KEY INFORMATION FOR EMERGENCY MANAGEMENT OF: KENNEDY'S DISEASE – SBMA

BACKGROUND

Kennedy's disease (KD) or spinal and bulbar muscular atrophy (SBMA), is a rare, **adult onset, X-linked neuromuscular disease** caused by a CAG repeat mutation in the Androgen Receptor gene.

It is characterised by **fasciculation, cramps, and muscle weakness, predominantly affecting the limbs, face and bulbar area**. These symptoms **may be accompanied by systemic effects** related to androgen insensitivity. Symptoms typically present between the ages of 30 and 50 and it predominantly affects males. Although disabling, 90 % of people with KD have a normal lifespan. Treatment is symptomatic and supportive.

AIRWAY

- Muscle weakness can affect the jaw, face, mouth, tongue, soft palate and larynx leading to **difficulties in chewing**, **dysphagia** and **mild dysarthria** characterised by nasal speech.
- Mild episodes of **choking episodes are common due to bulbar involvement**. Rarely, these can be severe and life threatening.
- Patients can experience **laryngospasm**, often accompanied by stridor. This can be triggered and exacerbated by gastro-oesophageal reflux and upper airway infections. Although these events can be frightening and distressing, they seldom escalate to prolonged episodes. **Techniques such as straw breathing can be helpful** to manage these events. Pharmaceutical interventions may be indicated in severe cases and recurrent episodes.
- Particular attention is warranted during **endotracheal extubation** due to the **risk of laryngospasm**. Severe postoperative glottic oedema has also been reported.

BREATHING

- **Minor breathing difficulties have been reported in KD**, but non-invasive ventilation is rarely needed (0.5% of patients in a recent survey). Breathing muscles are typically spared from weakness.
- Dysphagia can lead to recurrent **aspiration pneumonias**. Pneumonia is identified as the most frequent cause of premature death (10% of patients).
- Pulmonary function testing may be advised pre-operatively.
- **The action of depolarising blocking agents, such as Succinylcholine, can be prolonged or enhanced in KD patients**. When using neuromuscular blocking drugs, it is necessary to observe the degree of relaxation using a muscle relaxation monitor.

CIRCULATION

- In rare cases (1-2% but possibly higher in patient of Asian descent), KD patients can develop **Brugada-like Syndrome**. **ECG atrial high lead placement** is useful in detecting Brugada syndrome. If detected, specific antiarrhythmic, psychotropic, anaesthetics and analgesic drugs should be avoided (a list can be found here: https://www.brugadadrugs.org/avoid/

- KD patients can present with **raised Troponin T and CK unrelated to cardiac damage. Troponin I testing should be preferred or used alongside Troponin T** when evaluating potential cardiac damage.
- Subclinical autonomic dysfunction has also been reported in KD patients.

DISABILITY

- KD affects predominantly the lower motor neurones and skeletal muscle. Deep tendon **reflexes will be diminished**.
- Patients may present with sensory disturbances **in the distal lower limbs**.
- **Balance and mobility issues are common**, so early assessment by physiotherapy and occupational therapy (PT/OT) is recommended to mitigate fall risks. Early mobilisation and sitting out of bed should be encouraged **as soon as medically appropriate** to reduce deconditioning and minimise care needs at discharge.
- **Cognitive and neuropsychological function is normal in KD.**

EXPOSURE

- High prevalence of insulin resistance, metabolic syndrome and non-alcoholic liver disease has been reported in KD.
- Statins are not usually prescribed due to the **potential for statin induced myopathy.**
- Elevated aspartate aminotransferase, alanine aminotransferase and lactate. dehydrogenase along with low creatinine are indicative of muscle pathology.
- **Osteopenia** is common and people with KD can be more susceptible to fractures following falls.

SOURCES

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- Practical Guidance for Patients : Living with KD : Kennedy's Disease Association (kennedysdisease.org)

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