

Severe retinitis pigmentosa with posterior pole staphyloma in a family harbouring c.886 A>G *RHO* mutation.

Vasily SMIRNOV(1,2), Claire-Marie DHAENENS (3), Bernard PUECH(1), Sabine DEFOORT-DHELLEMMES(1)

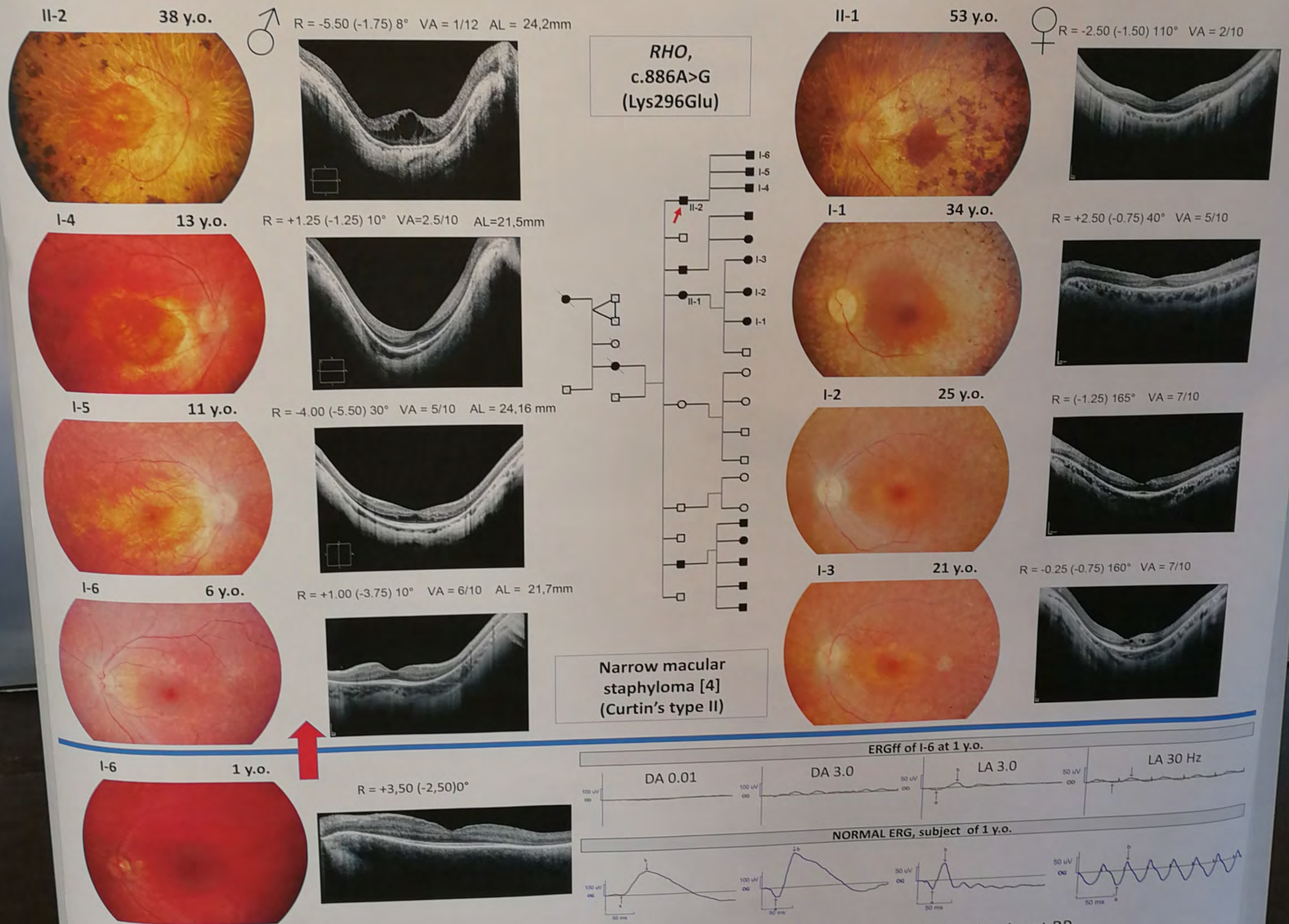
(1) Département of Visual Explorations and Neuro-ophthalmology
 (2) Université de Lille, Faculté de Médecine

(3) Biochemistry and Molecular Biology Department – UF Génopathies, University Hospital of Lille, France



Introduction. Posterior pole staphylomata were unfrequently reported in association with inherited retinal degenerations[1]. We report here a large family suffering from a peculiar form of retinitis pigmentosa (RP) with posterior pole staphyloma (PPS).

Patients: All the affected members complained of hemeralopia from their early childhood. The youngest patient (I-6) of the “male” branch was examined at the age of 11 months old. His scotopic ERG was already extinguished and the photopic ERG responses were severely reduced. He did not have PPS at this stage and was hyperopic, but the signs of posterior pole bowing became evident at 6 y.o. In all other affected members ERG was extinguished. 2 older brothers (I-5 and I-4) were first seen at the age of 5 et 10 years old, respectively. The eldest one (I-4) had already a large PPS and the younger one (I-5) developed it by the age of 10 years old. In another “female” branch of this family (II-1 and I-1-3), PPS was present in all but one (I-1) relatives. Interestingly, the **axial length was rather short** in these patients. A myopic shift was observed for all the patients at follow-up visits. Staphylomas were of narrow macular (Curtin’s II) type.



Discussion: We could clinically observe a PPS development in initially non-myopic patients of a family with a dominant RP. C.886 A>G *RHO* mutation is known to be unusually severe one[2] with the complete loss of function of rhodopsin[3]. The other reported family with the same mutation had a similar severe RP[2]. In our patient I-6 the myopic shift was secondary to PPS formation. The PPS concerned only the area with relatively preserved outer retinal structures (ONL and EZ). Does the outer retina guide choroid and scleral remodeling and thus emmetropization?

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