



## Mrs M

- 74 year old female
- Previously well, independent, caregiver for elderly husband
- 5 month history of;
  - Rash around eyes, dorsum of arms, thighs, back of hands, anterior chest.
    - Itchy. Has been putting topical steroid on it.
  - Severe muscle weakness – worse proximally.
    - Can't get hands to hair
    - Can't stand from sitting
    - No numbness or tingling



## Further symptoms

- 3-4 months of progressive breathlessness.
  - Can now walk only 10m
  - Minimal cough
- Sore throat, multiple mouth ulcers, nasal crusting, occasional epistaxis and nasal congestion
- New significant hearing loss
- Saw ENT -nasoendoscopy normal but middle ear effusions bilaterally (grommets planned)
- Significant dysphagia
  - Struggle to swallow solids
- New severe constipation – no PR blood/melena
- Weight loss of 10kg (but eating much less due to swallowing difficulties)

## On examination

- Dramatic erythematous rash on dorsum of arms, thighs
- Heliotrope rash around eyes
- Periorbital oedema
- Gottron's papules hands
- There is no synovitis
- Chest clear to auscultation
- Severe muscle weakness globally but worse in proximal muscles
  - Neck flexion weakness
  - 2/5 hip flexion
  - Significant quadriceps and biceps muscle wasting present

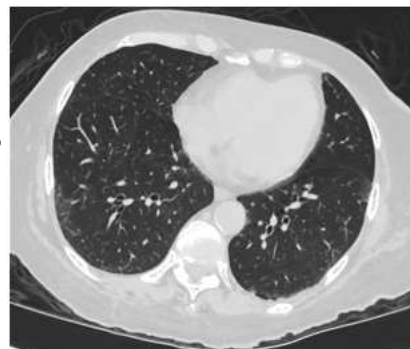


## Investigations

- CK normal (46)
- CRP 16
- ANA 1:1260 –basic ENA screen negative,
- ANCA negative
- Creatinine is low (was 60, now 30)
- Abdo CT with contrast
  - Small nodule associated with the body of the left adrenal is most likely an adrenal adenoma.

## HRCT Chest

- Multiple solid, spiculated, pulmonary nodules bilaterally, measuring up to 9mm in diameter.
- No evidence of interstitial lung disease.




### MRI Thighs

- MRI appearances confirm a diffuse proximal myositis surrounding pelvis and involving the thigh musculature with diffuse fatty atrophy.




### SAE1 antibody

• HMGR Abs	Negative
• <b>Ro-52 Ab (Myositis Blot)</b>	<b>Positive</b>
• OJ Ab (Myositis Blot)	Negative
• EJ Ab (Myositis Blot)	Negative
• PL-12 Ab (Myositis Blot)	Negative
• PL-7 Ab (Myositis Blot)	Negative
• SRP Ab (Myositis Blot)	Negative
• Jo-1 Ab (Myositis Blot)	Negative
• PM-Scl75 Ab (Myositis Blot)	Negative
• PM-Scl100 Ab (Myositis Blot)	Negative
• Ku Ab (Myositis Blot)	Negative
• <b>SAE1 Ab (Myositis blot)</b>	<b>Positive</b>
• NXP2 Ab (Myositis blot)	Negative
• MDA5 Ab (Myositis blot)	Negative
• TIF1 gamma Ab (Myositis blot)	Negative
• Mi-2 beta Ab (Myositis blot)	Negative
• Mi-2 alpha Ab (Myositis blot)	Negative



### SAE-1 antibodies

- Anti-small ubiquitin-like modifier-1 (SUMO-1) activating enzyme (SAE)
  - novel DM-specific autoantibody first reported by Betteridge et al. in 2007
- Myositis specific antibody
- Found in 3-8 % of adult DM patients
- Presenting with
  - Severe skin disease –often cutaneous amyopathic dermatomyositis
  - Dysphagia
  - Minimal lung involvement
  - Prognosis is favorable
- Strongly associated with the HLA-DQB1\*03, HLA-DRB1\*04, and HLA-DQA1\*03 haplotypes



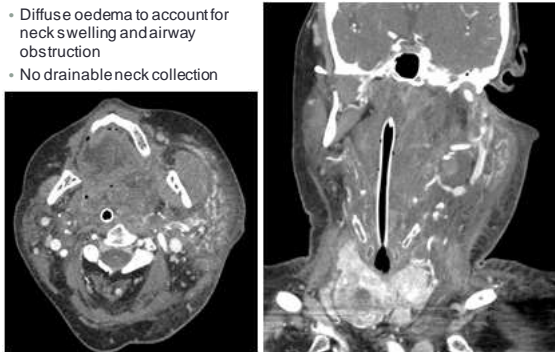
SUMO-1 is involved in post translational modification affecting a wide variety of cellular processes

### Long Holiday Weekend

- Started 1g IV methylpred (planned 3 days IV then oral) and hydroxychloroquine Friday
- Early hours of the morning Sunday
  - Worsening difficulty swallowing and pain in neck
  - A few hours later –struggling to breathe, stridorous
  - New, visible cheek and left neck swelling
  - Taken to theatre for awake fiberoptic intubation
  - Transferred to ICU –noted to have worsening extrinsic compression of ET tube
  - Also complication of known large goitre which would make emergency trachy hard
  - Returned to theatre for tracheostomy insertion

### CT Neck

- Diffuse oedema to account for neck swelling and airway obstruction
- No drainable neck collection



### Higo R et al. Laryngeal edema in a patient with dermatomyositis. Otolaryngology Case Reports. Published online Feb 2017

- 61 year old man initially presented with cervical oedema
- Went on to develop classic dermatomyositis rash and myopathy
- Classic myositis antibodies negative



Fig. 2. Laryngeal edema and tumorous lesion of the left aryepiglottic fold.

Fig. 3. Laryngeal endoscopic finding three months after initial therapy. Left aryepiglottic edema markedly improved.