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# Kearns-Sayre syndrome: presenting with vocal fold palsy

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**Abstract**
We present the first documented case of Kearns-Sayre syndrome presenting with a vocal fold palsy.

**Key words: Kearns Syndrome; Vocal Fold Paralysis**

## Introduction

Kearns-Sayre syndrome is a mitochondrial disorder first described in 1958,[1] that develops in early adult life. The full syndrome is characterized by progressive external ophthalmoplegia, atypical pigmentary degeneration of the retina and heart block. It has been known to present with sensorineural hearing loss.[2] It is sporadic, non-hereditary and cardiac problems may not develop for many years. There have been no documented reports of Kearns-Sayre or oculopharyngeal myopathy to date presenting with a true vocal fold palsy.

## Case report

A 27-year-old male furniture restorer presented to the ENT department with a sudden onset of hoarseness and ptosis of the right upper eye lid. The ptosis was progressive over the following six months and the left eye subsequently became affected. His hoarseness did not improve and a left vocal fold palsy was confirmed by clinical examination and computed tomography scan (CT). There were no other neurological symptoms and the subsequent neurological examination was unremarkable.

Investigations including blood tests, extra-ocular muscle biopsy and lumbar puncture were performed. The CSF analysis was normal and there was no evidence of oligoclonal IgG bands. Urea, electrolytes and glucose were normal but the lactate level was elevated. Fibre-optic nasendoscopy was unremarkable and CT scan did not reveal any pathological cause for the vocal fold palsy. The extra-ocular muscle biopsy and quadriceps biopsy were normal and did not show any classic features of mitochondrial disease. Although the histology did not confirm mitochondrial disease there is often no evidence of abnormal mitochondria on biopsy.

The history and clinical findings combined with his age and the absence of any family history were characteristic of Kearns-Sayre syndrome and this was confirmed by the neurologist involved. A left thyroplasty was performed to improve the quality of voice using a 3 mm prosthesis. The initial result was very good, however, six months following surgery for his ptosis, which involved laryngeal intubation, he developed further hoarseness. A revision thyroplasty was performed, that improved the voice quality, both subjectively and on laryngographic analysis, but it never returned to its pre- operative level (ie. the level after the first thyroplasty). He subsequently developed dysphagia and pooling of saliva. Video fluoroscopy was performed to dynamically assess the swallow. The oral phase was normal but the pharyngeal phase delayed, with trapping of the bolus in the vallecula, consistent with impaired pharyngeal motility. Speech therapy was commenced and the patient has not deteriorated further.

## Discussion

We present a case of early presentation of Kearns-Sayre syndrome to the department of Otolaryngology. There appears to be an overlap between the characteristic symptoms of Kearns-Sayre and oculopharyngeal myopathy in mitochondrial disorders; they are not mutually exclusive, from our interpretation of the literature.[3–6] Although our patient did not appear to have retinitis pigmentosa and cardiac myopathy, his age, clinical presentation and absence of family history suggest that the diagnosis is that of Kearns-Sayre syndrome. There has been one documented case of oculopharyngeal myopathy presenting to the ENT department with sensorineural hearing loss[2] and a further case has described vocal fold bowing in association with mitochondrial myopathy.[7] In view of the symptoms experienced by patients with mitochondrial diseases, it is not inconceivable to expect their presentation to the ENT department, and they should be considered in the differential diagnosis of disorders of the pharynx and larynx.

One would expect that if a mitochondrial disorder is to present in the ENT clinic, it would be for investigation of dysphagia due to involvement of the pharyngeal muscles. First presentation due to other symptoms, such as hoarseness in our case or sensorineural hearing loss as in the other case mentioned above, are extremely rare.

The patient's symptoms are progressive and, as noted in the literature, there may be later development of other associated conditions.[3]

It is important not to overlook the underlying causes of vocal fold paralysis and to remember the rarities. We have been able to alleviate his symptoms with a thyroplasty, however, the subsequent onset of dysphagia and his further ophthalmic surgery has compromised the original result. The sudden loss of his voice following surgery to correct his ptosis was attributed to the intubation during the general anaesthetic. With respect to his voice, this was

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formally assessed in the voice clinic pre- and post-thyroplasty both quantitatively and qualitatively. There have been documented anaesthetic considerations when treating these cases, and special attention should be given to optimizing oxygenation to prevent further stress to the dysfunctional system of aerobic metabolism. Careful dose titration of anaesthetic agents and optimum monitoring is essential to minimize abnormal responses.[8]

In conclusion, we highlight the importance of investigation of the cause of vocal fold palsy and the role of thyroplasty in improving symptoms in these unusual mitochondrial conditions. Otolaryngologists should be aware of the possibility of presentation of mitochondrial diseases to their clinic.

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