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·病例报告·

无明显外伤史的脉络膜破裂：1例病例报道并文献复习

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[摘　要] 脉络膜破裂大部分是由撞击引起的对冲伤，冲击力通过玻璃体传导眼底引起，因此一般脉络膜破裂会有比较明确的外伤或者钝挫伤病史。本文将报告1例16岁体校男生在无明显外伤史出现多发性脉络膜破裂伤，通过查阅文献发现有文献报道在隐匿性假性黄色瘤(pseudoxanthoma elasticum，PXE)疾病中可在无明显外伤或轻微外伤出现脉络膜破裂，并根据文献复习考虑本病例为隐匿性PXE可能。

[关键词] 脉络膜破裂；隐匿性假性黄色瘤；外伤史；病例报道；文献复习

Choroidal rupture without obvious trauma: a case report and literature review

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Abstract Most of the choroidal rupture is mostly caused by impact injury, and the impact force is caused by the vitreous conduction through the fundus. Therefore, the choroidal rupture generally has a clear history of trauma or blunt trauma. This article will report a case of a 16-year-old boy in a sports school who developed multiple choroidal ruptures without obvious trauma history. Through literature review, it was found that choroidal rupture can occur without obvious or minor trauma in subtle pseudoxanthoma elasticum (PXE) disease, and based on literature review, this case was considered as a possibility of subtle PXE.

Keywords choroidal rupture; pseudoxanthoma elasticum; history of trauma; case report; literature review

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脉络膜破裂[1]是在外伤事件中引起视网膜色素上皮(retinal pigment epithelium，RPE)、Bruch膜及脉络膜毛细血管层断裂，通常会引起视网膜下、视网膜膜内及脉络膜内出血造成视功能下降。5%~10%的眼外伤患者会出现脉络膜破裂[2]，其中约80%是由间接创伤引起[3-4]，一般是由撞击引起的对冲伤，冲击力通过玻璃体传导眼底引起脉络膜破裂[5]。因此一般脉络膜破裂会有比较明确的外伤或者钝挫伤病史，但也有文献[6]报道在无明显外伤或轻微外伤出现脉络膜破裂的病例。本文报告1例同样无明显外伤史出现脉络膜破裂伤并进行文献复习。

1 病例资料

患者，男，16岁，体校学生，汉族，因“右眼眼前黑影遮挡，伴视力下降2周”为主诉入院。2周前无明显诱因出现右眼眼前黑影遮挡，伴视力下降，不伴眼红、眼痛、畏光、流泪等症状。既往体健，生命征平稳，心肺腹未发现明显异常。眼部情况：右眼视力0.02，−3.00DS/−1.25DC*171=0.25，左眼戴镜视力0.4，−1.75DS/−1.50DC*9=1.0，双眼眼前节情况可，左眼眼底大致正常。右眼眼底及辅助检查(图1~6)：黄斑区见一不连续弧形黄白色病灶，其周围见类圆形出血性神经上皮脱离(图1)。自发荧光显示黄斑区多个弧形呈低荧光病灶，黄斑去血性神经上皮脱离，部分因血自发荧光增强(图2)。行光学相干断层扫描血流成像(optical coherence tomography angiography，OCTA)检查，在视网膜无血管层可见弧形低密度区及网状CNV结构(图3)，在脉络膜毛细血管层见经过黄斑中央脉络膜毛细血管弧形缺损区，暴露其下脉络膜中层血管(图4)。行荧光素钠造影(fluorescein angiography，FA)联合吲哚菁绿造影(indocyanine green angiography，ICGA)检查，ICGA见经过黄斑中心凹处一新月形弱荧光，视盘颞侧见多处小弧形弱荧光，均为脉络膜破裂部位。破裂病灶对应线扫光学相关断层扫描(optical coherence tomography，OCT)图RPE-Bruch膜复合体及脉络膜毛细血管断裂处，凹陷处见中高反射瘢痕信号，FA可见弧形染色并且中央有楔形荧光渗漏，考虑有脉络膜新生血管(choroidal neovascularization，CNV)形成可能(图5)。诊断：右眼多发性脉络膜破裂并CNV形成、黄斑视网膜下出血、双眼屈光不正。治疗上予玻璃体腔注射抗血管内皮生长因子(vascular endothelial growth factor，VEGF)治疗后1个月随访，视网膜下出血吸收，矫正视力提高到0.6(图6)。

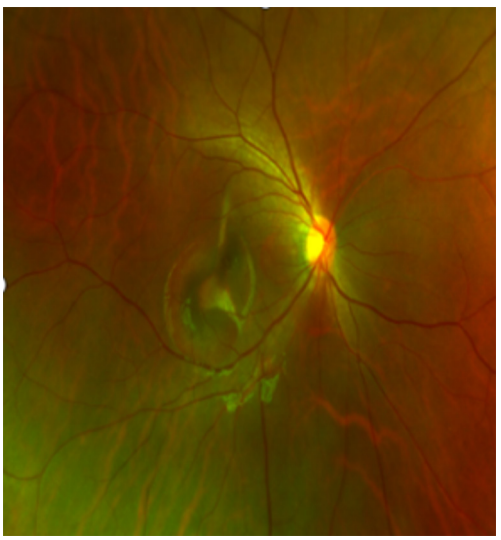

图1 黄斑区见一不连续弧形黄白色病灶，其周围见类圆形出血性神经上皮脱离

Figure 1 A discontinuous curved yellow-white lesion with quasi-circular hemorrhagic neuroepithelial detachment is seen around the macular area

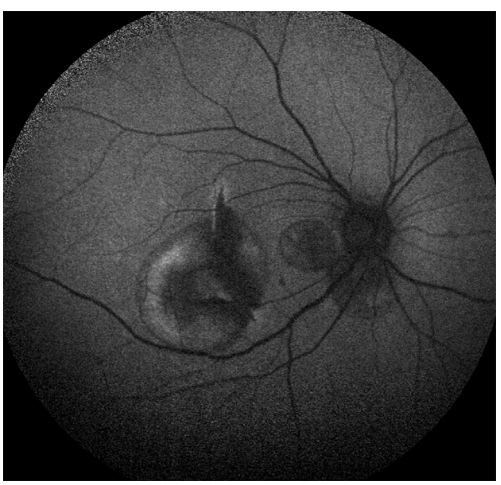

图2 自发荧光显示黄斑区多个弧形呈低荧光病灶，黄斑区血性神经上皮脱离，部分因血液引起强荧光

Figure 2 Autofluorescence shows multiple arc-shaped low-fluorescence lesions in the macular area, macular dehemorrhagic neuroepithelial detachment, partly caused by blood

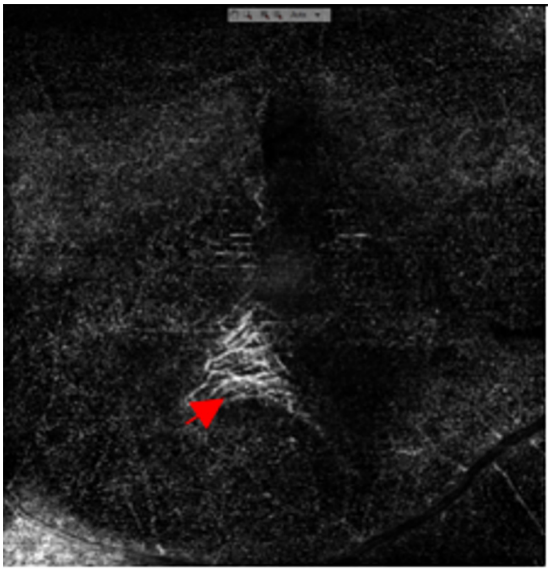

图3 OCTA黄斑区无血管层见两处弧形低密度区及网状CNV结构(箭头)

Figure 3 In the vascularized layer of OCTA macular area, there were two curved low density areas and reticular CNV structure (arrow)

图4 OCTA脉络膜毛细血管层见经过黄斑中央脉络膜毛细血管弧形缺损区，暴露其下脉络膜中层血管

Figure 4 Choroidal capillary layer of OCTA showed the arc-shaped defect area of choroidal capillaries passing through the central macular, exposing the inferior choroidal middle vessels

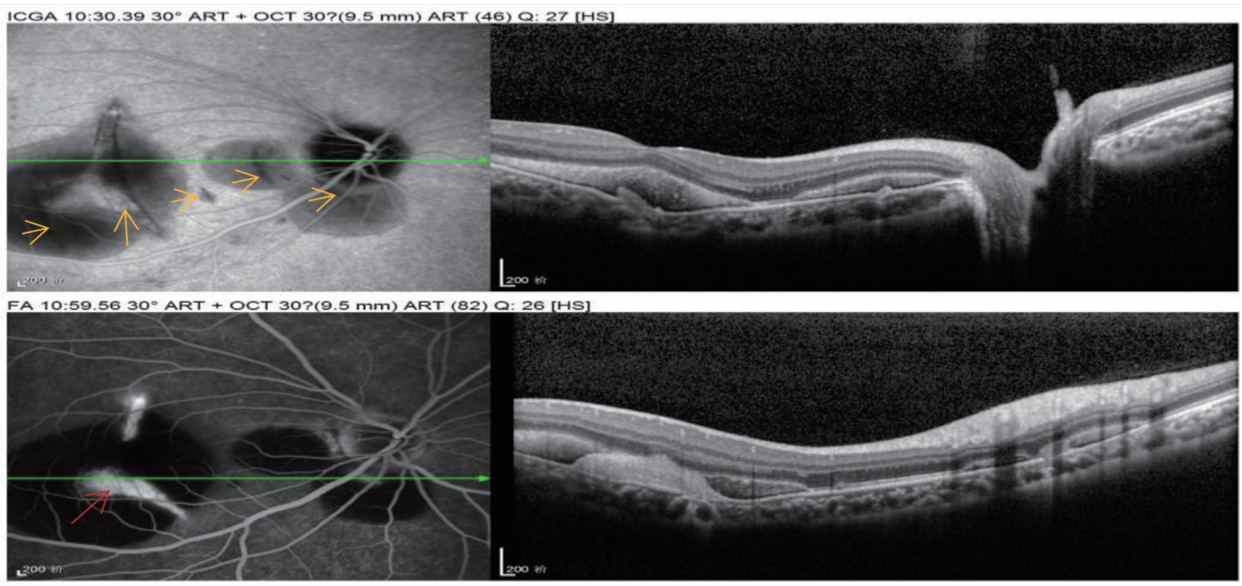

图5 ICGA对弧形病灶内强荧光显示得很清晰，视盘周围还可见好几处弧形病灶(黄色箭头)，对应OCT见RPE及脉络膜毛细血管凹陷，凹陷处见中高反射瘢痕信号，FFA可见弧形染色并且中央有楔形荧光渗漏，考虑有CNV(红色箭头)

Figure 5 ICGA clearly showed strong fluorescence in curved lesions, and several curved lesions (yellow arrows) could be seen around the optic disc. Corresponding to OCT, there were RPE and choroidal capillary depressions, moderate and high reflection scar signals in depressions, arc staining in FFA and wedge-shaped fluorescence leakage in the center. CNV (red arrow) was considered

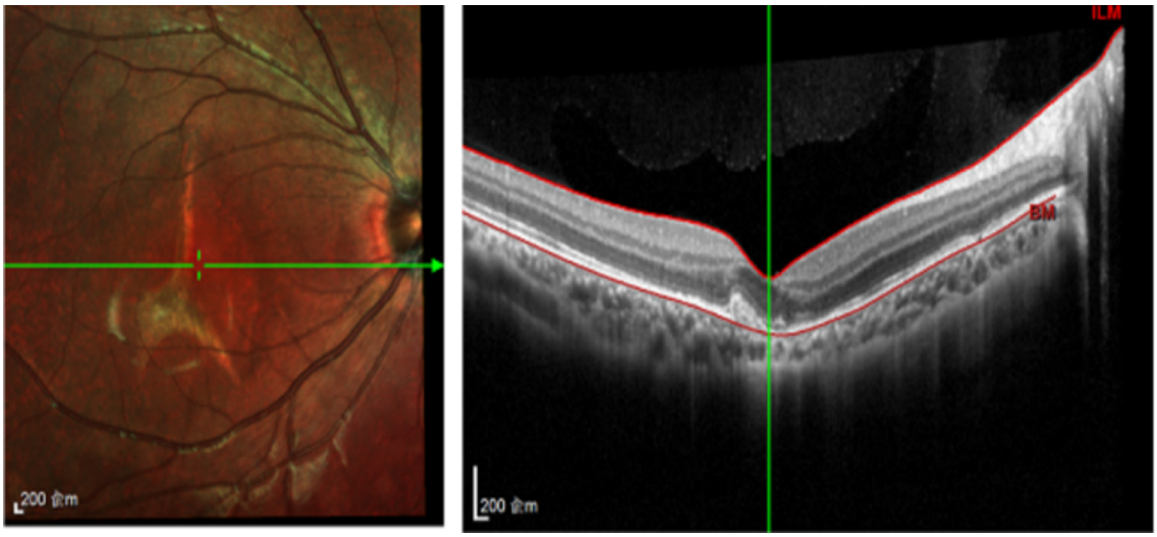

图6 玻璃体内抗VEGF注射1个月后复查，视网膜下出血吸收，视力提高到0.6

Figure 6 One month after intravitreal anti-VEGF injection, the subretinal hemorrhage was absorbed and the visual acuity was improved to 0.6

2 讨论

在没有外伤或顿挫伤的情况下出现如此严重的脉络膜破裂伤，似乎很难解释。为了阐明病因，查找文献发现有些疾病会有类似脉络膜破裂的表现。一是高度近视高度近视漆裂纹改变，在OCT上表现为RPE-bruch膜缺失及强反射、脉络膜组织变薄[7]，在其早期阶段可出现视网膜下出血或CNV[8]。本病例并非高度近视，眼底未见漆裂纹改变，因此暂时不考虑该病可能。二是眼底血管条纹，血管条纹是Bruch膜断裂，表现为以视盘或者黄斑处呈放射状的红褐色条纹，容易并发CNV[9-10]，同时因为脉络膜毛细血管脆性增加，钝挫伤后容易引起破裂出血[11]。血管条纹可以与不同的全身疾病相关，例如假性黄色瘤(pseudoxanthoma elasticum，PXE)、Paget疾病、Ehlers-danlos综合征、血管基因症或胶原蛋白疾病[9]。无赤光和自发荧光对早期的血管条纹有较好的敏感性，可以发现视盘或者黄斑条纹状改变[9]，本病例自发荧光除了脉络膜破裂灶，自发荧光及无赤光，甚至FA和ICGA均没有发现血管条纹改变，似乎也可以排除血管条纹。继续查找文献发现Oztas等[6]发现1例儿童受到轻微创伤后出血视网膜下出血及脉络膜破裂，并做了病例报道。病例中提到12岁男孩，被母亲拇指弹伤后出现脉络膜破裂、黄斑视网膜下出血，并且通过检查任一眼均无可见的血管样条纹改变，但该研究发现患者颈部和肚周皮肤有“拔鸡毛外观”，且对皮肤进行活检显示网状真皮的碎片，短而波浪状的不规则弹性纤维，考虑该患者是隐匿性PXE。

PXE是一种罕见的常染色体隐性遗传性多系统疾病，主要影响皮肤、心血管系统和视网膜，由16p13.1号染色体上ABCC6基因的突变引起[12]。其特征是受影响的组织缓慢，进行性异常钙化和弹性纤维断裂。儿童或年轻人中PXE的表现通常仅限于细微、无症状的皮肤病变，而典型的皮肤病变，如淡黄色丘疹，可能会凝结成斑块，使皮肤在颈部和弯曲区域出现“拔毛鸡”的外观[13]。心血管方面，通过钙沉积和中型动脉弹性层的变性引起心血管事件，这些变化可能导致间歇性跛行、肾性高血压、心绞痛、脑卒中、二尖瓣脱垂、心肌梗塞甚至心脏死亡[14]，尽管Oztas等[6]报道的病例心血管检查和心电图检查正常，但仍跟踪发现了严重的心脏表现。胃肠道方面，有学者[15]发现儿童期PXE会由于动脉壁中的弹性纤维变性而导致胃肠道出血。PXE的眼部受累可能是轻度到重度，轻度无肉眼可见的异常，重度的特征性病变为棕褐色外观，有细小、黄色、玻璃疣样的

色素不规则，最主要出现在黄斑颞侧[13,16]。PXE患者头部震荡或眼钝挫伤中导致钙化和易碎的Bruch膜破裂[17]。这类患者在轻微的眼外伤或者头部剧烈震动下可诱发脉络膜破裂和视网膜下出血，避免暴力和接触性运动及其他活动有眼外伤的风险。虽然本例患者皮肤无“拔鸡毛”外观，且由于患者及其家属拒绝行基因检测及皮肤活检，无法提供基因检测和皮肤活检结果，仍考虑假性黄色瘤可能性大。

再次详细回顾患者基本情况，了解到患者是体校学生，常打篮球。而有研究[18-19]发现体育活动，尤其是使用球的运动，是引起脉络膜破裂的最常见原因。虽然没有基因检测及皮肤活检结果，经过文献复习认为患者在有隐匿性PXE基础上，在打篮球过程中发生过头部撞击或者震荡导致脉络膜破裂可能性大，而因为有CNV形成，予抗VEGF治疗，虽然目前CNV已经疤痕化，但仍需要长期随访，研究支持抗VENG的PRN治疗方案是合理的[20]，嘱咐患者定期检查，重点关注是否有心血管系统疾病和胃肠道出血，建议定期自查Amsler表，同时注意避免暴力和接触性运动及其他有眼外伤风险的活动。

开放获取声明

参考文献

1. Patel MM, Chee YE, Eliott D. Choroidal rupture: a review[J]. Int Ophthalmol Clin, 2013, 53(4): 69-78.
2. Williams DF, Mieler WF, Williams GA. Posterior segment manifestations of ocular trauma[J]. Retina, 1990, 10(Suppl 1): S35-S44.
3. Raman SV, Desai UR, Anderson S, et al. Visual prognosis in patients with traumatic choroidal rupture[J]. Can J Ophthalmol, 2004, 39(3): 260-266.
4. Venkatesh R, Bavaharan B, Yadav NK. Predictors for choroidal neovascular membrane formation and visual outcome following blunt ocular trauma[J]. Ther Adv Ophthalmol, 2019, 11: 2515841419852011.
5. Dawson SL, Hirsch CS, Lucas FV, et al. The contrecoup phenomenon. Reappraisal of a classic problem[J]. Hum Pathol, 1980, 11(2): 155-166.
6. Oztas Z, Karadeniz C, Afrashi F, et al. Minor trauma resulting in subretinal haemorrhage with choroidal rupture: a case of subtle pseudoxanthoma elasticum in a child[J]. Clin Exp Optom, 2016, 99(1): 84-86.
7. Liu CF, Liu L, Lai CC, et al. Multimodal imaging including spectral-domain optical coherence tomography and confocal near-infrared reflectance for characterization of lacquer cracks in highly myopic eyes[J]. Eye (Lond), 2014, 28(12): 1437-1445.
8. Xu X, Fang Y, Uramoto K, et al. Clinical features of lacquer cracks in eyes with pathologic myopia[J]. Retina, 2019, 39(7): 1265-1277.
9. Chatziralli I, Saitakis G, Dimitriou E, et al. ANGIOID STREAKS: A comprehensive review from pathophysiology to treatment[J]. Retina, 2019, 39(1): 1-11.
10. Marchese A, Parravano M, Rabiolo A, et al. Optical coherence tomography analysis of evolution of Bruch's membrane features in angioid streaks[J]. Eye (Lond), 2017, 31(11): 1600-1605.
11. Pandolfo A, Verrastro G, Piccolino FC. Retinal hemorrhages following indirect ocular trauma in a patient with angioid streaks[J]. Retina, 2002, 22(6): 830-831.
12. Hu X, Plomp AS, van Soest S, et al. Pseudoxanthoma elasticum: a clinical, histopathological, and molecular update[J]. Surv Ophthalmol, 2003, 48(4): 424-438.
13. Finger RP, Charbel Issa P, Ladewig MS, et al. Pseudoxanthoma elasticum: genetics, clinical manifestations and therapeutic approaches[J]. Surv Ophthalmol, 2009, 54(2): 272-285.
14. Sherer DW, Sapadin AN, Lebwohl MG. Pseudoxanthoma elasticum: an update[J]. Dermatology, 1999, 199(1): 3-7.
15. Antiga E, Melani L, Caproni M, et al. An unusual cause of gastrointestinal bleeding in a young girl[J]. CMAJ, 2006, 175(6): 583.
16. Georgalas I, Tservakis I, Papaconstaninou D, et al. Pseudoxanthoma elasticum, ocular manifestations, complications and treatment[J]. Clin Exp Optom, 2011, 94(2): 169-180.
17. Naouri M, Boisseau C, Bonicel P, et al. Manifestations of pseudoxanthoma elasticum in childhood[J]. Br J Dermatol, 2009,

161(3): 635-639.

18. Ament CS, Zacks DN, Lane AM, et al. Predictors of visual outcome and choroidal neovascular membrane formation after traumatic choroidal rupture[J]. Arch Ophthalmol, 2006, 124(7): 957-966.
19. Wood CM, Richardson J. Indirect choroidal ruptures: aetiological factors, patterns of ocular damage, and final visual outcome[J]. Br J Ophthalmol, 1990, 74(4): 208-211.
20. Lupidi M, Muzi A, Castellucci G, et al. The choroidal rupture: current concepts and insights[J]. Surv Ophthalmol, 2021, 66(5): 761-770.

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