Expansion of the Clinical Spectrum of Oguchi Disease

Caroline A. Tawfik [1], Noha I. Khater [2]

[1] Department of Ophthalmology, Ain Shams University, Cairo and Al Watany Eye Hospital, Electrophysiology of vision lab, Cairo, Egypt
[2] Al Mouneer Diabetic Eye Center, Dokki, Giza, Egypt

Introduction

Oguchi disease is a rare autosomal recessive congenital stationary night blindness with an abnormal-appearing fundus first described in the Japanese population by Oguchi in 1907.[1]

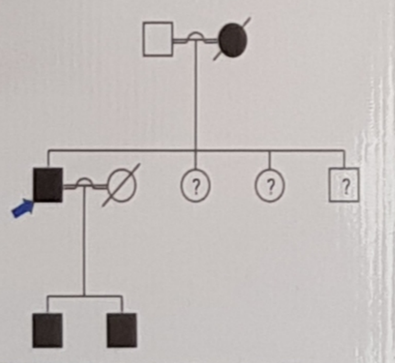
It is characterized by non-progressive retinal dysfunction with fundus appearance having a golden-yellow metallic sheen, with normal fundus coloration restored after prolonged dark adaptation > 2 hours (Mizuo-Nakamura phenomenon).[2]

Two genes have been implicated in Oguchi disease, both are involved in rod phototransduction: SAG (encoding arrestin) and GRK1 (encoding rhodopsin kinase)[3,4]

Fewer than 60 cases of Oguchi have been reported worldwide being primarily concentrated in Europe and Asia. To date, there have been no reports of Oguchi in the Middle East or Africa regions

Purpose

To describe a new clinical observation in 3 cases of a highly-consanguineous Egyptian family



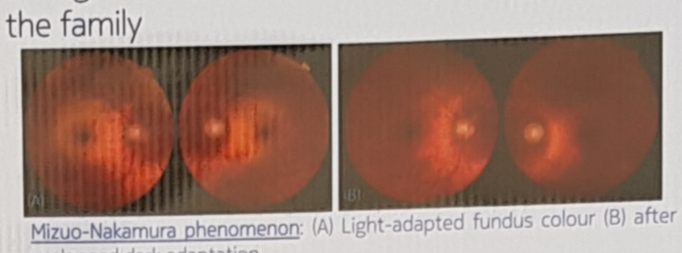
Methods

Full ophthalmological examination including Visual acuity (VA), intraocular pressure (IOP) by Goldmann applanation tonometery, fundus photography in light adaptation and after prolonged dark adaptation, optic disc assessment, visual field perimetry (VF), Central corneal thickness (CCT) and OCT ONH were done to all patients.

Criteria	Case 1	Case 2	Case 3
Age	66	31	28
VA	0.7	1.0	1.0
Initial IOP	36 OU	14 OU	17 OU
VCDR	0.75 OD 0.65 OS	0.6 OU	0.7 OU
CCT	560	545	555

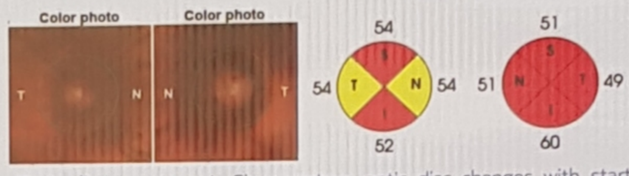
Case 1

Proband is a 66 years old man who was aware of night blindness since childhood, running in the family

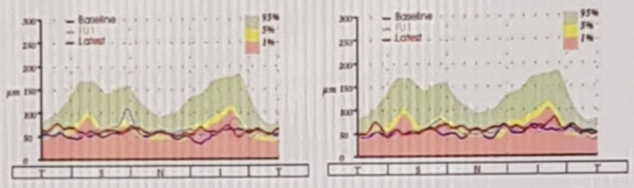


Mizuo-Nakamura phenomenon: (A) Light-adapted fundus colour (B) after prolonged dark adaptation

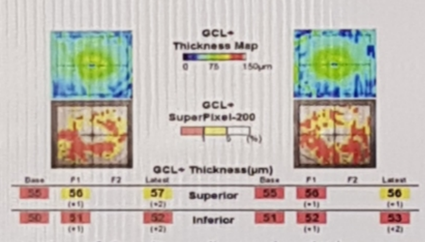
IOP: initially 36 mmHg stabilized on Latanoprost 0.005% down to 19 mmHg. Compliance was good initially then declined on subsequent follow-up visits



Optic disc assessment: Glaucomatous optic disc changes with starting inferior notching of neuroretinal rim, more evident in the left eye. Corresponding thinning of RNFL on OCT ONH



Serial Examination of TSNIT Graph: revealed loss of bi-hump configuration with corresponding thinning of RNFL correlating well with disc appearance

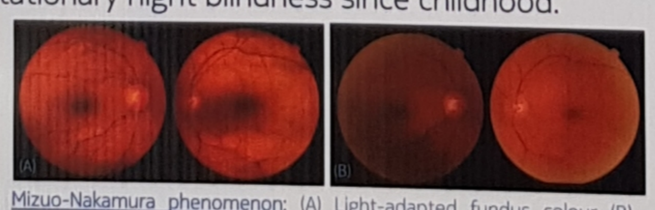


Serial Examination of Ganglion cell complex thickness map: superior and inferior thinning correlating well with optic disc changes and TSNIT graph

Serial Examination of VF: revealed corresponding field defect in the form of superior arcuate scotoma in right eye, and denser scotoma with nasal step in the left eye with progression in the last field

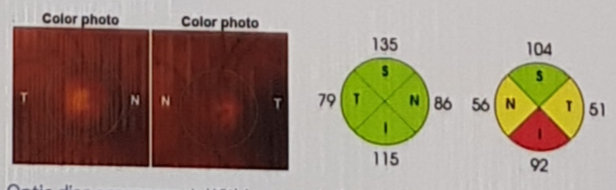
Case 2

Proband's elder son, 31 years old man with stationary night blindness since childhood.

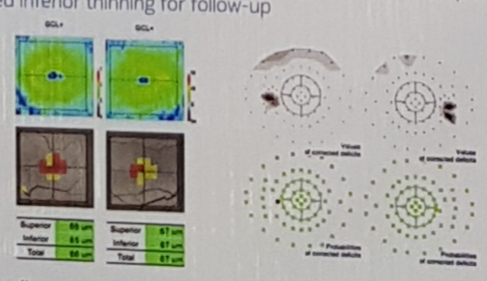


Mizuo-Nakamura phenomenon: (A) Light-adapted fundus colour (B) after prolonged dark adaptation

IOP: within normal IOP (14 OU)



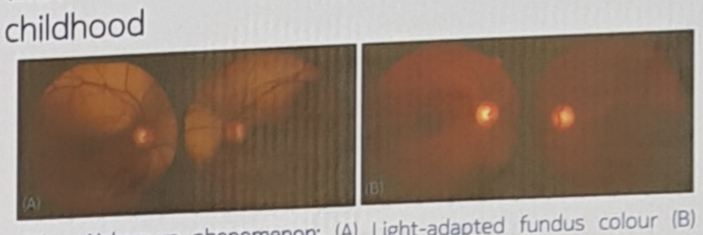
Optic disc assessment: Within normal disc appearance with deep cups. Left eye showed inferior thinning for follow-up



Ganglion cell complex thickness map and VF: revealed within normal parameters, and no scotomata of particular significance or pattern

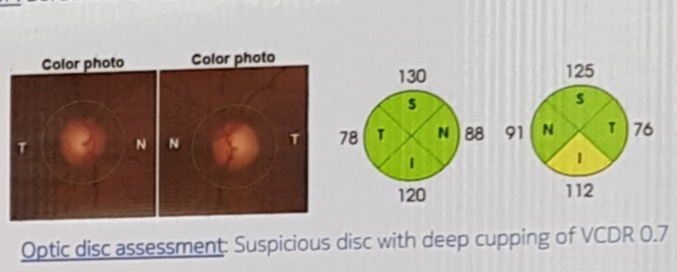
Case 3

Proband's younger son, 28 years old man with stationary night blindness since childhood

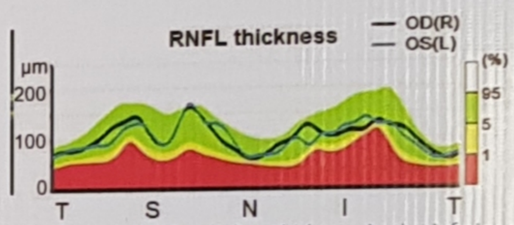


Mizuo-Nakamura phenomenon: (A) Light-adapted fundus colour (B) after prolonged dark adaptation

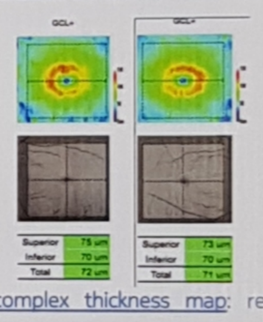
IOP: Borderline IOP (20 mmHg) initially as well on follow-up visit



Optic disc assessment: Suspicious disc with deep cupping of VCDR 0.7



TSNIT Graph: revealed borderline thickness in the inferior rim of left eye



Ganglion cell complex thickness map: revealed within normal parameters

Serial Examination of VF: revealed a superior field defect in right eye, and a denser superior scotoma in the left eye with improvement in the last field possibly due to learning effect

Discussion

To our knowledge, glaucoma was not described before as a possible association to Oguchi disease.

The highly-consanguineous nature of the family could be responsible for condensation of glaucoma risk in the family.

Conclusion

Open angle glaucoma could be a new feature associated with Oguchi disease.

Genetic testing to exclude the presence of other genes responsible for glaucoma in the family is pending.

Acknowledgment

Special thanks go to Prof. Azza Shehab; my mentor for her continuous support

References

1. Oguchi C. Ueber eine Abart von Hemeralopie. Acta Soc Ophthalmol Jpn 1907;11:123–134.
2. Mizuo G. On new discovery in dark adaptation in Oguchi's disease. Acta Soc Ophthalmol Jpn 1914;18:73–127.
3. Dryja TP. Molecular genetics of Oguchi disease, fundus albipunctatus, and other forms of stationary night blindness: LVII Edward Jackson memorial lecture. *Am J Ophthalmol* 2000;130(5):547–563.
4. Yamamoto S, Sippel KC, Berson EL, Dryja TP. Defects in the rhodopsin kinase gene in the Oguchi form of stationary night blindness. *Nat Genet* 1997;15(2):175–178.

International Society of Clinical Electrophysiology of Vision 56th Symposium, Reims, France, June 2018