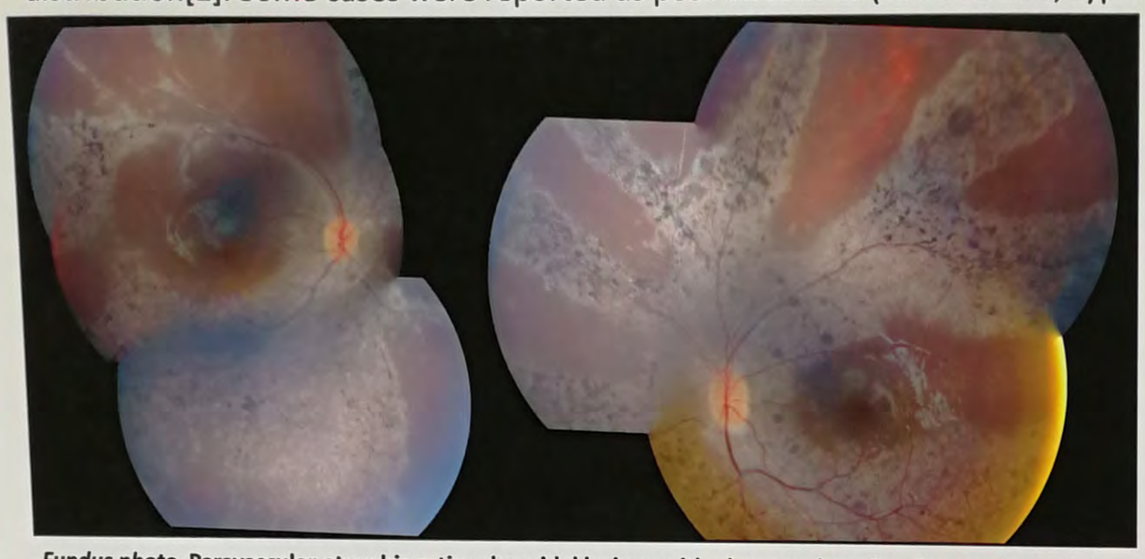
Pigmented paravenous chorioretinal atrophy in a boy with Inflammatory Bowel phenotype of Chronic Granulomatous Disease.

Sabine DEFOORT-DHELLEMMES(1), Vasily SMIRNOV(1), Bernard PUECH(1), Frédérique GOTTRAND(2), Brigitte NELKEN(3)

1. Visual Explorations and Neuro-Ophthalmology Department
2. Pediatric Gastroenterology, Hepatology and Nutriton Department
3. Pediatric Hematology Department

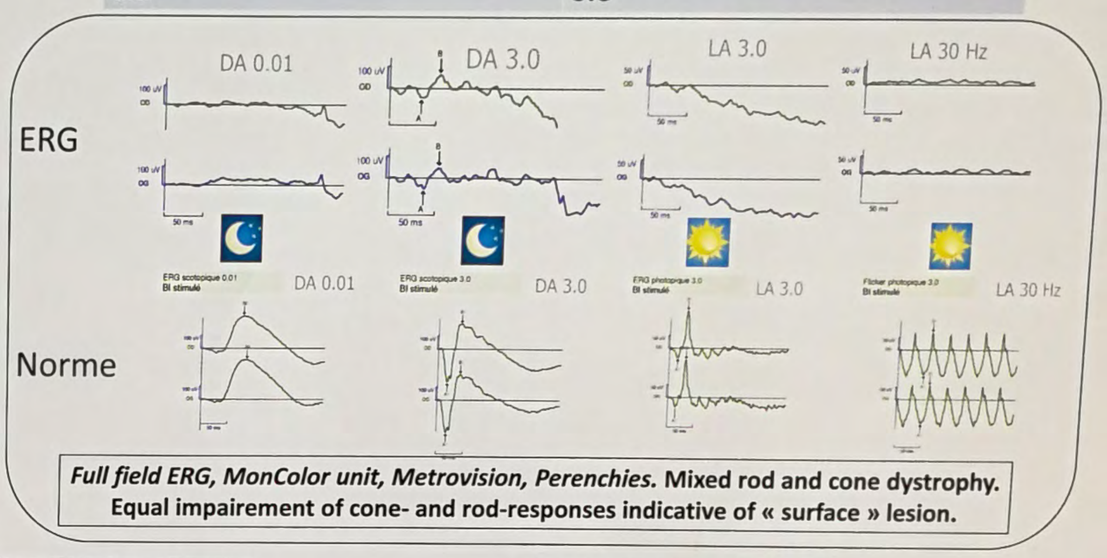
CHRU de Lille, Lille, France
Université de Lille, Faculté de Médecine, Lille, France

Introduction. Pigmented paravenous chorioretinal atrophy (PPCRA) is a relatively stationary degeneration of external retina with a specific pattern of lesion distribution[1]. Some cases were reported as post-infectious (tuberculosis, syphilis, measles and rubella) or post-vaccinal[2].

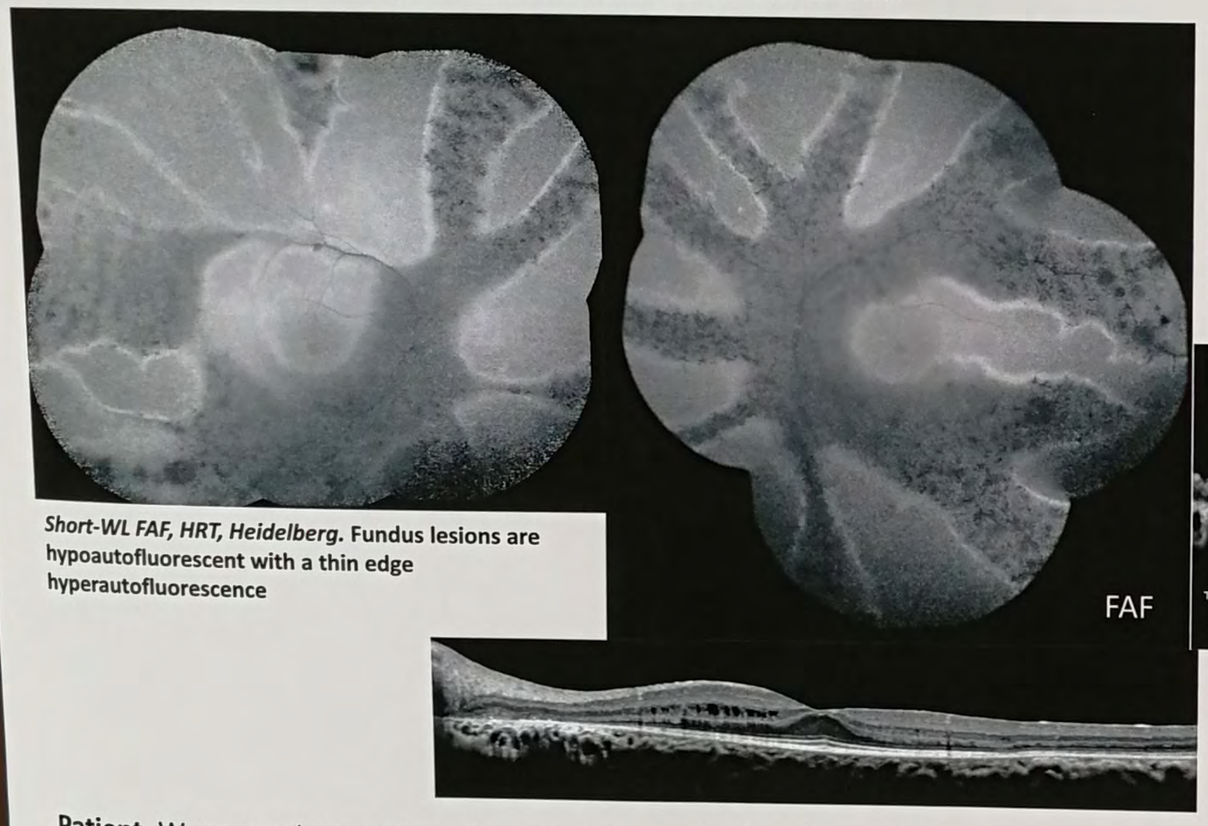


Fundus photo. **Paravascular atrophic retinochoroidal lesions with pigment clumping disposed in radial pattern (« retinochoroiditis radiata »). All images obtained at 4 y.o. without sedation.**

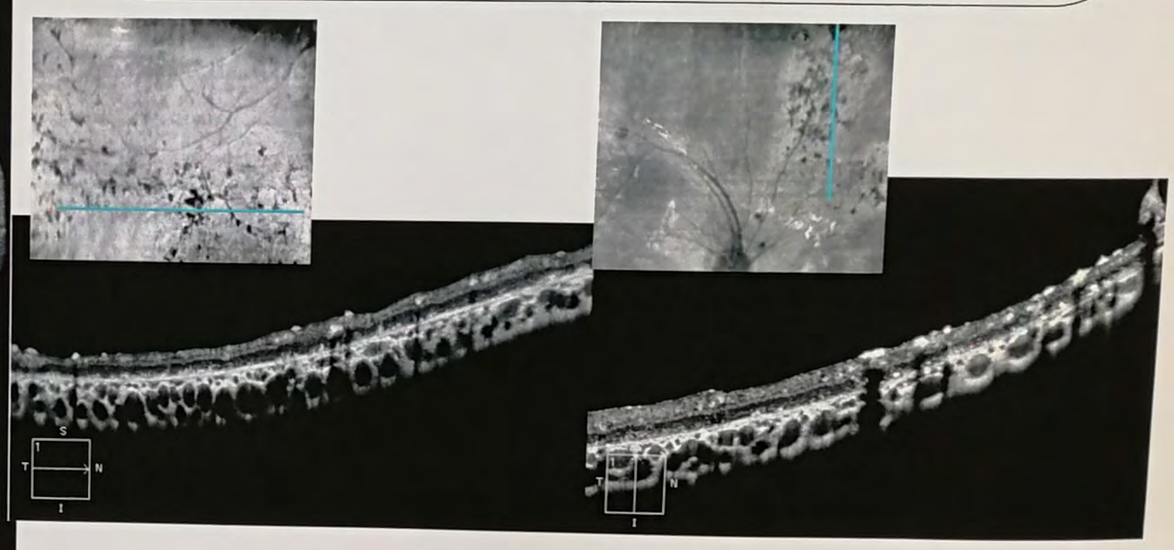
Best corrected VA at 4 y.o.	
OD	OS
+3,0 (- 3,0) 160°	+1,50(-1,50)165°
0.5	0.6



Full field ERG, MonColor unit, Metrovision, Perenchies. **Mixed rod and cone dystrophy. Equal impairement of cone- and rod-responses indicative of « surface » lesion.**



Short-WL FAF, HRT, Heidelberg. **Fundus lesions are hypoautofluorescent with a thin edge hyperautofluorescence**

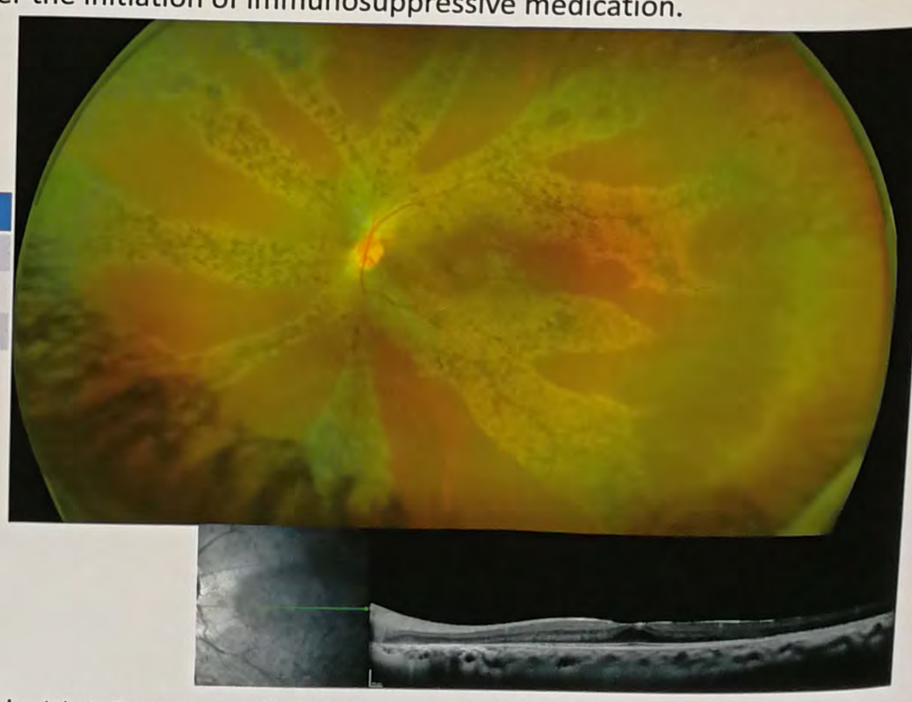


SD-OCT, Cirrus, Zeiss. **Destruction of outer retinal layers and global retinal thinning in affected areas. « Sponge» appearance of choroid. Note macular intraretinal cysts. This edema resolved after initiation of immunosupressive drugs.**

Patient. We report here a case of a 4 year-old boy with a PPCRA discovered at the routine ophthalmic examination after a febrile seizure. At the repeated ophthalmic assessment we noted a chronic diarrhoea with a weight stagnation. Gastroenterological examination revealed a severe Crohn-like inflammatory bowel disease. Standard therapy (azathioprine and infliximab) was inefficient and led to multiple complications (colic perforation, subcorneal pustular dermatosis) . Entero-MRI revealed a diffuse infiltration of mesenteric fat. Further hematologic investigation revealed altered neutrophilic function (Nitroblue tetrazolium test was 0%). Recurrent homozygous mutation *c.75_76delGT* were found in *NCF1* gene thus confirming the diagnosis of Chronic Granulomatous Disease (CGD). Clinical status improved after introduction of preventive antibiotic and antifungal treatment. Fundus lesions were non progressive. Macular edema resolved after the initiation of immunosuppressive medication.

Optos® UWF fundus photos **at 7 y.o. Chorioretinal lesions are non progressive.**

Best corrected VA at 7 y.o.	
OD	OS
+4,0 (- 3,0) 0°	+3,25(-1,25)165°
0.8	0.8



Discussion. The inflammatory origin of PPCRA is usually discussed but few cases were undoubtedly linked with inflammatory/infectious event (tuberculosis, syphilis, measles, rubella, MMR vaccination, chronic uveitis, Behçet disease). Predominantly perivascular outer retinal destruction supporting the probable immune mechanism of lesions. Some family cases of PPCRA were described[3-5], but there is no definitive evidence for hereditary origin[2].

CGD is an inherited immunodeficiency syndrome characterized by incapacity of neutrophils to achieve phagocytosis[6]. Thus, recurrent bacterial and fungal infections of barrier tissues occur. Active chorioretinal granulomas and punched-out scars were seen in 20-40% of patients with CGD[7,8]. To our best knowledge, PPCRA was never reported in association with CDG. Noteworthy, the discovery of PPCRA in our patient preceded and precipitated the diagnosis of CDG.

Our case of PPCRA is remarkable for both inflammatory and hereditary causes are intermingled.

1. Brown TH: Retino-choroiditis radiata. Br J Ophthalmol 21: 645-648, 1937.
2. Shona OA, Islam F, Robson AG, Webster AR, Moore AT, Michaelides M. Pigmented Paravenous chorioretinal atrophy - detailed clinical study of a large cohort. Retina. January 2018:1.
3. Traboulsi EI, Maumenee IH. Hereditary pigmented paravenous chorioretinal atrophy. Arch Ophthalmol Chic Ill 1960. 1986 Nov;104(11):1636–40.
4. Noble KG. Hereditary pigmented paravenous chorioretinal atrophy. Am J Ophthalmol. 1989 Oct 15;108(4):365–9.
5. Obata R, Yanagi Y, Iriyama A, Tamaki Y. A familial case of pigmented paravenous retinochoroidal atrophy with asymmetrical fundus manifestations. Graefes Arch Clin Exp Ophthalmol Albrecht Von Graefes Arch Klin Exp Ophthalmol. 2006 Jul;244(7):874–7.
6. Segal BH, Leto TL, Gallin JI, Malech HL, Holland SM. Genetic, biochemical, and clinical features of chronic granulomatous disease. Medicine (Baltimore). 2000;79(3):170-200.
7. Kim S-J, Kim JG, Yu YS. Chorioretinal lesions in patients with chronic granulomatous disease. Retina Phila Pa. 2003;23(3):360-365.
8. Valluri S, Chu FC, Smith ME. Ocular pathologic findings of chronic granulomatous disease of childhood. Am J Ophthalmol. 1995;120(1):120-122.