论著·临床医学

正常眼底型先天性静止性夜盲患儿的电生理特征与基因分析

齐冬梅*,霍姝佳*,周 序,余 涛 陆军军医大学(第三军医大学)第一附属医院眼科/西南医院眼科,重庆

［摘要］ **目的** 分析眼底正常型先天性静止性夜盲(congenital stationary night blindness,CSNB)患儿的电生理与基因检查结果。**方法** 回顾性分析2019年8月至2025年3月经临床检查及基因检测确诊为CSNB的儿童,共11例纳入研究。分析其全视野视网膜电图(full-field electroretinogram,ffERG)特征及基因检查结果。**结果** 11例患儿中男性10例,女性1例,年龄为(7.5±2.1)岁。患儿戴镜后矫正视力为0.48±0.11,验光结果中1例远视,余均为近视。眼底照相检查除高度近视表现为豹纹状改变外,余眼底未见明显异常。ffERG检查所有患儿暗视ERG 3.0/10.0的a波幅值正常或轻中度降低,b波振幅显著下降,b波和a波的振幅比<1,为负相波,属于CSNB Schubert-Bornschein波型。基因检测结果发现,11例患儿均携带致病性基因突变,其中6例为CACNA1F基因突变,2例为GPR179基因突变,2例为NYX基因突变,1例为GRM6基因突变。**结论** 对于眼底表现正常但戴镜矫正视力低于正常的患儿,应尽早进行ffERG检查,排查CSNB,并结合基因检查进行确诊,有助于评估疾病的遗传性及其预后,提高早期发现机会,减少过度的弱视训练和治疗。

［关键词］ 儿童;先天性静止性夜盲;眼底照相;视网膜电图;基因检测
［中图法分类号］ R394.1;R591.412;R770.43 ［文献标志码］ A

Electrophysiological characteristics and gene analysis of children with congenital stationary night blindness without fundus abnormalities

QI Dongmei, HUO Shujia, ZHOU Xu, YU Tao Department of Ophthalmology, First Affiliated Hospital/Southwest Hospital, Army Medical University (Third Military Medical University), Chongqing, China

［**Abstract**］ **Objective** To analyze the electrophysiological and genetic examination results of children with congenital stationary night blindness (CSNB) without fundus abnormalities. **Methods** A retrospective study was conducted on 11 children diagnosed with CSNB in our department from August 2019 to March 2025. Their full-field electroretinogram (ffERG) and the results of genetic testing were analyzed. **Results** Among the 11 patients, there were 10 males and 1 female, at a mean age of 7.5±2.1 years. In the optometry, 1 patient had hyperopia and the other had myopia, and all of them obtained a corrected visual acuity of 0.48±0.11 after wearing glasses. In fundus photography examination, except for the leopard pattern changes in high myopia, no obvious abnormalities were found in the remaining fundus. In ffERG examination for all patients, the amplitude of the a-wave of scotopic ERG 3.0/10.0 was normal or slightly to moderately decreased, the amplitude of the b-wave was decreased significantly, and the amplitude ratio of the b-wave to the a-wave was <1, showing a negative-phase wave, which belonged to the CSNB Schubert-Bornschein

* 同等贡献者

［通信作者］ 余 涛,E-mail:yt423@tmmu.edu.cn

2097-0927/Copyright © 2025, 陆军军医大学(第三军医大学)。这篇开放获取文章遵循 CC BY 许可协议 (https://creativecommons.org/licenses/by/4.0/)

引用格式:齐冬梅,霍姝佳,周序,等. 正常眼底型先天性静止性夜盲患儿的电生理特征与基因分析[J]. 陆军军医大学学报,2025,47(20):2540-2547. DOI:10.16016/j.2097-0927.202507102.

Citation:QI D M, HUO S J, ZHOU X, *et al*. Electrophysiological characteristics and gene analysis of children with congenital stationary night blindness without fundus abnormalities[J]. J Army Med Univ,2025,47(20):2540-2547. DOI:10.16016/j.2097-0927.202507102.

waveform. The results of genetic test revealed that all the 11 patients carried pathogenic mutations, including 6 cases with CACNA1F mutations, 2 cases with GPR179 mutations, 2 cases with NYX mutations, and 1 case with GRM6 mutation. **Conclusion** For children with normal fundus manifestations but corrected visual acuity lower than normal after wearing glasses, ffERG examination should be carried out as soon as possible to screen for CSNB, and a diagnosis should be made in combination with genetic testing. This is helpful for evaluating the heredity and prognosis of the disease, increasing the chance of early detection, and reducing excessive amblyopia training and treatment.

[**Key words**] children; congenital stationary night blindness; fundus photography; electroretinogram; genetic testing

QI Dongmei and HUO Shujia contributed equally to the article. Corresponding author: YU Tao, E-mail: yt423@tmmu.edu.cn

先天性静止性夜盲(congenital stationary night blindness,CSNB)是一种罕见的遗传性眼部疾病,以先天性非进行性夜盲为临床特点。患者均自幼发病,典型的临床表现为视力不同程度低下,合并夜盲、眼球震颤、斜视[1]。在一些典型病例中,CSNB患者眼底可能观察到视网膜或脉络膜色素沉着。但大部分儿童眼部检查难以发现异常,主要以视力差、屈光不正为临床表现,因此该病容易被误诊为弱视,导致不必要的治疗或延误干预。目前,对该病具有诊断价值的特征性改变是全视野视网膜电图(full-field electroretinogram,ffERG)异常,其主要特征为:杆体ERG无波、暗视ERG a波正常或降低、b波振幅下降,部分患者明视ERG也表现异常,表明锥体系统功能也受累[2]。根据CSNB临床表现,分为眼底正常的CSNB和眼底异常的CSNB。眼底异常的CSNB包括小口病(oguchi disease, OD)、白点状眼底[3]。眼底正常型CSNB根据ffERG改变,分为Riggs型和Schubert-Bornshein型,Schubert-Bornschein型根据视杆细胞是否存在功能,又分为1型即完全型CSNB(complete form of CSNB, cCSNB)和2型不完全型CSNB(incomplete form of CSNB, iCSNB)。CSNB具有高度遗传异质性,涉及多个致病基因,不同基因突变可能导致不同的临床亚型。因此,基因检测在CSNB的精准诊断、分型及遗传咨询中具有重要价值,尤其是当临床表现不典型或家族史不明确时。本研究回顾性分析了11例眼底正常型CSNB患儿的电生理与基因检查结果,通过整合临床表现、电生理与基因检测数据,旨在提高对儿童CSNB的认知和诊断水平,进而为推动个性化诊疗及遗传咨询提供依据。

1 资料与方法

1.1 一般资料

本研究为回顾性研究,选择2019年8月至2025年3月在陆军军医大学第一附属医院眼科检查,经ffERG及基因报告确诊为CSNB的儿童共11例。其中男性10例,女性1例,年龄2~9(7.5±2.1)岁。

1.2 方法

记录所有患儿的症状,患儿均接受裸眼视力、眼压、验光,以及裂隙灯显微镜、眼底彩照、黄斑OCT、ffERG和基因全外显子测序(whole exome sequencing, WES)。WES由上海韦翰斯生物医药科技有限公司完成。参照美国医学遗传学和基因组学学会(American College of Medical Genetics and Genomics, ACMG)相关指南对基因变异进行致病性评价。本研究已通过陆军军医大学第一附属医院伦理委员会审查[批件号:(B)KY2025101]。研究人员向每位父母/监护人解释研究目的、程序及潜在风险和益处,并获得书面知情同意。

2 结果

2.1 视力与屈光

所有患儿的主要症状表现为视力下降或是体检时发现视力差。患儿的初次矫正视力为0.26±0.16(0.05~0.50),验光结果中1例远视,余均为近视。低度近视患儿2例,中度近视患儿3例,高度近视患儿5例。经戴镜治疗半年后复查时,有4例患儿视力无改善,余均有一定提高,矫正视力为0.48±0.11(0.3~0.6),但与正常年龄段儿童比较,视力仍低于正常值(表1)。

cCSNB的患儿最佳矫正视力(best-corrected visual acuity,BCVA)(戴镜后半年)为0.4~0.6(0.52±0.13),屈光状态为-9.0D~-3.0D,平均为-6.95D。iCSNB的患儿BCVA(戴镜后半年)为0.3~0.6(0.43±0.08),屈光状态为-8.25D~+2.0D,平均为-2.96D。

2.2 眼部检查

裂隙灯检查:所有患儿的角膜、虹膜、晶状体未见

表1 患儿基本情况、基因检测以及ffERG分型

Table 1 Demographics, genetic results and ffERG findings in the patients

No.	Sex	Age/years	PxSx	RS	BCVA	BCVA (6mo f/u)	FP	Macular OCT	ffERG class	GF/IP
1	M	6	Decreased vision	(-)1.25ds-3.00dc (-)0.50ds-3.00dc	0.2/0.3	0.5/0.6	Normal	Normal	iCSNB	CACNA1F/XL
2	M	8	Poor vision	(-)2.00ds-3.25dc (-)1.25ds-3.75dc	0.5/0.5	0.6/0.7	Normal	Normal	iCSNB	CACNA1F/XL
3	M	4	Poor vision	(-)7.75ds-2.75dc (-)8.25ds-3.50dc	0.05/0.05	0.3/0.3	Tigroid fundus	Normal	iCSNB	CACNA1F/XL
4	M	4	Poor vision	(-)3.50ds-3.00dc (-)3.00ds-3.50dc	0.4/0.4	0.4/0.4	Normal	Normal	iCSNB	CACNA1F/XL
5	M	9	Poor vision	(-)3.75ds-0.50dc (-)4.75ds-1.00dc	0.3/0.2	0.3/0.3	Normal	Normal	iCSNB	CACNA1F/XL
6	M	9	Nyctalopia	(+)1.75ds (+)2.00ds	0.5/0.5	0.5/0.6	Normal	Normal	iCSNB	CACNA1F/XL
7	M	3	Poor vision	(-)9.00ds-1.00dc (-)8.75ds-1.00dc	0.10/0.15	0.5/0.3	Tigroid fundus	Normal	cCSNB	NYX/XL
8	M	4	Poor vision	(-)5.75ds-1.00dc (-)6.50ds-0.75dc	0.4/0.4	0.4/0.4	Tigroid fundus	Normal	cCSNB	NYX/XL
9	M	6	Decreased vision	(-)3.00ds-1.00dc (-)3.00ds-0.50dc	0.20/0.15	0.5/0.3	Normal	Normal	cCSNB	GPR179/AR
10	F	4	Poor vision	(-)6.75ds-1.50dc (-)7.00ds-1.50dc	0.2/0.1	0.6/0.6	Tigroid fundus	Normal	cCSNB	GPR179/AR
11	M	2	Decreased vision	(-)8.50ds-1.25dc (-)9.25ds-1.25dc	0.2/0.2	0.6/0.6	Tigroid fundus	Normal	cCSNB	GRM6/AR

ffERG: Full-field electroretinogram; PxSx: Presenting symptoms; RS: Refractive status; BCVA: Best-corrected visual acuity; f/u: Follow up; FP: Fundus photography; GF: Genetic findings; IP: Inheritance patterns; iCSNB: Incomplete form of congenital stationary night blindness; cCSNB: Complete form of congenital stationary night blindness; XL: X-linked recessive; AR: Autosomal recessive; DS: Diopter sphere; DC: Diopter cylinder

异常,瞳孔光反射灵敏。眼底检查5例高度近视的患儿均有豹纹状改变,余眼底未见明显异常。

辅助检查:眼底照相中5例患儿为豹纹状改变,黄斑OCT检查所有患儿黄斑结构未见异常。ffERG检查所有患儿暗视3.0/10.0 ERG的a波正常或轻度降低,b波振幅显著下降,b波幅值和a波的幅值比<1,为负相波,属于Schubert-Bornschein波型,其中5例患儿暗适0.01不能诱发波形,为cCSNB,6例患儿为iCSNB(图1、2)。图1为6号患儿,9岁男性儿童,以夜盲就诊;图2为8号患儿,4岁男性儿童,以视力差就诊。

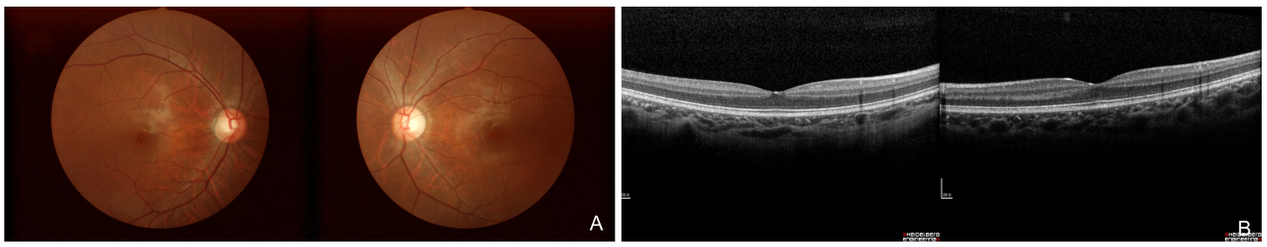
2.3 基因检测

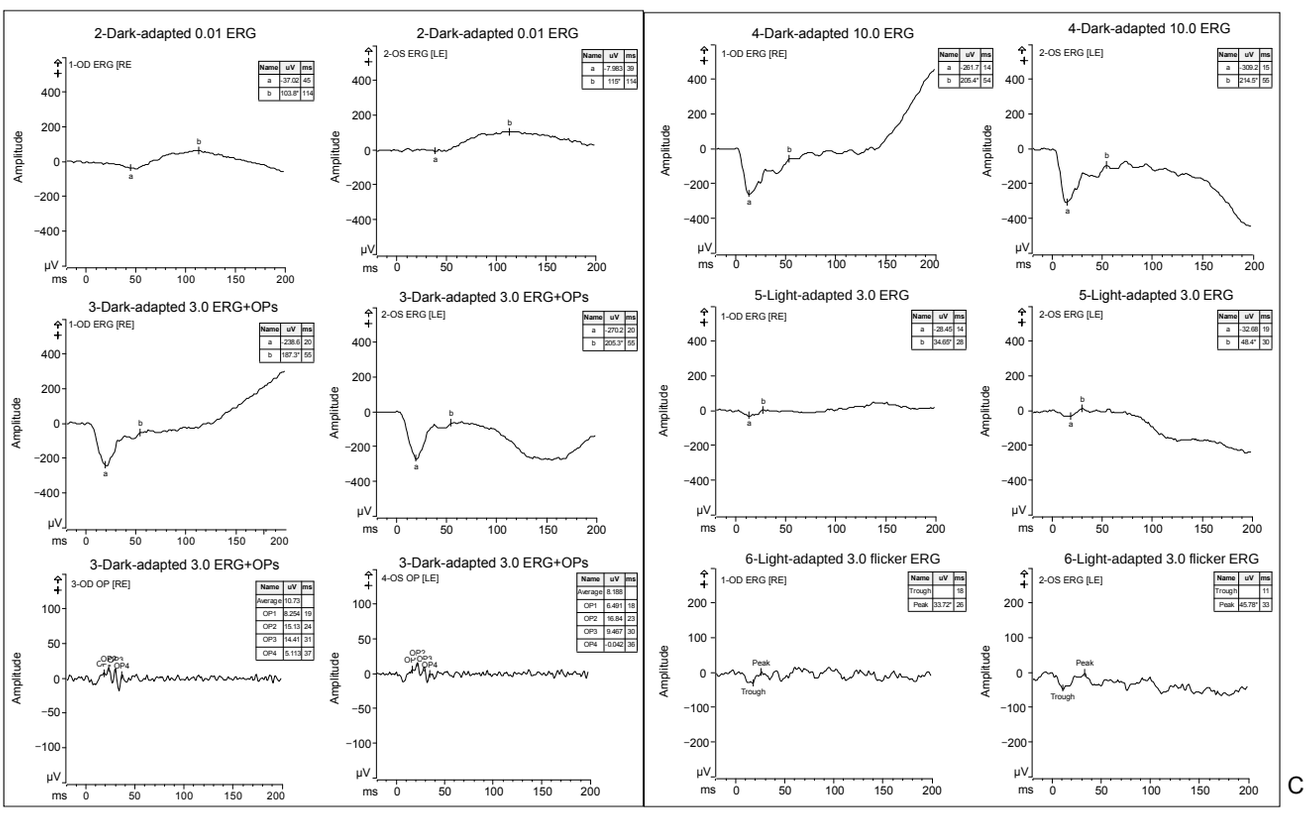
11例患儿中有6例为CACNA1F基因突变,该6例患儿ffERG检查均表现为不完全型CSNB。5例患儿ffERG均表现为完全型CSNB,其中2例为GPR179基因突变,2例为NYX基因,1例为GRM6基因(表1)。

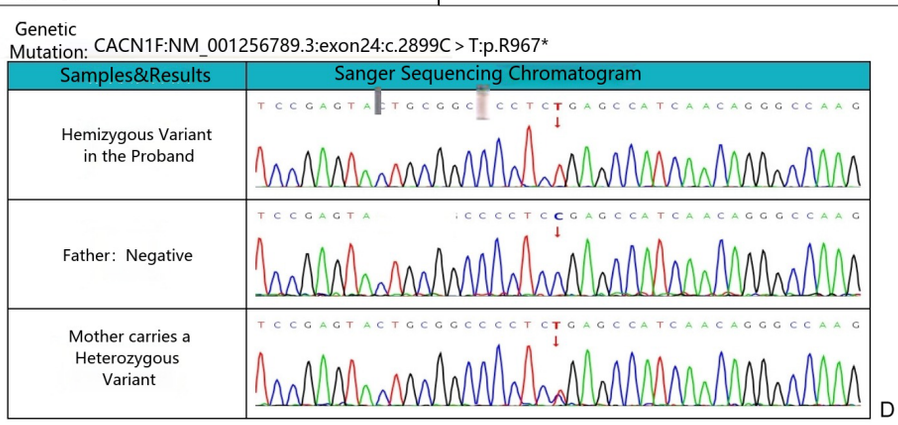
3 讨论

CSNB是一组具有遗传和临床异质性的疾病,主要影响视网膜光感受器内的信号处理。视网膜色素上皮(retinal pigment epithelium, RPE)中的视黄醇循环,经由视网膜双极细胞的信号传递,其临床特征包括自幼夜盲,可伴有近视、远视和视力下降,少数可合并眼球震颤和视乳头发育不良[4]。

眼底正常型CSNB患者以早发性近视为最常见临床表现,其次是斜视以及眼球震颤,眼部检查除发现近视相关改变外,其余眼部结构均正常,但在ffERG上有特征性表现[5]。根据ffERG特征可分为Riggs型和Schubert-Bornschein型两种类型[6]。Riggs型ffERG表现为暗反应a波振幅明显下降,伴b/a比值下降,呈负波形,患者主要表现为夜盲,通常视力无明显下降,不



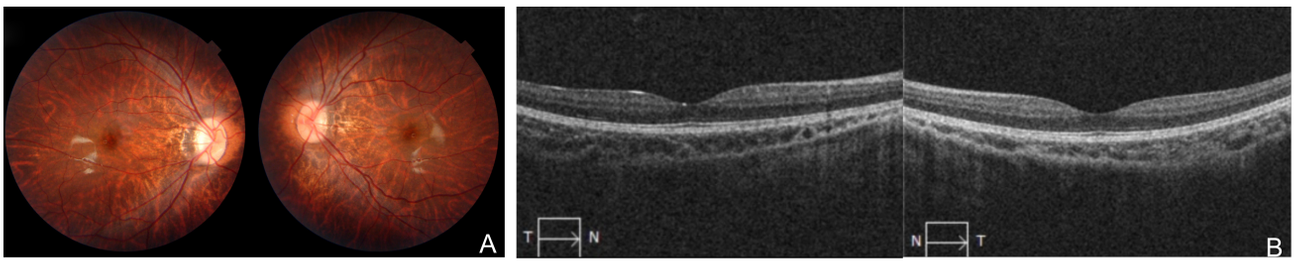


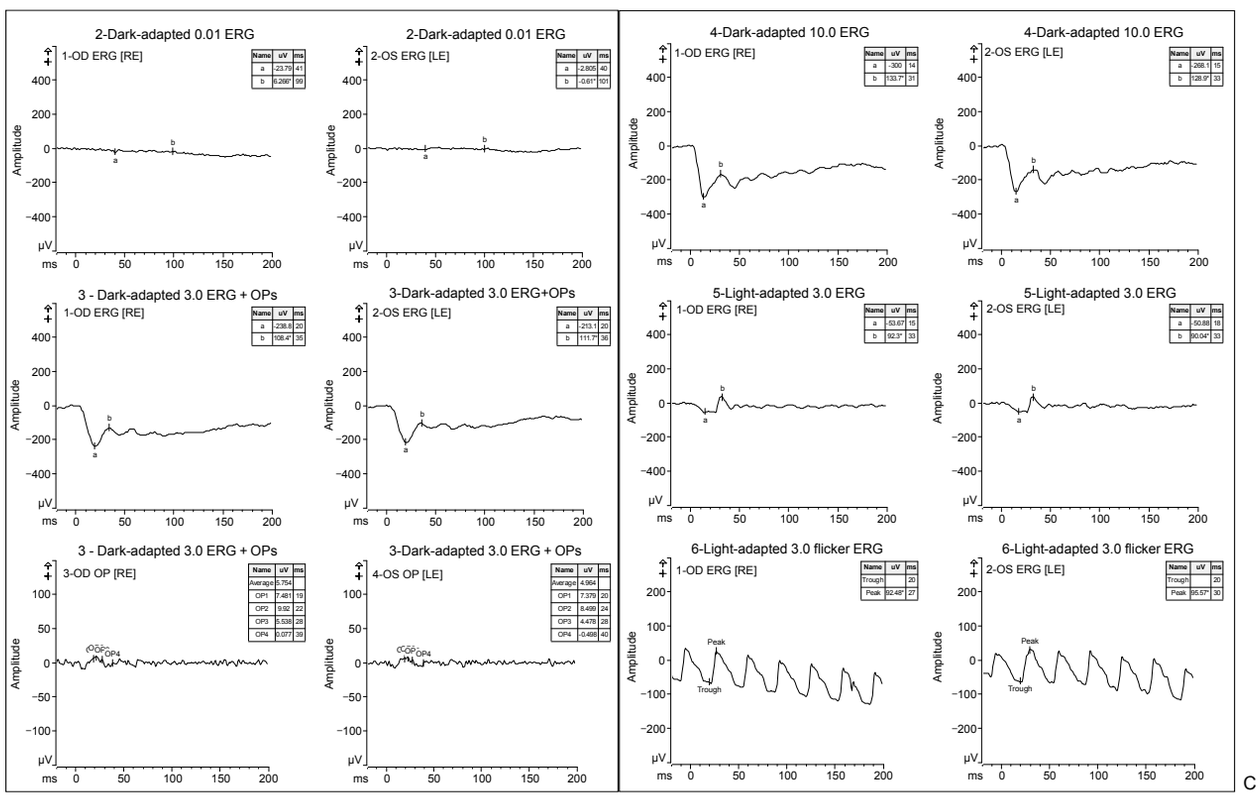


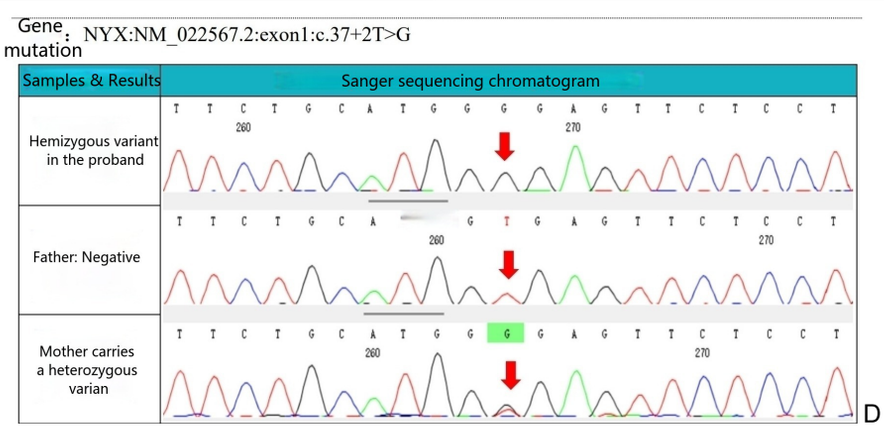
A: Representative fundus photogram showing no abnormalities; B: Unremarkable macular OCT; C: ffERG findings. The examination reveals reduced b-wave amplitudes under all scotopic conditions (dark-adapted 0.01, 3.0, and 10.0); The waveforms under scotopic 3.0 and 10.0 conditions shows a b/a ratio of less than 1, presenting as a negative waveform; These findings are consistent with iCSNB; D: Genetic testing report. Whole exome sequencing analysis reveals that the proband carries a hemizygous nonsense variant, c. 2899C>T (p. Arg967*), in the CACNA1F gene; This mutation in the CACNA1F gene is a known cause of iCSNB, which is consistent with the patient's ffERG findings

图1 iCSNB患儿的眼底照相、黄斑OCT、ffERG以及基因检查结果

Figure 1 Fundus photogram, macular optical coherence tomogram (OCT), full-field electroretinogram (ffERG) and result of genetic testing of an iCSNB patient







A: Representative fundus photogram showing a tessellated fundus; B: Unremarkable macular OCT; C: ffERG findings. Non-recordable b-wave under 0.01 dark-adapted conditions; The waveforms under scotopic 3.0 and 10.0 conditions shows a b/a ratio of less than 1, presenting as a negative waveform; Photopic responses are normal; Impression: These findings are consistent with cCSNB; D: Genetic testing report; Whole exome sequencing analysis revealed that a hemizygous canonical splice-site variant (c. 37+2T>G) in the NXYgene; This variant is predicted to disrupt normal mRNA splicing; Sanger sequencing confirmed the variant is authentic and has been maternally inherited; Mutations in this gene are a known cause of X-linked cCSNB; The genetic finding is entirely consistent with the patient's clinical presentation and ffERG results

图2 cCSNB患儿眼底照相、黄斑OCT、ffERG以及基因检查结果

Figure 2 Fundus photogram, macular optical coherence tomogram (OCT), full-field electroretinogram (ffERG) and results of genetic testing of a cCSNB patient

合并高度近视,眼球震颤,因此这类患者在临床上就诊率不高,极易被漏诊。Schubert-Bornschein型根据视杆细胞是否存在功能,又分为cCSNB和iCSNB。在典型病例中,代表视杆信号传导通路功能的ERG的b波在cCSNB中完全缺失,在iCSNB中波峰振幅降低减弱[7]。Schubert-Bornschein型是最常见类型,发生率可高达96.61%[8]。cCSNB患者通常有自幼夜盲史,伴有明显的视力下降,视力的中位数约为0.5,患者屈光状态主要特征为早发性中度至高度近视,文献报道100%的cCSNB为近视,且屈光的中位数为-7.4D[9]。而iCSNB患者有不同程度的屈光不正,从远视到近视,约16%的患者表现为远视,iCSNB患者的视力通常比cCSNB的患者更差,其视力中位数约为0.3[10]。本研究收集的11例眼底正常型CSNB患儿中,根据ffERG检查结果均属于Schubert-Bornschein型,其中cCSNB型5例,iCSNB型6例。所有患儿中有10例为近视,发生率为90.91%。11例患儿经戴镜治疗后,cCSNB的患儿BCVA为0.52±0.13,屈光状态平均为-6.95D。iCSNB的患儿BCVA为0.43±0.08,屈光状态平均为-2.96D。与iCSNB患儿进行比较,cCSNB患儿矫正视力稍好,近视程度更重。这些临床特点与文献报道一致。但是由于本研究患儿大多数年龄较小,对于症状描述不清,仅有1例以夜盲为首诊表现,其余均是以视力差就诊。所有患儿无眼球震颤表现,可能与患儿视力0.3以上有关系。

截至目前,共发现18个与CSNB相关致病基因[11]。在这些CSNB致病基因中以CACNA1F和NYX基因突变最为常见,两者共占到50%以上,其次为TRPM1[12]。cCSNB常见的致病基因为NYX、TRPM1、GRM6、GPR179、LRIT3,iCSNB常见的致病基因为CACNA1F、CACNA2、CABP4、RIMS2。按照遗传学方式CSNB可分为常染色体显性遗传(autosomal dominant,AD)、常染色体隐性遗传(autosomal recessive,AR)和X连锁隐性遗传(X-1inked recessive,XL)3种,大多数CSNB患者是按XL遗传模式进行遗传。本研究11例患儿中有8例为XL方式,占72.7%,突变的基因分别为CACNA1F 6例,NYX基因突变2例。3例为AR遗传方式,突变的基因为GPR179 2例,GRM6 1例。所有检测出突变的基因均为Schubert-Bornschein型中已报道的基因,与文献具有一致性。

引起iCSNB型致病基因主要是CACNA1F基因,约占90%[12]。本研究所检测的致病基因均为CACNA1F。CACNA1F位于人类染色体Xp11.23,编码Cav1.4离子通道的α1F亚基。α1F亚基缺失,可使光感受器细胞带状突触数量、形态以及功能发生异常,从而引发光感受器细胞与二级神经元之间视觉信号传导障碍,导致CSNB 2A型、AlandIsland眼病(Aland Island eye disease, AIED)、X连锁隐性遗传的视锥视杆细胞营养不良3型(Cone-rod Dystrophy, CORD)等疾病。三者具有较多共同临床特征,如视力下降、色觉缺陷、近视、眼球震颤、视网膜和视神经发育异常、ffERG负波形等[13-14]。AIED其特征为眼底色素沉着、视力下降、眼球震颤、散光、普罗坦色觉缺陷、进行性近视、暗适应不良,与CSNB 2A型有相似的临床特征,但主要区别在于黄斑发育不良[15]。CORD以视锥细胞受损为主,伴不同程度的视杆细胞损伤,ERG检查视锥细胞反应可呈熄灭型或明显下降。黄斑区视网膜外层光感受器受损,随年龄增长范围扩大,可累及周边部甚至全视网膜。本组6例患儿均表现为暗适b波重度降低,a波正常,b波和a波的振幅比<1,为负相波,明适损伤较暗适轻,同时所有患儿黄斑OCT检查未见异常,结合患儿临床表现及ffERG检查结果,故排除AIED和CORD,符合iCSNB。

引起cCSNB型致病基因有NYX、TRPM1、GRM6、GPR179、LRIT3。TRPM1最常见,约占70%。本研究所检测的致病基因有GPR179、NYX及GRM6,三者之间无明显的差异,主要原因为患者病例数较少。GPR179基因位于染色体17q12,功能实验表明GPR179基因敲除导致ffERG b波的振幅显著降低,表明该基因功能缺失可导致cCSNB[16-17]。NYX基因位于人类染色体Xp11.4,NYX在光感受器细胞、双极细胞、无长突细胞和神经节细胞中均有表达,其突变破坏了视网膜细胞间的连接,导致视杆、视锥细胞与ON-双极细胞间的信号传递发生异常,因此其突变导致cCSNB[18]。GRM6基因由10个外显子组成,位于5q35,是编码一种代谢型谷氨酸受体蛋白6型,主要在视网膜的双极细胞起作用,是cCSNB致病基因[19]。GPR179和GRM6遗传方式为AR,NYX为XL。该4例患儿的ffERG结果显示暗适b波均不能诱发波形,符合典型的cCSNB。结合基因检查结果,诊断为cCSNB。

本研究中11例患儿除1例以夜盲就诊外,均以视力下降、视力差就诊。所有患儿在经过正规戴镜治疗后,视力有提高,但BCVA仍低于相应年龄的视力。根据中国儿童弱视防治专家共识(2021年)[20],所有患儿无发生弱视的危险因素,因此排除弱视诊断。对于这部分眼底表现无明显异常、无弱视危险因素的患儿,应进一步完善电生理检查。ffERG是视网膜接受到全视野闪光刺激后,光感受器细胞产生的生物电反应总

和,主要反映视神经节细胞前整个视网膜的功能,在眼底病变的诊断中具有特定的临床意义。当ffERG提示杆状细胞功能障碍,而锥状细胞相对保存,应高度怀疑CSNB。同时必要的基因检测结果提高了CSNB的诊断准确率。明确的诊断可以帮助家长理解病症的遗传风险、疾病的传递方式及其预后,有助于评估疾病的遗传性及其对家庭的影响,提高早期发现和干预的机会。

由于眼底正常型CSNB患儿缺乏特异的临床表现,容易被误诊及漏诊。这部分患儿眼部结构无明显异常,且多伴有屈光不正,常常被误诊为弱视,被实施弱视训练,增加了不必要的家庭经济负担。因此,对于正规戴镜后随访矫正视力仍低于正常的患儿,除了常规眼科检查外,还应进行眼电生理检查,必要时行基因检测以明确诊断,为患儿提供个性化的治疗方案、遗传咨询及长期随访计划,从而改善患儿的生活质量并为家庭提供合理的治疗以及生育指导。

利益冲突 本文所有作者声明不存在利益冲突

参考文献:

[1] DE SILVA S R, ARNO G, ROBSON A G, *et al*. The X-linked retinopathies: Physiological insights, pathogenic mechanisms, phenotypic features and novel therapies[J]. Prog Retin Eye Res, 2021, 82: 100898. DOI: 10.1016/j.preteyeres.2020.100898.

[2] 睢瑞芳, 赵家良. 先天性静止性夜盲[J]. 中华眼科杂志, 2006(5): 472-475.
SUI R F, ZHAO J L. Congenital stationary night blindness[J]. Chin J Ophthalmol, 2006(5): 472-475.

[3] TSANG S H, SHARMA T. Congenital stationary night blindness[M]//Atlas of Inherited Retinal Diseases. Cham: Springer International Publishing, 2018: 61-64. DOI: 10.1007/978-3-319-95046-4_13.

[4] MIYAKE Y. Establishment of the concept of new clinical entities: complete and incomplete form of congenital stationary night blindness[J]. Nippon Ganka Gakkai Zasshi, 2002, 106(12): 737-755;discussion756.

[5] 李蕙, 刘丽英, 徐海燕, 等. 先天性静止性夜盲患者的临床特征[J]. 中华医学杂志, 2012, 92(39): 2756-2759.
LI H, LIU L Y, XU H Y, *et al*. Clinical features of congenital stationary night blindness[J]. Natl Med J China, 2012, 92(39): 2756-2759.

[6] ZEITZ C, ROBSON A G, AUDO I. Congenital stationary night blindness: an analysis and update of genotype-phenotype correlations and pathogenic mechanisms[J]. Prog Retin Eye Res, 2015, 45: 58-110. DOI: 10.1016/j.preteyeres.2014.09.001.

[7] MIYAKE Y. Congenital stationary night blindness with negative electroretinogram: a new classification[J]. Arch Ophthalmol, 1986, 104(7): 1013. DOI: 10.1001/archopht.1986.01050190071042.

[8] BIJVELD M M C, FLORIJN R J, BERGEN A A B, *et al*. Genotype and phenotype of 101 Dutch patients with congenital stationary night blindness[J]. Ophthalmology, 2013, 120(10): 2072-2081. DOI: 10.1016/j.ophtha.2013.03.002.

[9] POELS M M F, DE WIT G C, BIJVELD M M C, *et al*. Natural course of refractive error in congenital stationary night blindness: implications for myopia treatment[J]. Invest Ophthalmol Vis Sci, 2024, 65(14): 9. DOI: 10.1167/iovs.65.14.9.

[10] HUANG L J, BAI X Q, XIE Y, *et al*. Clinical and genetic studies for a cohort of patients with congenital stationary night blindness[J]. Orphanet J Rare Dis, 2024, 19(1): 101. DOI: 10.1186/s13023-024-03091-3.

[11] DURAJCZYK M, LUBIŃSKI W. Congenital stationary night blindness (CSNB): case reports and review of current knowledge[J]. J Clin Med, 2025, 14(4): 1238. DOI: 10.3390/jcm14041238.

[12] 申眉, 李世迎. 先天性静止性夜盲疾病谱及其基因型-表型研究现状与进展[J]. 眼科学报, 2025, 40(2): 144-154. DOI: 10.12419/24111001
SHEN M, LI S Y. Congenital stationary night blindness: an update and review of the disease spectrum and genotype-phenotype correlations[J]. Eye Sci, 2025, 40(2): 144-154. DOI: 10.12419/24111001.

[13] MAHMOOD U, MÉJÉCASE C, ALI S M A, *et al*. A novel splice-site variant in CACNA1F causes a phenotype synonymous with åland island eye disease and incomplete congenital stationary night blindness[J]. Genes, 2021, 12(2): 171. DOI: 10.3390/genes12020171.

[14] 王平, 谢黎, 郗晓芸, 等. CACNA1F基因变异相关眼病临床表型特征[J]. 中华眼底病杂志, 2024(6): 443-448.
WANG P, XIE L, XI X Y, *et al*. Clinical characteristics in ocular diseases with CACNA1F genetic variants[J]. Chin J Ocul Fundus Dis, 2024(6): 443-448.

[15] MIHALICH A, CAMMARATA G, TREMOLADA G, *et al*. Two novel CACNA1F gene mutations cause two different phenotypes: Aland Eye Disease and incomplete Congenital Stationary Night Blindness[J]. Exp Eye Res, 2022, 221: 109143. DOI: 10.1016/j.exer.2022.109143.

[16] PEACHEY N S, RAY T A, FLORIJN R, *et al*. GPR179 is required for depolarizing bipolar cell function and is mutated in autosomal-recessive complete congenital stationary night

blindness[J]. Am J Hum Genet, 2012, 90(2): 331-339. DOI:10.1016/j.ajhg.2011.12.006.

[17] 但汉东, 宋秀胜, 李家璋, 等. Schubert-Bornschein型先天性静止性夜盲致病的分子机制[J]. 中国实用眼科杂志, 2016(5): 407-410.
DAN H D, SONG X S, LI J Z, *et al*. Molecular mechanism of Schubert-Bornschein congenital static night blindness[J]. Chin J Pract Ophthalmol, 2016(5): 407-410.

[18] 徐敏, 杜伟. NYX基因新致病突变引起X连锁隐性遗传先天性静止性夜盲1例[J]. 中华实验眼科杂志, 2022, 40(12): 1206-1207.
XU M, DU W. A novel NYX mutation causes X-linked recessive congenital stationary night blindness: a case report[J]. Chin J Exp Ophthalmol, 2022, 40(12): 1206-1207.

[19] ZEITZ C, VAN GENDEREN M, NEIDHARDT J, *et al*. Mutations in GRM6 cause autosomal recessive congenital stationary night blindness with a distinctive scotopic 15-Hz flicker electroretinogram[J]. Invest Ophthalmol Vis Sci, 2005, 46(11): 4328-4335. DOI:10.1167/iovs.05-0526.

[20] 中华医学会眼科学分会斜视与小儿眼科学组,中国医师协会眼科医师分会斜视与小儿眼科学组.中国儿童弱视防治专家共识(2021年)[J].中华眼科杂志,2021,57(5):336-340.
Group of Strabismus and Pediatric Ophthalmology of Chinese Ophthalmology Society. Expert consensus on prevention and treatment of amblyopia in children[J].Chin J Ophthalmol, 2021,57(5):336-340.

(**收稿**:2025-07-29;**修回**:2025-10-17)

(编辑 张 维)