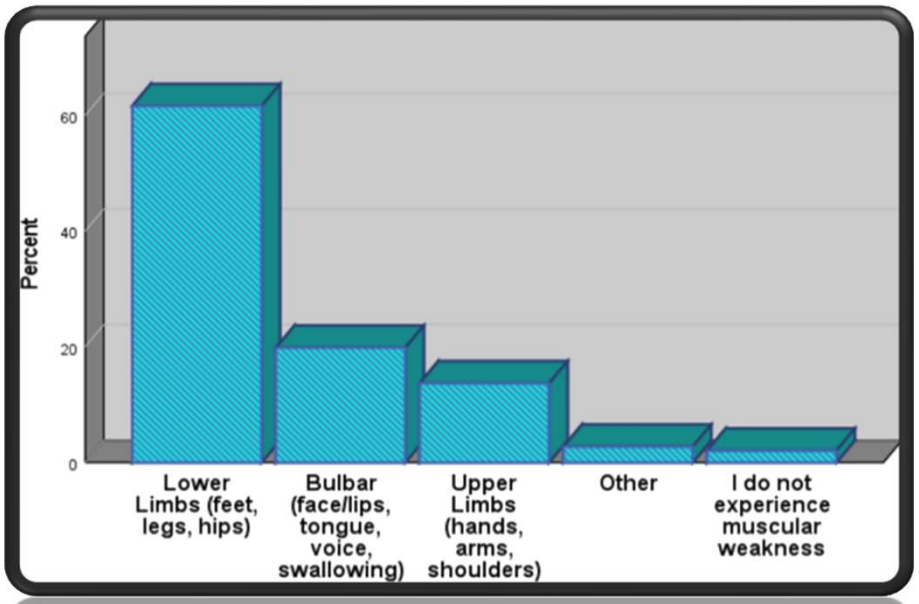
- KD is a type of motor neuron disease – Slowly Progressive Motor Neuropathy
- LMN 

UMN
- Diagnostic Odyssea: Can be Misdiagnosed as ALS
- Age of onset depends on what symptom is considered (Finsterer et al, 2015)
- Weakness and fatigability are the symptoms that most commonly bring patients to medical care (Fratta et al, 2014)

Age of Onset Muscle weakness (Survey 2020)



Onset – and Progression (Survey 2020)

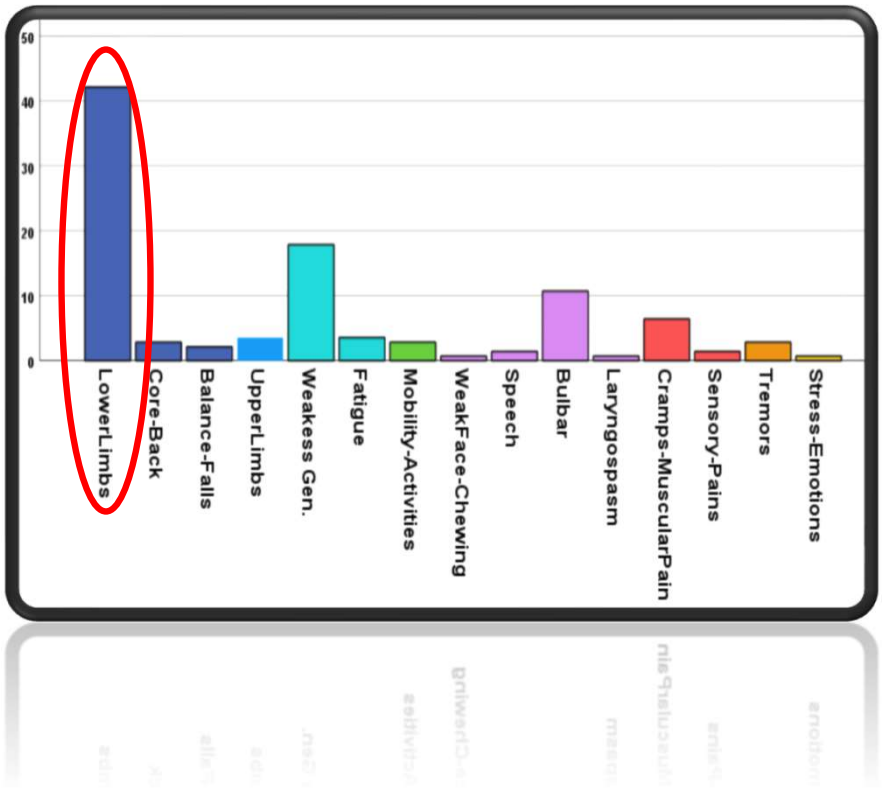


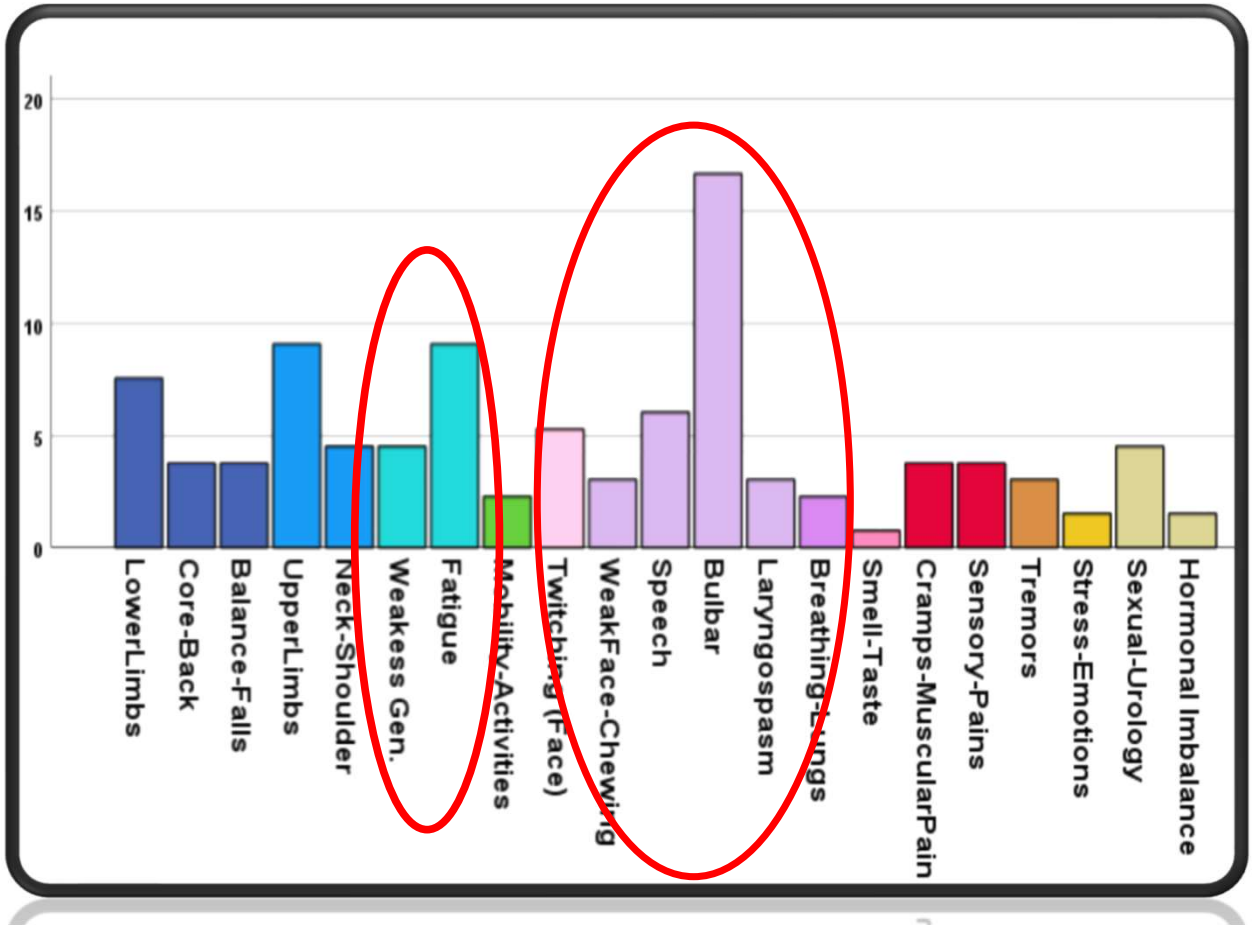
Lower Limbs Bulbar Upper Limbs

Mixed Presentation

SLOW PROGRESSING DISEASE

Main Symptom/problem v Third most important problems (Survey 2020)





LOWER LIMBS: THE MRI STUDY

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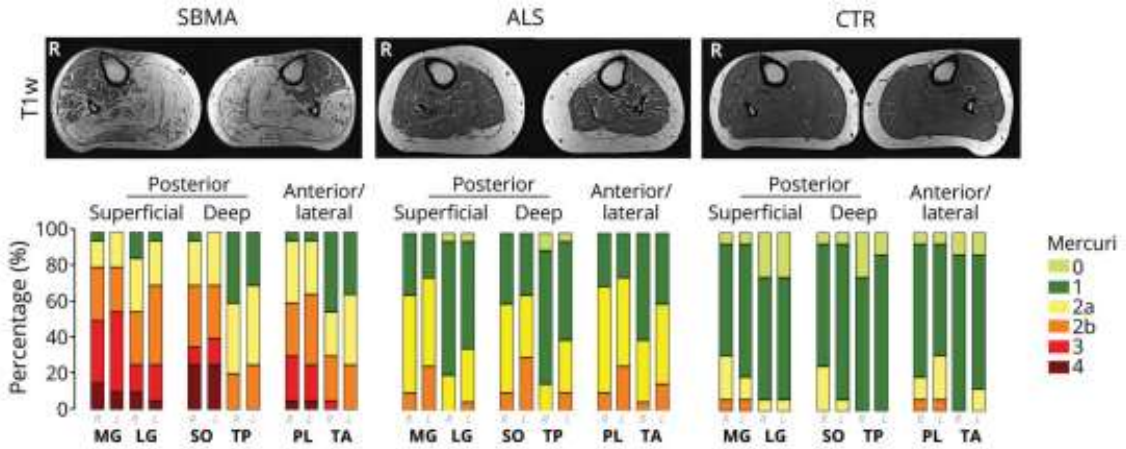
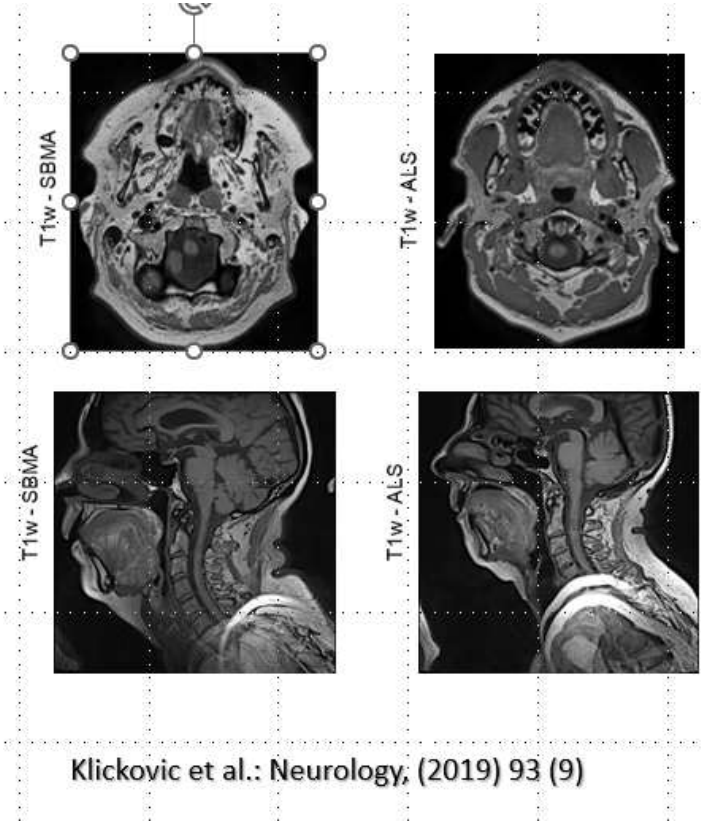
ARTICLE OPEN ACCESS

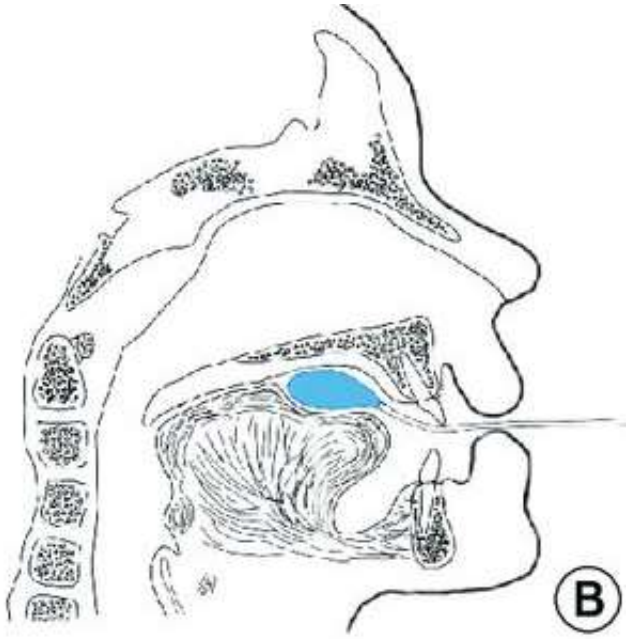
Skeletal muscle MRI differentiates SBMA and ALS and correlates with disease severity



MEASURIGN BULBAR FUNCTION



Klickovic et al.: Neurology, (2019) 93 (9)



BULBAR: KD v ALS

	KD	ALS
Fasciculations	On the tongue quasi constant, lips, chin, perioral area	Frequent on the tongue
Tongue atrophy	Reshaped aspect	Present
Dysarthria and dysphagia	Moderate contrasting with the severity of tongue involvement. Slow progression.	Frequent use of assistive devices for communication and enteral feeding
Respiratory Insufficiency	Rare	Common

Pradat et al.; Orphanet Journ of rare diseases (2020); 15(90)

Pradat *et al. Orphanet Journal of Rare Diseases* (2020) 15:90
https://doi.org/10.1186/s13023-020-01366-z

Orphanet Journal of Rare Diseases

RESEARCH Open Access

The French national protocol for Kennedy's disease (SBMA): consensus diagnostic and management recommendations

Pierre-François Pradat[1*], Emilien Bernard[2], Philippe Corcia[3], Philippe Couratier[4], Christel Jublanc[5], Giorgia Querin[1], Capucine Morélot Panzini[6,7], François Salachas[8], Christophe Vial[9], Karim Wahbi[10], Peter Bede[1,11], Claude Desnuelle[12] and on behalf of the French Kennedy's Disease Writing Group

FATIGUE SEVERITY SCALE

Modified Fatigue Severity Scale

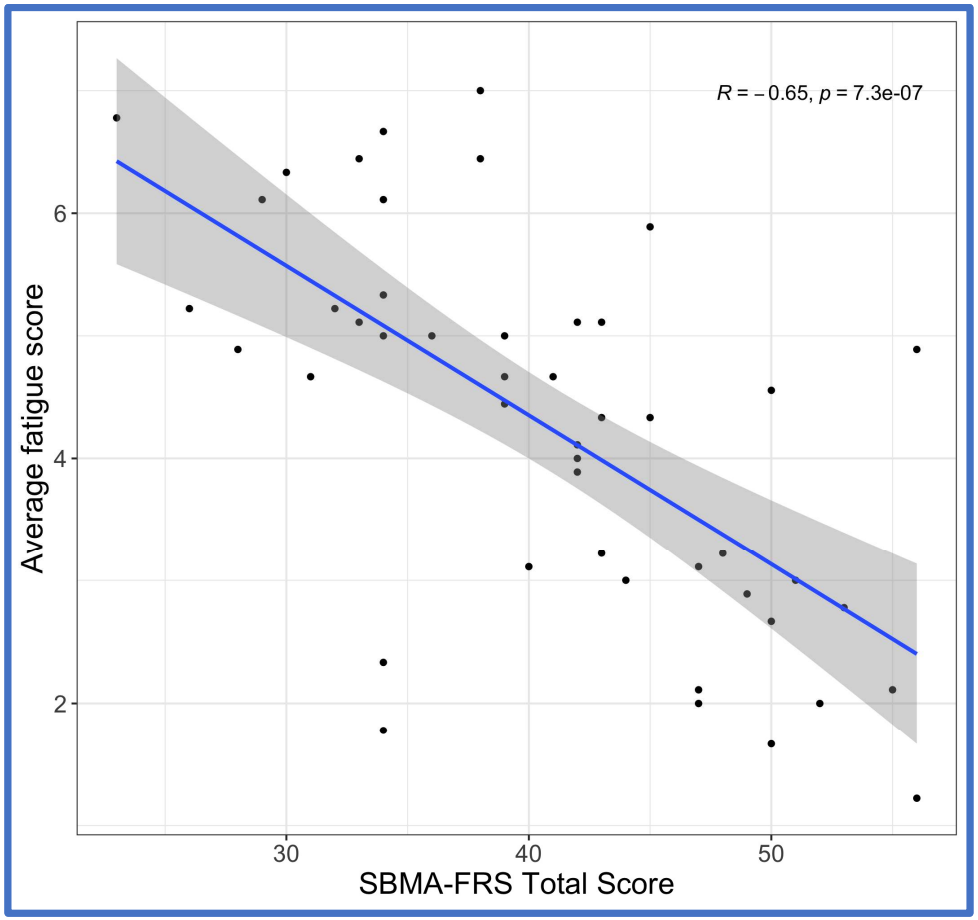
Please read each statement and circle a number from 1 to 7, depending on how appropriate you feel the statement applies to you **over the last week**. A low value indicates that the statement is not very appropriate whereas a high value indicates agreement.

During the past week, I have found that …	Never	Hardly any time	Some of the time	About half of the time	A lot of the time	Nearly all of the time	All of the time	Average
1. My motivation is lower when I am fatigued	1	2	3	4	5	6	7	3.85%
2. Exercise brings on my fatigue	1	2	3	4	5	6	7	4.95%
3. I am easily fatigued	1	2	3	4	5	6	7	4.44%
4. Fatigue interferes with my physical functioning	1	2	3	4	5	6	7	4.53%
5. Fatigue causes frequent problems for me	1	2	3	4	5	6	7	4.00%
6. My fatigue prevents sustained physical functioning	1	2	3	4	5	6	7	4.36%
7. Fatigue interferes with carrying out certain duties and responsibilities	1	2	3	4	5	6	7	3.89%
8. Fatigue is among my three most disabling symptoms	1	2	3	4	5	6	7	4.36%
9. Fatigue interferes with my work, family or social life	1	2	3	4	5	6	7	3.80%

- 63% of participants (31 in 49) experience an average score in FFS above 4
- Item 2: Exercise – scored highest
- Items 1,7, and 9: Motivation, Duties and Responsibility and work

FATIGUE: Correlates with loss of functionality

SBMA-FRS	SCORES
1. SPEECH	0-4
2. CONTROL OF SALIVATION	0-4
3. SWALLOWING	0-4
4. TONGUE	0-4
5. PUFFING CHEECKS	0-4
6. WRITING	0-4
7. EATING ACTION	0-4
8. DRESSING ACTIVITY	0-4
9. RISING FROM SITTING	0-4
10. ARISING FROM SUPINE	0-4
11. BOWING	0-4
12. WALKING	0-4
13. STAIRS	0-4
14. BREATHING	0-4
TOTAL MAXIMUM SCORE	56



TYPE	INTERVENTION	COMMENTS
PHARMACOLOGICAL	Upcoming Disease modifying drugs	Severity of fatigue as a secondary outcome measure in trials? β agonist?
	Trials of symptomatic treatments	Participants similar pathologies / issues?
NUTRACEUTICAL	Q10 enzyme, L Carnitine, Arginine	Anecdotal evidence. More evidence required
EXERCISE CVS Fitness	Breathing techniques, Yoga, Pilates, other	Can improve fitness. May be useful in managing symptoms
	Proprioceptive/balance training	Reducing abnormal movement compensation? – NB Avoid falls
	High intensity	Reduces time exposure to fatiguing exercise
	Low intensity	Light, graded, or minimal exercise – Hydrotherapy.
NUTRITION	Pre/post-exercise	Safe protein intake
	Healthy Diet	Sugar intake, use of stimulants, balance of nutrients
	Softer diet	Reduces masticatory effort
BEHAVIOURAL INTERVENTIONS	Planning, pacing, “boom and bust”, balance rest, work, leisure	Budgeting energy reserve / muscle strength – Use of activity diary
	Sleep hygiene	Identifying, addressing sleep difficulties
	Assistive/digital technologies, mobility aids	Reduce fatigue inducing tasks
STRESS REDUCTION	Relaxation, talking therapies, CBT, ACT, family, friends, patient associations	Stress can have a detrimental effect on fatigue Fatigue can be detrimental to self-esteem

CONCLUSIONS

- IMPACT OF SYMPTOMS AFFECTS QUALITY OF LIFE
- ETHEROGENEOUS CONDITION
  - KD – AGEING – CONCOMITANT PATHOLOGIES – EXTERNAL FACTORS
- SLOWLY PROGRESSIVE DISEASE
- CAN WE SLOW PROGRESSION WITHOUT DISEASE MODIFYING DRUGS?
  - MANAGING KD: MAINTAIN FUNCTIONS / ? SLOW PROGRESSION
  - MANAGING ALS: SUPPORT FUNCTIONS
- FATIGUE: MUSCULAR FATIGUE – IMPACTS ON MOTIVATION
- BULBAR INVOLVEMENT IN KD V ALS
- NB: RISK OF PNEUMONIA
  - NEED FOR DIETICIAN – SPEECH and LANGUAGE THERAPIST INPUT @ FIRST INDICATIONS OF BULBAR INVOLVEMENT
  - ADVANCED DECISIONS

MANY THANKS TO

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