Ophthalmologist Saving Lives

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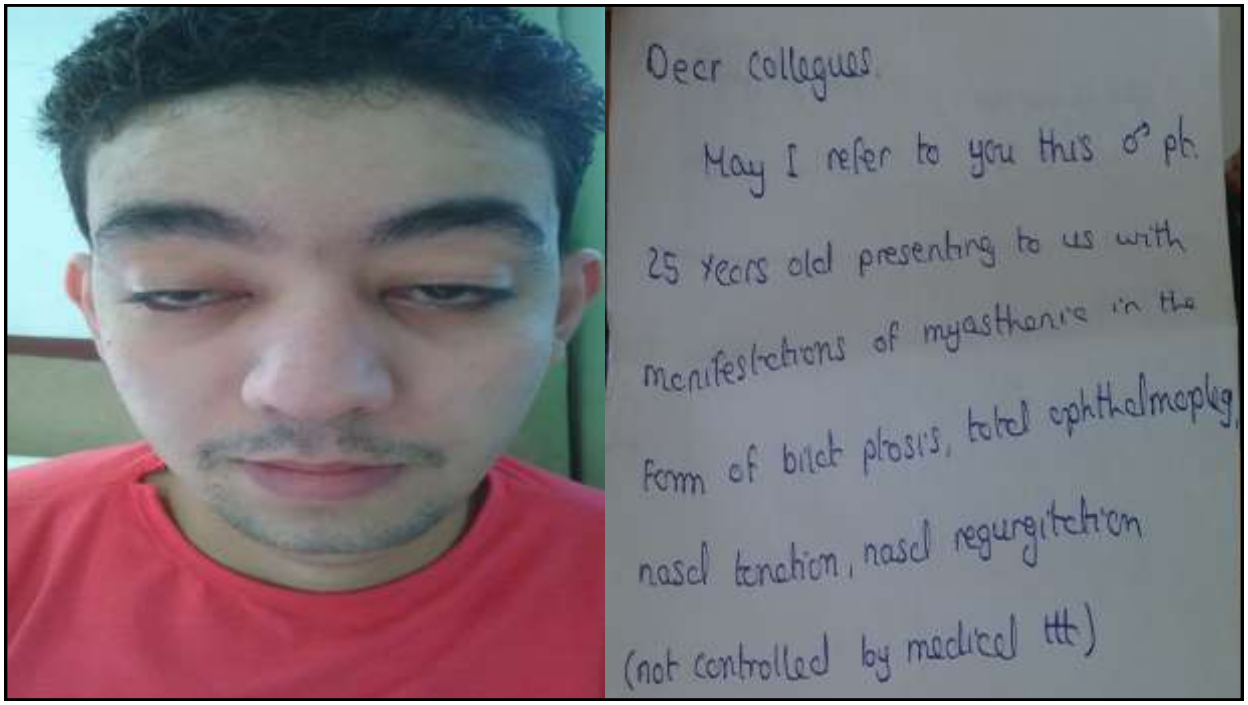
Medical

NLP

means

No Light Perception (Ophthalmic)



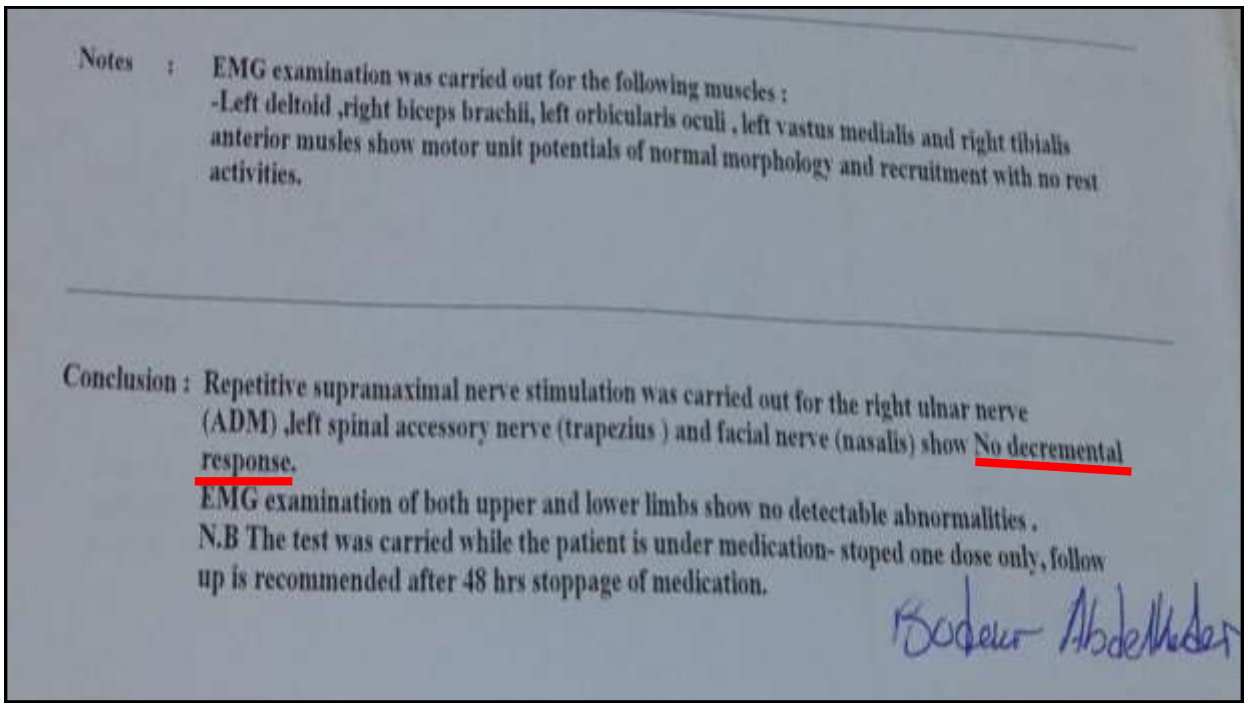
- 25 year-old male.
- H/O: Bil upper lid surgeries for ptosis (age of 15), Total Ophthalmoplegia (age of 20) and diagnosed to have MG.
- Medications: Pystinone 2 tabs/6hr , Solupred 20 mg 4 tabs daily.

Examination

- Severe Ptosis (MRD 1 = -1)
- Total Ophthalmoplegia
- Hearing loss ??
- Respiratory Rate (Normal !!)

Examination

- Asymmetric ptosis & fluctuates (fatiguable).
- Ice test & Cogan's lid twitch sign.
- Edrophonium (Tensilon) test.
- Electromyography.
- CT chest ------- No thymoma.



Notes : EMG examination was carried out for the following muscles :
-Left deltoid ,right biceps brachii, left orbicularis oculi , left vastus medialis and right tibialis anterior musles show motor unit potentials of normal morphology and recruitment with no rest activities.

Conclusion : Repetitive supramaximal nerve stimulation was carried out for the right ulnar nerve (ADM) ,left spinal accessory nerve (trapezius) and facial nerve (nasalis) show No decremental response.
EMG examination of both upper and lower limbs show no detectable abnormalities .
N.B The test was carried while the patient is under medication- stoped one dose only, follow up is recommended after 48 hrs stoppage of medication.

POST NON IONIC CONTRAST MULTISLICE C.T EXAMINATION OF THE CHEST REVEALED

FINDINGS:

- Lung fields and costodiaphragmatic recesses are clear.
- No pulmonary parenchymatous deposits seen.
- No signs of mediastinal lymphadenopathy detected.
- Normal appearance of the heart and its great vessels.
- Sections taken in the upper abdomen are unremarkable.

OPINION:

- Normal CT study of the chest.

Graves' disease??

- Proptosis & other signs.
- Thyroid gland examination.
- T3 , T4 , TSH.
- CT Orbit.

Laboratory Report

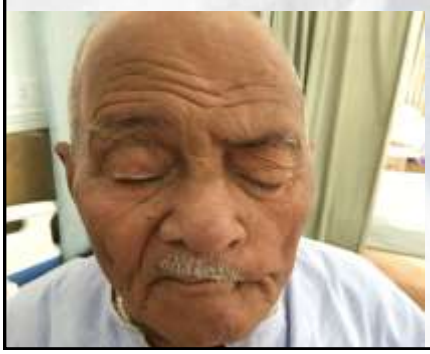
Test	Result	Unit	Reference Range
TSH	1.39	uIU/mL	0.40 - 4.00
Free T3	2.5	pg/mL	2.0 - 4.4
Free T4	1.5	ng/dl	0.9 - 1.8

OPINION:
Unremarkable post contrast CT examination of the orbit

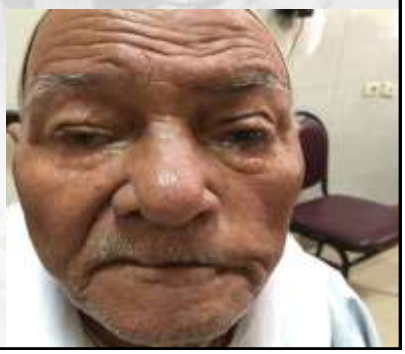
Chronic Progressive External Ophthalmoplegia (CPEO)

Oculopharyngeal muscular dystrophy (OPMD)

- Symptoms after 40.
- Ptosis then dysphagia.
- Proximal muscles weakness.

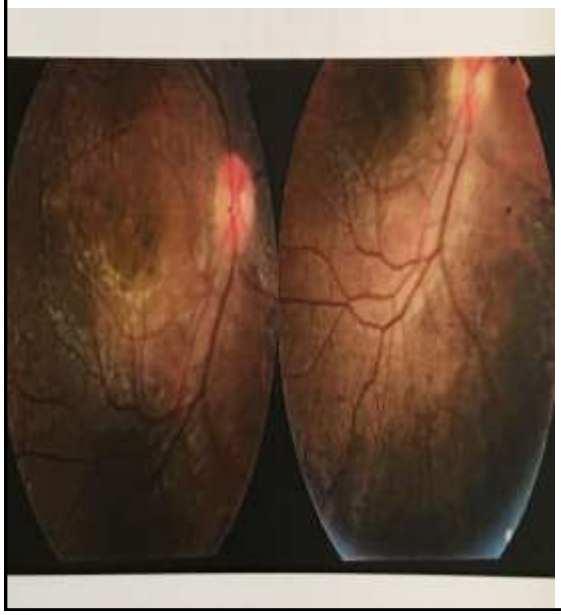


Courtsey Prof Dr Khaled Abo El Enin



Examination

- VA OD 6/18 → 6/9 by plano/-1.25 X 30
- OS 6/12 → 6/9 by plano/-1.0 X 120
- Normal Slitlamp.
- Fundus ??

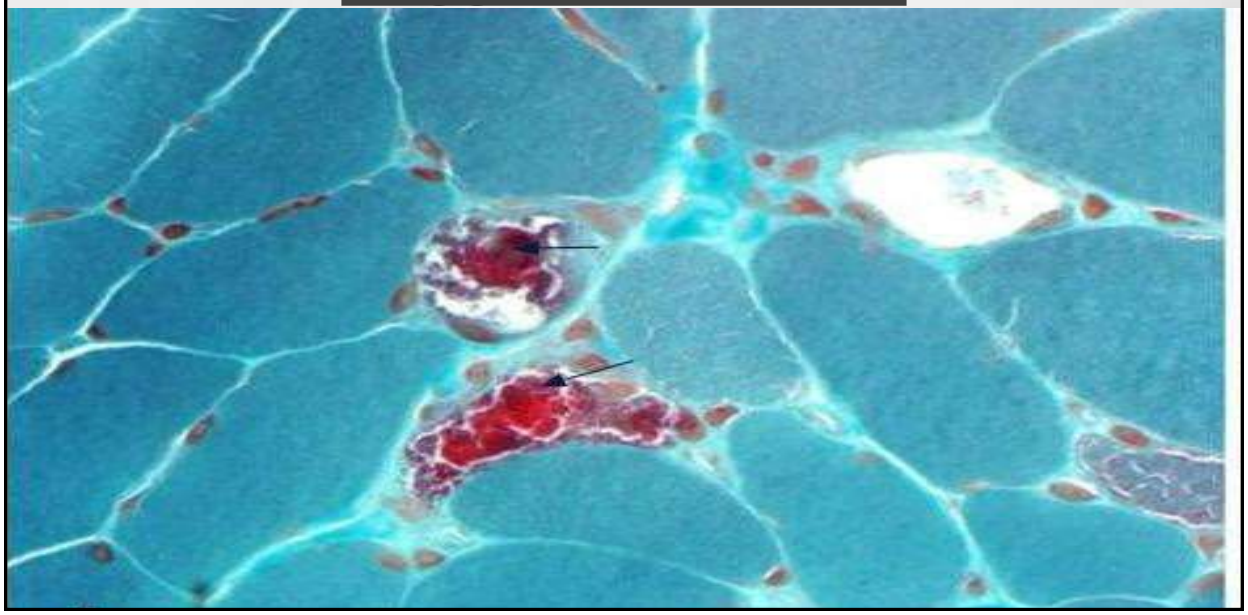


Kearns-Sayre Syndrome (KSS)

Confirmatory investigations

- mtDNA large deletion (ranging 1,000 to 10,000 nucleotides)
- Most common deletion removes 4997 nucleotides.
- PCR amplification of complete deletions in mtDNA by EDTA blood collected for separation of leukocytes.

Ragged Red Fibers



Speech discrimination

Left Severe Sensorineural hearing loss with fair Speech discrimination

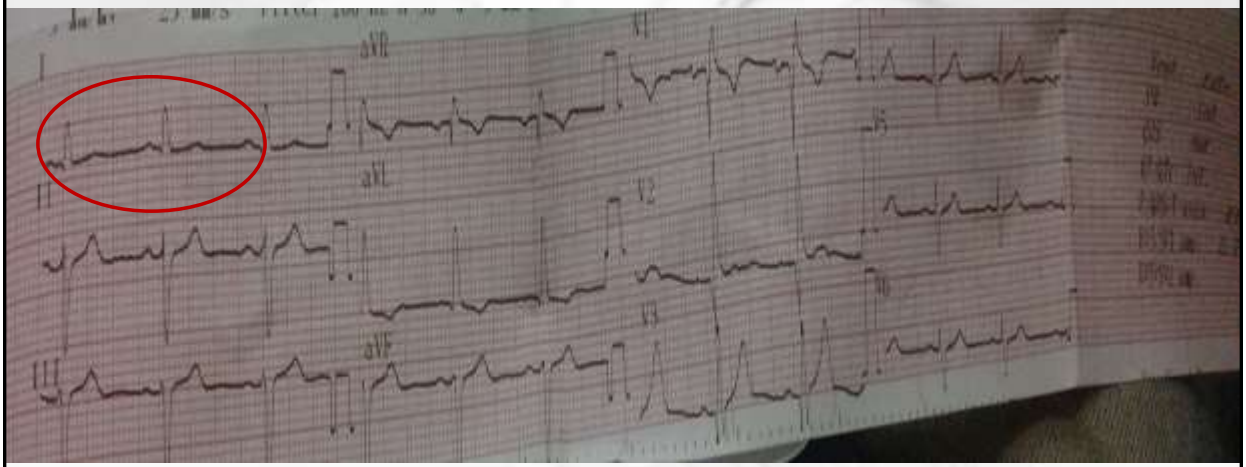
Bilateral type (A) tympanograms

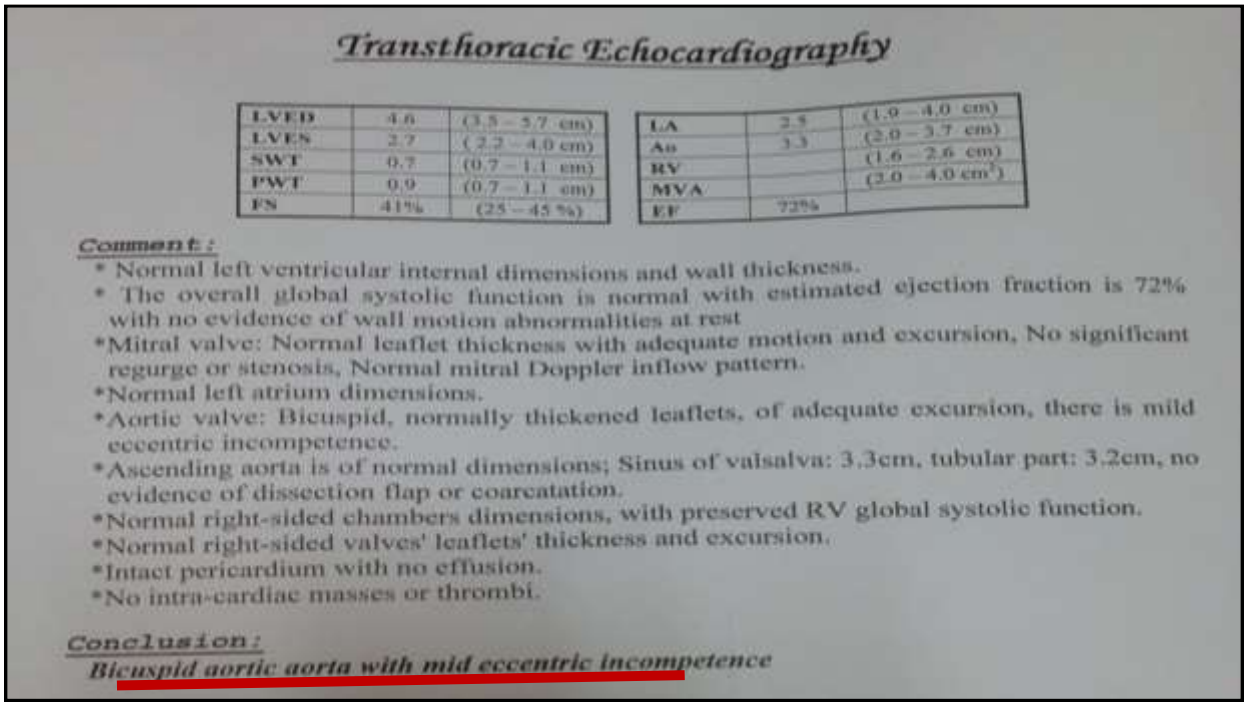
Recommendations:

~ for H.A fitting

IS KSS DEADLY??

Cardiac conduction block





Transthoracic Echocardiography

LVED	4.6	(3.5 – 5.7 cm)	LA	2.5	(1.9 – 4.0 cm)
LVES	2.7	(2.2 – 4.0 cm)	Ao	3.3	(2.0 – 3.7 cm)
SWT	0.7	(0.7 – 1.1 cm)	RV		(1.6 – 2.6 cm)
PWT	0.9	(0.7 – 1.1 cm)	MVA		(3.0 – 4.0 cm^2)
FS	41%	(25 – 45 %)	EF	72%	

Comment :

- Normal left ventricular internal dimensions and wall thickness.
- The overall global systolic function is normal with estimated ejection fraction is 72% with no evidence of wall motion abnormalities at rest
- Mitral valve: Normal leaflet thickness with adequate motion and excursion, No significant regurge or stenosis, Normal mitral Doppler inflow pattern.
- Normal left atrium dimensions.
- Aortic valve: Bicuspid, normally thickened leaflets, of adequate excursion, there is mild eccentric incompetence.
- Ascending aorta is of normal dimensions; Sinus of valsalva: 3.3cm, tubular part: 3.2cm, no evidence of dissection flap or coarcatation.
- Normal right-sided chambers dimensions, with preserved RV global systolic function.
- Normal right-sided valves' leaflets' thickness and excursion.
- Intact pericardium with no effusion.
- No intra-cardiac masses or thrombi.

Conclusion:

Bicuspid aortic aorta with mid eccentric incompetence

Management

- KSS is a complex disorder requiring the involvement of physicians from various specialties, including neurology, cardiology, ophthalmology, and endocrinology.
- In KSS, progressive cardiac conduction block is common and timely placement of pacemaker can extend lifespan.
- Coenzyme Q10 (CoQ10) administration has proven beneficial, although effects are transient.

What's new?

Coenzyme Q10 in the Treatment of Corneal Edema in Kearns-Sayre: Is There an Application in Fuchs Endothelial Corneal Dystrophy?

Kim J, et al. Cornea. 2016.
Show full citation

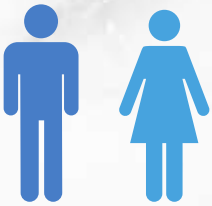
Abstract

PURPOSE: Corneal involvement in mitochondrial disease is seldom described. Kearns-Sayre syndrome (KSS) is a mitochondrial disorder characterized by retinitis pigmentosa, external ophthalmoplegia, and heart block. We report 2 patients with KSS with corneal lesions involving the endothelium, which improved with Coenzyme Q10 (CoQ10). Based on recent research regarding the role of dysfunctional oxidative metabolism in Fuchs Endothelial Corneal Dystrophy (FECD), we propose that mitochondrial diseases and FECD share a final pathway.

METHODS: A chart review was performed and a review of the literature was completed with a PubMed search using the terms "Kearns-Sayre Syndrome", "mitochondria", "endothelium", "Fuchs endothelial corneal dystrophy", and "cornea".

Take Home Message

Lives




Thank you
Dr. Nader Momtaz