A 43-year-old Caucasian female, with known 46XY chromosome pure gonadal agenesis and systemic hypertension, presented with upper motor neuron weakness of the right face and upper limb. She was also noted to have livedo reticularis and advanced finger clubbing. Left-sided cerebral infarction was confirmed on brain computed tomography (CT) and, following further investigations, a diagnosis of Sneddon's syndrome (SS) was made. Three years later, she went on to develop adenocarcinoma of the right lung, which sadly claimed her life within 4 months. To our knowledge, this is the first reported case of SS in association with finger clubbing and pure gonadal agenesis.

**Key words:** Sneddon's syndrome, pure gonadal agenesis, finger clubbing, adenocarcinoma

**Sneddon's Syndrome**

**Unusual Features and Associations**

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INTRODUCTION

Sneddon's syndrome (SS) is a rare disease of unknown aetiology, characterised by ischaemic cerebrovascular accident and generalised idiopathic livedo reticularis. It is more common in females between the third and fifth decade, with an estimated incidence of four cases per million per year. It has a mortality rate of 9.5% over 6.2 years, likely secondary to recurrent stroke and vascular dementia (1). Systemic hypertension, headaches, venous thrombosis, and valvulopathy have also been mentioned as possible clinical manifestations (1-3).

It has been found that around 59% of cases are positive for antiphospholipid antibody. However, the link between SS and antiphospholipid antibody syndrome remains the subject of much debate. Diagnosis consists of cerebral imaging, laboratory studies, including autoantibody screening, and skin biopsy (2). The mainstay of treatment is with anticoagulant agents (3).

CASE

A 43-year-old Caucasian female presented with sudden onset right facial and upper limb weakness. She had a past medical history of 46XY chromosome pure gonadal agenesis, for which she was on long-term hormone replacement therapy (HRT). She had a history of aortocoronary bypass surgery at 36 years of age and a past cardiac history of atrial fibrillation on warfarin. At the time of her presentation, she was found to have peri-orbital xanthemata, plethoric facies, widespread livedo reticularis, and newly reported finger clubbing (drum stick appearance). Upper motor neuron weakness of the seventh cranial nerve and the right upper limb were also observed. Carotid Doppler revealed left internal carotid occlusion, whilst brain CT scan demonstrated a left-sided cerebral infarct in the parieto-occipital region. Trans-thoracic and later transoesophageal contrast echocardiography showed no evidence of cardiac or pulmonary shunting, whilst pulmonary function tests were consistent with mild obstructive airways disease. Thrombophilia and autoantibody screening included anti-phospholipid antibody and anti-thrombin III, were negative. Skin biopsy of the livedo reticularis showed patchy mild perivascular inflammation in the dermis only and was non-specific. She had hypertension, and was also a smoker. On examination, she was found to have peri-orbital xanthemata, plethoric facies, widespread livedo reticularis, and newly reported finger clubbing (2). The mainstay of treatment is with anticoagulant agents (3).

In concluding, it is our view that this case does present a collection of uncommon disorders and potentially new clinical features affecting the same patient, and is thus worthy of note within the pantheon of case reports of SS.

REFERENCES
Figure 5. Both of inner and outer lumens are associated with the branches

Figure 6. Both of inner and outer lumens are associated with the branches
Lung cancer D-dimer