Clinical and Genetic Characteristics of enhanced S-cone syndrome in a Tunisian cohort: experience of the oculogenetic laboratory LR14SP01

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Introduction

Enhanced S-cone syndrome (ESCS) is a rare autosomal recessive vitreoretinal degeneration that is characterized by night blindness and increased sensitivity to blue light. Clinically, there may be various fundus appearances, but there are pathognomonic electroretinographic features that helps in confirming the diagnosis.

The aim of this study was to report the clinical, multimodal imaging and genetic characteristics of patients with ESCS in a Tunisian cohort.

Methods

Descriptive retrospective clinical and genetic study including 480 patients with inherited retinal dystrophies who consulted the oculogenestics laboratory LR14SP01, at Hédi Rais Institute of ophthalmology between 2006 and 2017.

A total of 9 patients from 6 consanguineous families were diagnosed with ESCS based on typical clinical and ERG abnormalities.

Patients underwent thorough ophthalmologic examination, including best-corrected visual acuity, slit lamp biomicroscopy, fundus photography, fundus autofluorescence, swept source optic coherence tomography and electrophysiology.

Results

F	P	Gender	Age years	Visual acuity RE	Visual acuity LE	Ophthalmoscopy	OCT	Gene	Mutation
1	1	F	32	1/10	2/10	Radial subtil pigmentary changes in the PP. Mid-peripheral nummular pigment clumping and yellow dots	Macular cysts	***NR2E3***	p.R311Q
	2	F	40	3/10	3/10	Radial subtil pigmentary changes in the PP. Mid-peripheral nummular pigment clumping and atrophy	Cystoid ME	***NR2E3***	p.R311Q
2	3	F	19	1/20	1/10	Mid-peripheral nummular pigment clumping and atrophy	Diffuse cystoid ME	***NR2E3+ RHO***	***NR2E3*** p.[R311Q],[R3 1Q] + ***RHO*** p.[G51A],[=]
	4	F	68	LP	LP	Diffuse retinal atrophy	Macular atrophy	***NR2E3+ RHO***	
3	5	M	55	LP	LP	Mid-peripheral nummular pigment clumping and atrophy	ME	***NR2E3***	p.G56R
4	6	M	26	5/10	4/10	minimal pigmentary changes along the arcades	Macular cysts	***NR2E3***	p.R311Q
	7	M	22	5/10	3/10	minimal pigmentary changes along the arcades	Macular cysts	***NR2E3***	p.R311Q
5	8	F	8	2/10	2/10	Mid-peripheral nummular pigment clumping	Cystoid ME	-	-
6	9	M	13	2/10	2/10	Mid-peripheral nummular pigment clumping	Cystoid ME	-	-

F: family; P: patient; RE: right eye; LE: left eye; PP: posterior pole; ME: macular edema

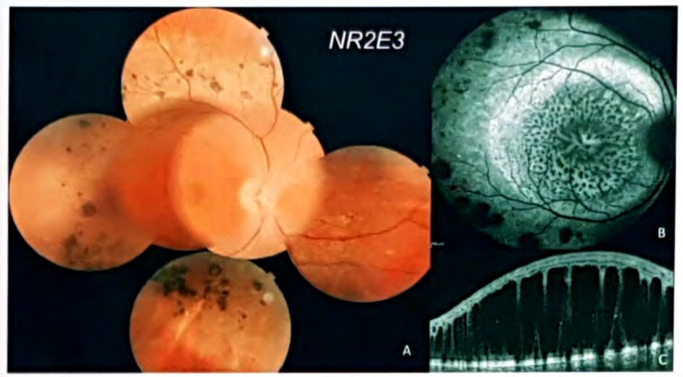


Figure 1: Patient 5 with Mid-peripheral nummular pigment clumping (A), cystoid autofluorescence (B) and important cystoid ME (C)

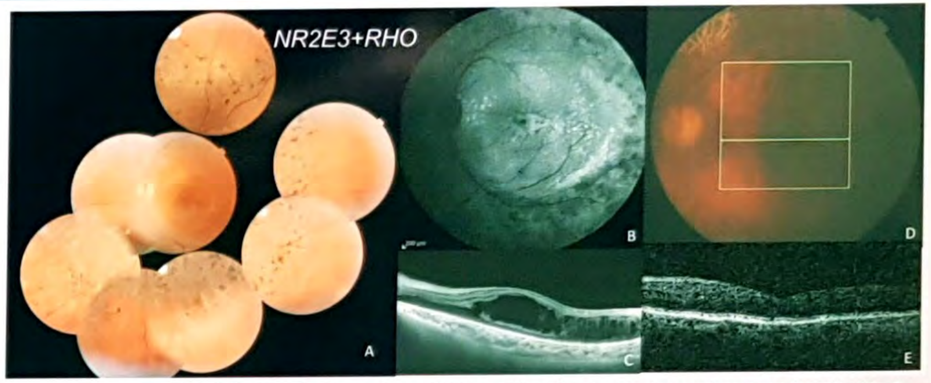


Figure 2: F 2. (A): Fundus imaging of LE of P3 with mid-peripheral nummular pigment clumping and atrophy ; (B): FAF showing areas of atrophy in the mid periphery;; C: SS-OCT showing an important macular edema. (D): fundus photo of the right eye of the mother P4, E: SS-OCT showing diffuse chorio-retinal atrophy

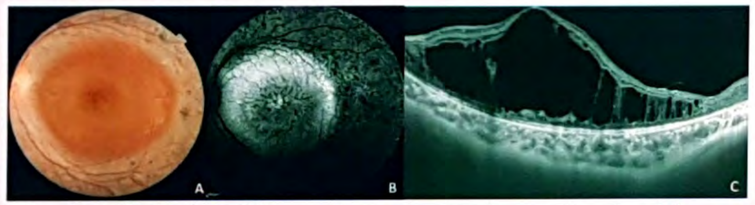
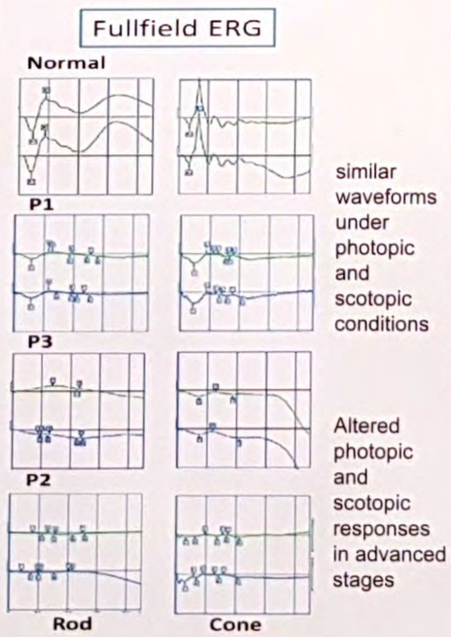


Figure 3: Patient 9 with posterior nummular pigment clumping (A), diffuse heterogenous FAF(B) and important cystoid ME (C)



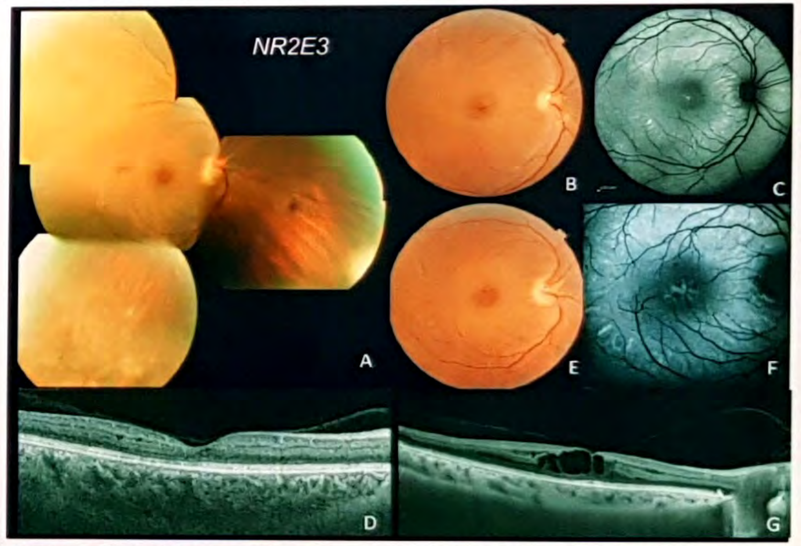


Figure 4: Clinical features of patients from F1 .A and B: Fundus imaging of RE of P1E: fundus photo of RE of P2. C and F: radial hyper FAF; D and G: SS-OCT showing cystoid macular edema.

Discussion and conclusions

This is to our knowledge the first report on a Tunisian cohort with ESCS. The phenotype in ESCS is highly variable and multimodal imaging may help to the diagnosis in addition to pathognomonic electrophysiology results.

In advanced stages, diagnosis became difficult because of diffuse retinal atrophy.

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