doi: 10.3978/j.issn.1000-4432.2019.09.05
View this article at: http://dx.doi.org/10.3978/j.issn.1000-4432.2019.09.05

无色素性视网膜色素变性1例

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[摘　要] 报告1例2019年1月因无色素性视网膜色素变性而就诊的病例。患者因发现视力差，常规眼科检查及全身的检查未发现异常，给予眼底荧光造影后确诊。给予眼底荧光造影后最终确诊为罕见的无色素性视网膜色素变性，防止了疾病的漏诊和误诊。对于缺乏视网膜色素变性典型的三联征的无色素性视网膜色素变性患者，临床要谨防漏诊，眼底荧光血管造影(fundus fluorescein angiography，FFA)可明确诊断。

[关键词] 无色素性视网膜色素变性；视网膜色素变性

Retinitis pigmentosa sine pigmento: A case report

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Abstract We reported a case of achromatic retinitis pigmentosa admitted to the hospital in January 2019. The patient was diagnosed by fundus fluorescein angiography because of poor vision, and no abnormality was found by routine ophthalmological examination and general examination. After fundus fluorescein angiography (FFA), a rare non-pigmented retinitis pigmentosa was finally diagnosed, which prevented misdiagnosis and missed diagnosis of the disease. For the patients without typical triad of retinitis pigmentosa, we should pay attention to missing the diagnosis in clinical practice. FFA can make a definite diagnosis of retinitis pigmentosa.

Keywords retinitis pigmentosa sine pigmento; retinitis pigmentosa

无色素性视网膜色素变性(retinitis pigmentosa sine pigmento，RPSP)是一种特殊类型的原发性视网膜色素变性(retinitis pigmentosa，RP)，眼底表现除无典型的骨细胞样色素沉着以外，其眼底表现与典型性RP相同，临床上比较罕见[1]，目前仅可见少量零星的病例报道[2-3]，容易被漏诊。云南省第二人民医院眼科收治1例因无色素性视网膜色素变性而就诊的病例，现报道如下。

收稿日期 (Date of reception)：2019–08–17

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基金项目 (Foundation item)：云南省眼部疾病临床医学研究中心内设课题 (YXZX -2019012)。This work was supported by Yunnan Ophthalmic Disease Clinical Medical Center, China (YXZX -2019012).

1 临床资料

患者，女，51岁，因“双眼夜视力下降明显5年”就诊。患者既往有夜间视力差25年，否认父母近亲联姻，有1个妹妹，目前妹妹、侄女患有无色素性视网膜色素变性，否认父母亲、父母的兄弟姐妹、祖父母患此病及否认其他低视力家族史。眼部检查：视力，双眼0.5，矫正无提高。双眼裂隙灯前节检查见轻度白内障。眼底检查：患者双眼底可见视盘颜色变淡，C/D=0.4。视网膜血管管径变细、轻度迂曲，后极部见不同程度的弥漫性斑点状色素脱失，色污黄，未见明显视网膜骨细胞样色素沉着(图1A，B)。眼底荧光血管造影(fundus fluorescein angiography，FFA)：双眼视网膜广泛弥漫性斑点状透见荧光，RPE萎缩区域呈斑驳状高荧光，未见充盈缺损区域(图1C，D)。双眼视网膜电图(electroretinogram，ERG)检查(图2)：基本呈熄灭型。双眼视野(图3)：管状视野。双眼黄斑光学相干断层成像(optical coherence tomography，OCT)(图4)：黄斑区视网膜变薄。综合上述相关检查及病史、症状、体征，诊断：“双眼无色素性视网膜色素变性”。

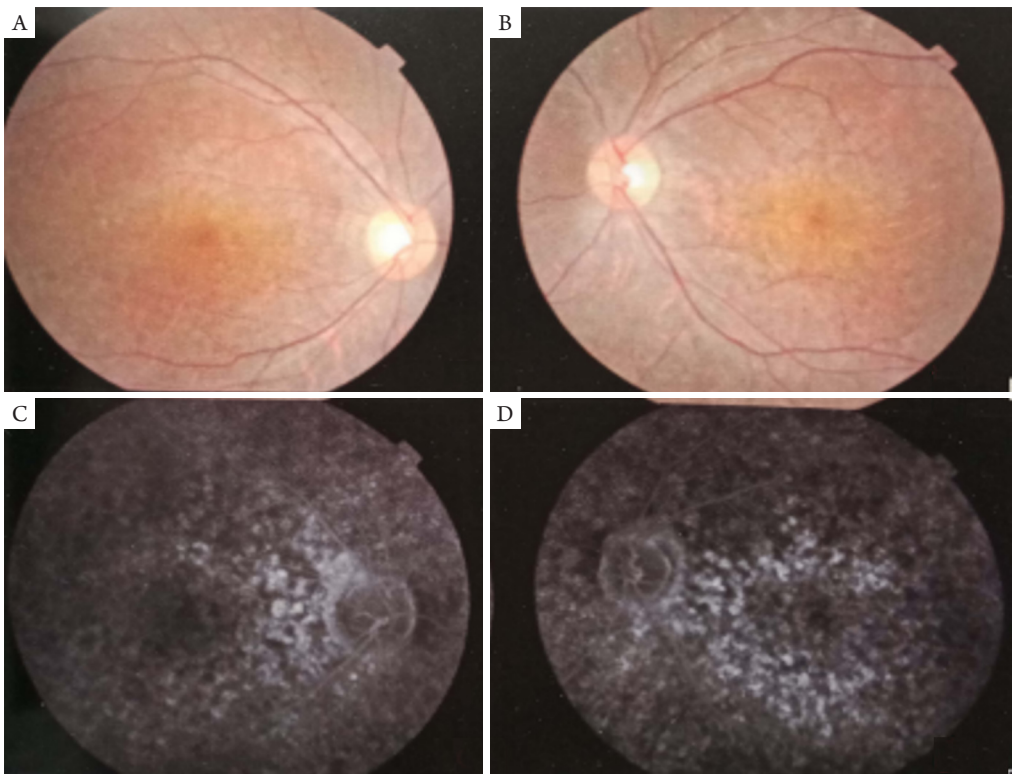

图1 无色素性视网膜色素变性眼底影像学检查

Figure 1 Fundus imaging examination of retinitis pigmentosa sine pigmento

右眼(A)、左眼(B)眼底彩色照相：眼底可见视盘颜色变淡，C/D=0.4，视网膜血管管径变细、轻度迂曲，后极部见不同程度的弥漫性斑点状色素脱失，色污黄，周边少许视网膜骨细胞样色素沉着。右眼(C)、左眼(D)眼底荧光造影：双眼视网膜后极部弥漫性斑点状透见荧光，RPE萎缩区域呈斑驳状高荧光，未见充盈缺损区域。

Right eye (A) and left eye (B) fundus color photography: fundus disc color became lighter, C/D=0.4, retinal vascular diameter became thinner and slightly tortuous, posterior pole showed diffuse spotted pigmentation, yellowish, with A few peripheral retinal osteoblast-like pigmentation. Right eye (C) and left eye (D) fundus fluorescein angiography: diffuse patchy fluorescence was seen in the posterior pole of the retina in both eyes, and patchy hyperfluorescence was seen in the atrophic area of RPE, without filling defect.

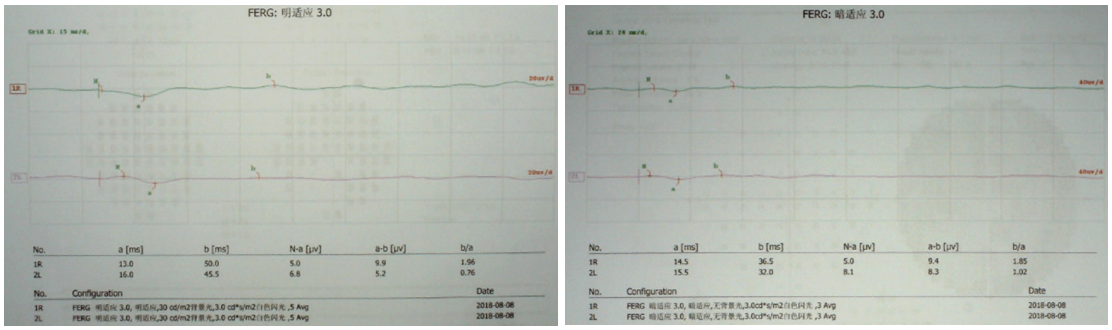

图2 双眼ERG：双眼(未散瞳)明适应3.0a-波b-波振幅降低，暗适应3.0b-波振幅降低

Figure 2 Binocular ERG: binocular (undisturbed) visual field 3.0a-wave b-wave amplitude decreases, and visual field 3.0b-wave amplitude decreases

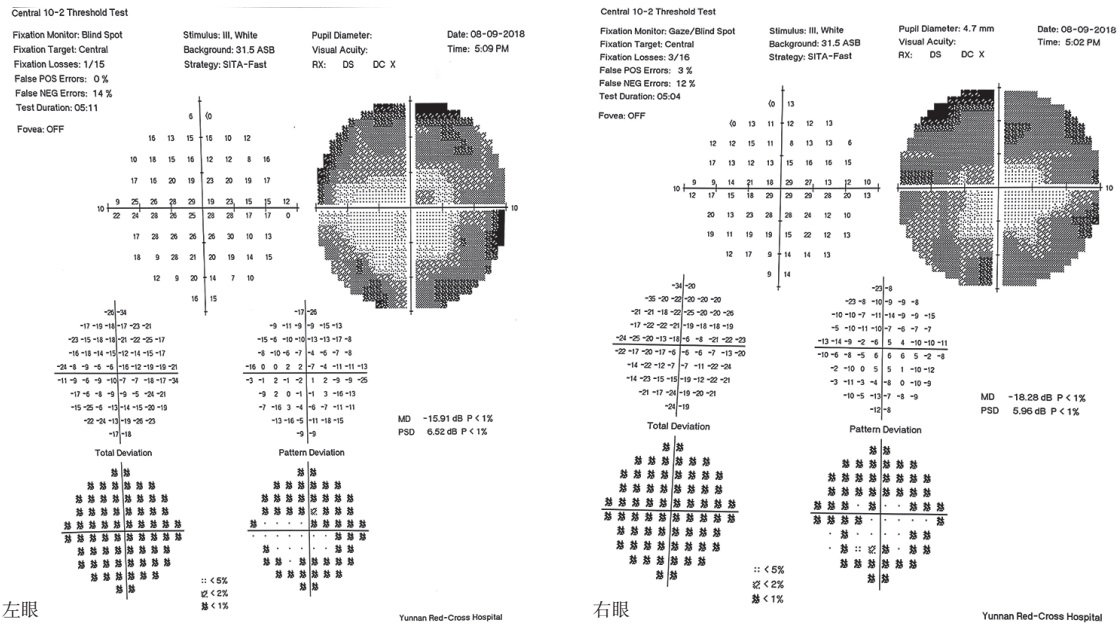

图3 双眼视野：管状视野

Figure 3 Binocular vision: tubular vision

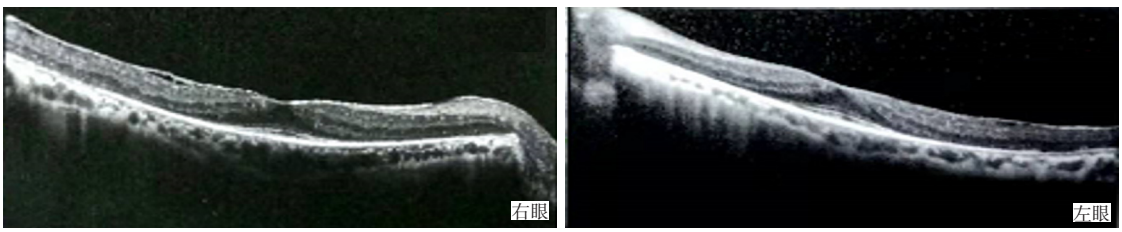

图4 双眼黄斑OCT：色素上皮层萎缩，周边视网膜变薄，各层结构明显萎缩

Figure 4 Binocular macular OCT: pigment epithelium atrophy, peripheral retina thinning, obvious atrophy of each layer structure

2 讨论

RPSP与典型的RP相比，无色素型视网膜色素变性眼底无或仅有极少色素沉着，但不能凭色素沉积的多少来决定疾病的严重性[4]。但同样可出现夜育、典型的眼底改变如视网膜色素脱失、视网膜血管变细、视盘蜡黄以及典型性视野和ERG改变。也有学者[5]认为随着患者年龄增大，病变进展，眼底会逐渐出现骨细胞样色素沉着。据文献[6]报道：RPSP FFA表现为弥散性椒盐状透见荧光，未见骨细胞样遮蔽荧光，本病早期以影响视杆细胞为主，全视野ERG出现A波及B振幅值的降低，随着病情的逐渐发展，累及视锥细胞，晚期出现视功能的完全丧失[7]。本病与文献报道基本一致，本例患者的病史、临床表现与FFA，ERG和视野检查均符合RPSP的典型表现，故不难诊断。

因为该病缺乏视网膜色素变性典型三联征的眼底表现，因此掌握视网膜色素变性患者眼底有无色素沉着，对漏诊和误诊具有重要的意义[8]。日前对于无色素型色素变性尚无有效的治疗方法，而全身应用扩张血管药物和补充维生素A可能对其治疗有所帮助[5]，另外针刺、中药对其治疗亦有所帮助。

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本文引用：杜福生, 彭华. 无色素性视网膜色素变性1例[J]. 眼科学报, 2019, 34(4): 274-277. doi: 10.3978/j.issn.1000-4432.2019.09.05
Cite this article as: DU Fusheng, PENG Hua. Retinitis pigmentosa sine pigmento: A case report[J]. Yan Ke Xue Bao, 2019, 34(4): 274-277. doi: 10.3978/j.issn.1000-4432.2019.09.05