白化病并发视网膜脱离1例报道

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【摘要】 眼皮肤白化病(oculocutaneous albinism,OCA)患者由于虹膜、脉络膜等缺乏黑色素,一旦发生视网膜脱离,诊断和手术干预极其困难。OCA患者眼底黑色素缺失,"红色眼底"缺乏对比度,视网膜组织难以辨认,无论是眼底检查还是手术操作都非常困难。术后视网膜愈合时间长,处理并发症需要非常谨慎。本文对1例白化病患者孔源性视网膜脱离的发病、诊断及治疗过程进行报道。患者经过治疗,视网膜成功复位,随访3个月无复发。因为发生继发性青光眼,视神经损害,预后视力差。本例提示,对于OCA患者的视网膜脱离,尽管通过手术使视网膜成功复位,但预后视力仍不佳,因此需要继续探索更为有效的诊治策略。

【关键词】 眼皮肤白化病(OCA); 黑色素缺失; 视网膜脱离; 手术; 继发性青光眼

【中图分类号】 R774.1+2,R758.5+4 【文献标志码】 B doi:10.3969/j.issn.1672-8467.2022.05.030

Albinism complicated with retinal detachment: a case report

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【**Abstract**】 For the lack of pigment in oculocutaneous albinism (OCA), especially in the iris and choroid, the diagnosis and surgical intervention of retinal detachment in albinism patients is extremely difficult. Because of the loss of melanin in the fundus of OCA patients, the "red fundus" lacks contrast, and the retinal tissue is difficult to recognize, both fundus examination and surgical operation are very difficult. Due to the long time of postoperative retinal healing, the management of complications also needs to be very careful. This report introduces the pathogenesis, diagnosis and treatment of rhegmatogenous retinal detachment in an albinism patient. With treatment, the patient's retina was successfully reattached and there was no recurrence during a 3-month follow-up. However, due to secondary glaucoma optic nerve damage, the patient ended up with a poor prognosis. The patient was eventually assessed for disability. This case suggests that the therapeutic effect of retinal detachment in albinism patients is still not satisfactory despite the most careful surgical treatment. In the future, clinicians should continue to explore more effective diagnosis and treatment strategies.

【**Key words**】 oculocutaneous albinism (OCA); absence of melanin; retinal detachment; operation; secondary glaucoma

*** This work was supported by the Talent Training Program of Shanghai Pudong Hospital (PY201901) and the Innovation Fund for Clinical Medicine of Zhangjiang Branch, Fudan University (KP7202107).**

眼皮肤白化病(oculocutaneous albinism,OCA)是由黑色素细胞中黑色素生物合成完全缺乏或减少导致头发、皮肤和眼部色素减退的一种常染色体隐性遗传疾病[1]。OCA患者的眼部表现

上海市浦东医院院级人才培养项目(PY201901);复旦张江临床医学创新基金(KP7202107)

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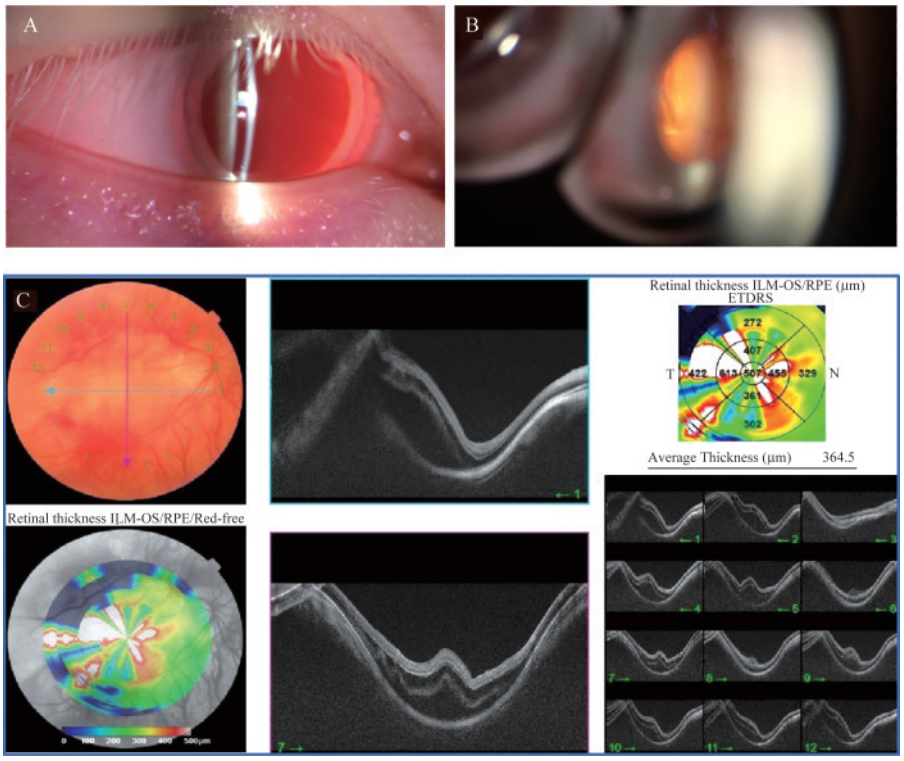
网络首发时间:2022-09-22 20:43:37 网络首发地址:https://kns.cnki.net/kcms/detail/31.1885.r.20220921.1328.004.html

主要为不同程度的先天性眼球震颤、虹膜色素减退、视网膜色素上皮色素减少等。由于眼底黑色素缺失，“红色眼底”缺乏对比度，眼底结构难以辨认，眼底检查难度极大。当OCA患者发生视网膜脱离，术前视网膜裂孔的辨认和术中玻璃体后脱离、玻璃体切除、视网膜激光光凝等都很困难，术后视网膜愈合时间长，处理并发症需要非常谨慎，对眼科医师极具挑战性。本文报告1例白化病患者孔源性视网膜脱离（rhegmatogenous retinal detachment，RRD）的发病、诊断及治疗过程，旨在进一步增加对疾病的认识，为今后的诊治提供参考。

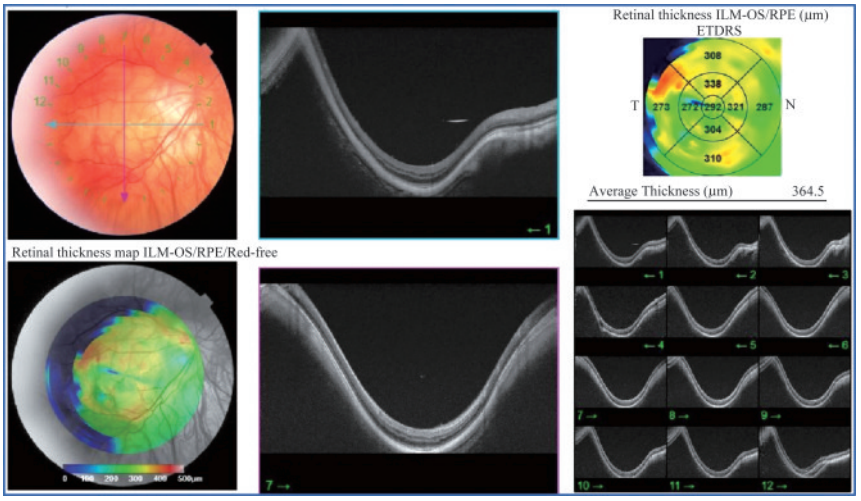
病例资料 男性患者，41岁，因“右眼突发眼前黑幕遮挡1周”，于2021年9月下旬就诊于上海市浦东医院眼科。既往病史：自幼左眼视力不佳（具体原因不详）；出生时诊断为白化病。入院检查：右眼视力0.15，矫正无助，左眼视力0.1，矫正无助；右眼眼压16 mmHg（1 mmHg=0.133 kPa），左眼眼压17 mmHg；双眼眼球水平震颤，不能固视；双眼眼睑皮肤及睫毛呈白色，角膜透明；前房轴深约3.5 CT，周边前房1/3 CT；房水清，虹膜透见红光，晶状体明，眼底色素缺失，脉络膜血管清晰可见，透见白色巩膜，视网膜结构分辨不清；右眼10～12点钟位隐约见视网膜隆起，累及黄斑区，10点位可见一疑似马蹄形裂孔（图1），光学相干断层扫描（optical coherence tomography，OCT）提示右眼黄斑区神经上皮层隆起。入院诊断：（1）右眼视网膜脱离（孔源性可能）；（2）左眼弱视；（3）眼皮肤白化病。入院后予右眼视网膜内路复位术，术中见11点钟位视网膜有一裂孔，行玻璃体切除，复位视网膜后予以CO_2冷冻裂孔及周围区域，气液交换，眼内填充硅油，术毕。术后1周复诊，查眼底见术眼上方少许视网膜下液，眼压27 mmHg。此后每周复诊，眼压波动在28～34 mmHg，视网膜下液体逐渐吸收，视网膜平伏（图2），期间予以强化局部降眼压药物治疗，具体方案：卡替洛尔滴眼液滴右眼（bid）；布林佐胺滴眼液滴右眼（bid）；溴莫尼定滴眼液滴右眼（bid）；妥布霉素地塞米松滴眼液滴右眼（qid）。术后3个月取出眼内硅油，右眼视力0.05，矫正无助，眼压27 mmHg，视网膜复位良好（图3），视盘色淡。



A：Anterior segment；B：One horseshoe hole in the upper temporal retina；C：OCT suggested detachment of the macular retinal nerve epithelium. The palpebral skin，iris，fundus and other parts of the plain color was missing. A suspected horseshoe hole and retinal eminence were seen in the peripheral retina at 10：00.

图1 患者就诊时右眼的情况

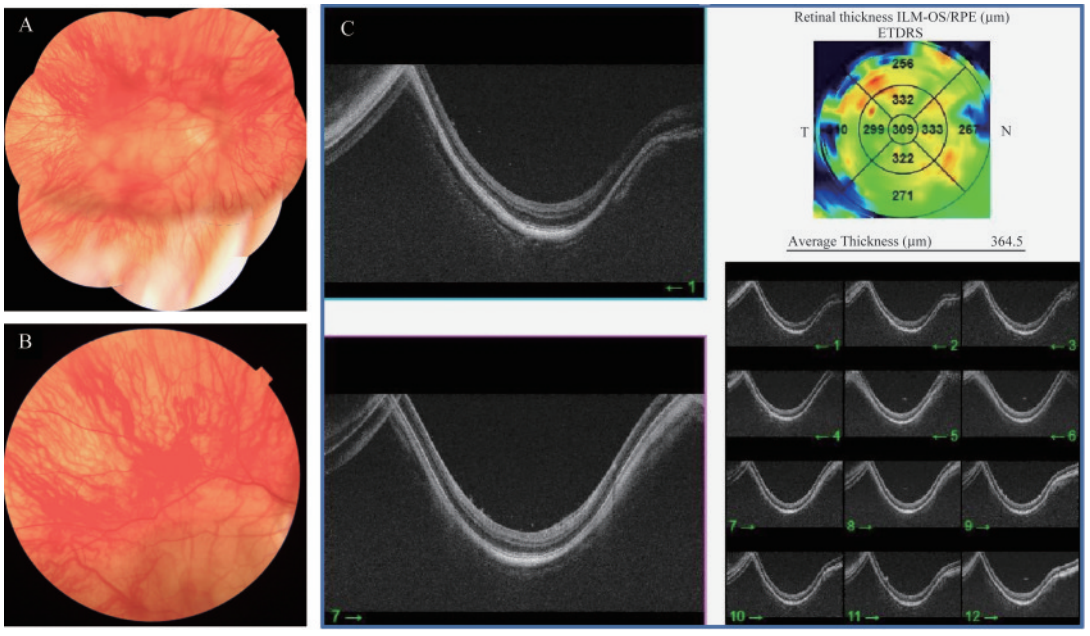
Fig 1 Initial images of the patient's right eye



The retinal nerve epithelium in macular area was well reset and the silicone oil interface was seen above.

图2 右眼术后1周的OCT检查结果

Fig 2 Results of OCT examination of the right eye 1 week after surgery



A: Fundus images from nine directions; B: Superior temporal retinal hole is difficult to recognize; C: OCT showed reduction of the thickness in retinal nerve epithelium after oil extraction. The original horseshoe hole was difficult to see, the retina was well attached, and the optic disc was pale.

图3 右眼硅油取出后的眼底表现

Fig 3 Fundus of the right eye after silicone oil removal

讨论 OCA是一组黑色素生物合成障碍的遗传性疾病[1],特征是头发、皮肤和眼睛的色素沉着减少。眼部临床表现包括:不同程度的先天性眼球震颤、虹膜色素减退和呈半透明状、视网膜上皮色素减少、中心凹发育不全、视力下降(通常为20/60至20/400)、屈光不正、色觉障碍、畏光、斜视、立体视觉降低等[2]。OCA患者具有正常寿命、智力和生理功能,使其获得有用视力,确保生活质量是诊疗的终极目标。

RRD在此类患者中非常罕见[3],关于这种罕见临床情况的大规模病例报道及研究较少。OCA患者会发生眼球震颤,快速眼球扫视运动会使液化玻璃体产生连续运动,从而导致早发性玻璃体后脱离(posterior vitreous detachment,PVD)发生率较高[4]。本例进一步补充了该领域的研究,深入了解OCA患者RRD的特点和治疗困难,以及对于治疗目标和治疗策略的合理判断。此类患者的视网膜复位手术难度较大,手术操作需要引起注意[5-7]。

OCA患者眼底黑色素缺失,“红色眼底”缺乏对比度,一旦发生RRD,术前和术中识别视网膜裂孔是第一个难点,视网膜裂孔在视网膜复位之后也难以辨认;术中行玻璃体后脱离、玻璃体切除切除、视网膜激光光凝等非常困难,即使是经验丰富的术者在操作过程中也可能发生医源性裂孔。

视网膜激光光凝以及冷冻是封闭视网膜裂孔的常用方法[8]。大多数激光光凝技术都依赖于视网膜色素上皮或脉络膜中色素的吸收。由于黑色素缺乏,OCA患者的视网膜难以通过激光光凝产生凝结反应而封闭裂孔。有研究提出,通过提高激光功率和增加持续时间使脉络膜毛细血管内的血红蛋白凝固,并间接导致OCA患者的视网膜粘连效应[9]。总之,在缺乏色素的眼睛中产生脉络膜视网膜粘连的效果很困难。本例通过对眼球壁组织进行冷冻,达到视网膜神经上皮层与视网膜色素上皮层的粘连,成功封闭视网膜裂孔。

本例患者经过治疗,视网膜成功复位,但术后出现继发性青光眼,考虑与以下因素有关:(1)术后炎症反应重,炎症刺激导致小梁网功能下降;(2)术中使用曲安奈德,术后可能有残留,导致眼压升高;(3)眼内硅油填充时间长,硅油乳化后堵塞小梁网,合并硅油滴对小梁网毒性损伤;(4)术后使用糖皮质激素滴眼液,对于糖皮质激素敏感个体,可能造成激素性青光眼。尽管采用联合降眼压药物治疗,眼压依然无法控制到正常水平。眼科医师面临两难的选择:如果取出硅油可能复发网脱,如果继续保留硅油可能出现不可逆的青光眼视神经损害。患者在加强局部降眼压治疗后长期随访观察。术后3个月,眼底检查结果确定视网膜复位良好,取出眼内硅油。患者术眼最佳矫正视力0.15,眼压31 mmHg,对侧眼自幼弱视,视力只有0.05。

由于缺乏黑色素,OCA患者视网膜脱离的诊断和治疗难度极大,经典治疗方式不能完全适用。在治疗过程中,视网膜复位是主要的治疗目标。手术并发症处理策略的选择及治疗时机的判断均会对视力预后有极大的影响,对于复杂性视网膜脱离的治疗仍需继续探索。

作者贡献声明 李婷婷 资料收集,文献检索,论文撰写。沙倩 论文设计、指导和修改。

利益冲突声明 所有作者均声明不存在利益冲突。

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(收稿日期:2022-01-24; 编辑:段佳)