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分担研究報告書

「無虹彩症の診療ガイドラインの普及・啓発活動および改訂に向けた検討」

研究代表者	**西田 幸二**	**大阪大学 脳神経感覚器外科学(眼科学)**	**教授**
研究分担者	大家 義則	大阪大学 脳神経感覚器外科学(眼科学)	助教
研究協力者	川崎 良	大阪大学 視覚情報制御学寄附講座	寄附講座教授
研究協力者	川﨑 諭	大阪大学 眼免疫再生医学共同研究講座	特任准教授
研究協力者	高 静花	大阪大学 視覚先端医学寄附講座	寄附講座准教授
研究協力者	松下 賢治	大阪大学 脳神経感覚器外科学(眼科学)	准教授
研究協力者	相馬 剛至	大阪大学 脳神経感覚器外科学(眼科学)	助教
研究協力者	渡辺 真矢	大阪大学 脳神経感覚器外科学(眼科学)	大学院生
研究協力者	河本 晋平	淀川キリスト教病院 眼科	医師
研究協力者	阿曽沼 早苗	大阪大学 医学部附属病院(眼科)	視能訓練士

【研究要旨】

無虹彩症は、完全または不完全な虹彩の形成異常に加えて角膜症、白内障、緑内障、黄斑低形成、眼球振盪症などを合併する難治性眼疾患である。責任遺伝子は眼の発生におけるマスター遺伝子として知られている *PAX6* 遺伝子であり、この遺伝子の片アリルの機能喪失によって機能遺伝子量が半減（ハプロ不全）することで発症すると考えられている。

今年度は、無虹彩症の診療ガイドラインの普及・啓発活動として日本眼科学会雑誌への掲載および学会 HP での公開に加え、Minds 専門部会による評価および Minds ガイドラインライブラリでの公開を実施した。また診療ガイドラインの普及率および使用実態の調査を目的としてアンケートを作成した。

A．研究目的

無虹彩症は、完全または不完全な虹彩の形成異常に加えて角膜症、白内障、緑内障、黄斑低形成、眼球振盪症などを合併する難治性眼疾患である。責任遺伝子は眼の発生におけるマスター遺伝子として知られている *PAX6* 遺伝子であり、この遺伝子の片アリルの機能喪失によって機能遺伝子量が半減（ハプロ不全）することで発症すると考えられている。

我々は日本眼科学会主導のもと、関連学会と連携して、これまでに無虹彩症の診断基準および重症度分類を策定した。本研究ではこれらをより質の高いものに改定するとともに、Minds に準拠した方法でエビデンスに基づく診療ガイドラインを策定し、医師、患者ならびに広く国民に普及・啓発活動を行うことで、国内における診療の均てん化を図ることを目的とする。

B．研究方法

昨年度は、無虹彩症の診療ガイドライン

を策定し、学会承認を得た。

今年度は診療ガイドラインの普及・啓発活動を実施する。まず初めに日本眼科学会雑誌への掲載および日本眼科学会 HP での公開を行い、次いで Minds に対してガイドラインの評価および Minds ガイドラインライブラリへの掲載依頼を行う。また海外へ向けて発信するため、英語版を作成する。令和 4 年度には普及・啓発活動に加え、眼科医師向けにアンケート調査を実施し、ガイドラインの普及率および使用実態の調査を行うとともに、改定に向けた検討を行う。

（倫理面への配慮）

すべての研究はヘルシンキ宣言の趣旨を尊重し、関連する法令や指針を遵守し、各施設の倫理審査委員会の承認を得たうえで行うこととする。また個人情報の漏洩防止、患者への研究参加への説明と同意の取得を徹底する。

C．研究結果

無虹彩症の診療ガイドラインについて、日本眼科学会雑誌へ掲載され、学会 HP にて公開を行った。また Minds 専門部会による評価および審議の結果選定となり、Minds ガイドラインライブラリへ掲載された。英語版（資料 1）については作成が終わり、海外雑誌へ投稿する予定である。アンケートについては調査票（資料 2）を作成し、令和 4 年度には日本眼科学会専門医制度認定研修施設 965 施設に向けてアンケートを実施する予定である。

D．考察

日本眼科学会雑誌および日本眼科学会 HP での診療ガイドラインの公開に加え、Minds HP へ掲載され公開されたことで、眼科医だけではなく一般の人もアクセスが容易になったと考える。また眼科医師に向けてアンケートを実施することにより、本診療ガイドラインの周知につながる事が期待される。

今後の課題として、ガイドラインの活用を促進する要因や阻害する要因等についての検討、患者・家族の価値観や希望の反映等が挙げられる。このうちガイドラインの活用を促進する要因や阻害する要因等については、アンケート調査結果をもとに検討したいと考える。本疾患は希少疾患であることから、診療ガイドライン作成過程への患者あるいは支援者の参加は困難であるといえる。そのため出来うる限り患者の視点に立ち、推奨や解説文の作成を行った。今後の改定に向けて、患者やその家族の価値観や意見を取り入れるためにどのような取り組みが可能であるかを検討するとともに、ガイドラインの内容を一般の人向けに分かりやすく解説した患者さん用小冊子の作成等についても検討したいと考えている。

E．結論

今年度は、無虹彩症の診療ガイドラインについて、日本眼科学会雑誌への掲載および学会 HP での公開に加え、Minds 専門部会による評価を受け Minds ガイドラインライブラリへ掲載された。また診療ガイドラインの普及率および使用実態の調査を目的としてアンケートを作成した。来年度には日本眼科学会専門医制度認定研修施設 965 施設に向けてアンケートを実施する予定である。

F．研究発表

1. 論文発表

1. Koh S, Soma T, Oie Y, Nishida K., First Diquafosol Treatment for Dry Eye: 10-Year Follow-Up. Asia Pac

J Ophthalmol (Phila). 2022 Mar 25. doi: 10.1097/APO.0000000000000493. Online ahead of print. PMID: 35342178 No abstract available.

2. Koh S, Inoue R, Maeno S, Mihashi T, Maeda N, Jhanji V, **Nishida K.**, Characteristics of Higher-Order Aberrations in Different Stages of Keratoconus. Eye Contact Lens. 2022 Mar 14. doi: 10.1097/ICL.0000000000000897. Online ahead of print.PMID: 35333804
3. Sakaue TA, Fujishima Y, Fukushima Y, Tsugawa-Shimizu Y, Fukuda S, Kita S, Nishizawa H, Ranscht B, **Nishida K**, Maeda N, Shimomura I., Adiponectin accumulation in the retinal vascular endothelium and its possible role in preventing early diabetic microvascular damage. Sci Rep. 2022 Mar 9;12(1):4159. doi: 10.1038/s41598-022-08041-2.PMID: 35264685
4. Fujimoto S, Wakabayashi T, Maruyama K, Hara C, Oguro-Igashira E, Nishide M, Sakaguchi H, Kumanogoh A, **Nishida K.**, Anterior chamber flare and ciliochoroidal detachment using flare photometry and anterior segment optical coherence tomography in acute lupus choroidopathy: A case report. Am J Ophthalmol Case Rep. 2022 Jan 31;25:101314. doi: 10.1016/j.ajoc.2022.101314. eCollection 2022 Mar.PMID: 35146195
5. Maruyama K, Iwahashi C, Hashida N, Ohguro N, **Nishida K.**, Relationship between vitreoretinal lymphoma and the site of lymphoma development in the central nervous system. Jpn J Ophthalmol. 2022 Mar;66(2):142-150. doi: 10.1007/s10384-021-00891-z. Epub 2021 Dec 3.PMID: 34860298
6. Shiraki N, Maruyama K, Hayashi R, Oguchi A, Murakawa Y, Katayama T, Takigawa T, Sakimoto S, Quantock AJ, Tsujikawa M, **Nishida K.**, PAX6-positive microglia evolve locally in hiPSC-derived ocular organoids. Stem Cell Reports. 2022 Feb 8;17(2):221-230. doi: 10.1016/j.stemcr.2021.12.009. Epub 2022 Jan 13.PMID: 35030319
7. Sugimoto I, Usui S, Okazaki T, Kawashima R, Miki A, Kawasaki R, Matsushita K, **Nishida K.**, Early Three-Dimensional Intrableb Structural Changes in Primary-Open Angle Glaucoma and Exfoliation Glaucoma After Ex-PRESS Surgery. Transl Vis Sci Technol. 2022 Feb 1;11(2):32. doi: 10.1167/tvst.11.2.32.PMID: 35191962
8. Shigeyasu C, Yamada M, Fukuda M, Koh S, Suzuki T, Toshida H, Oie Y, Nejima R, Eguchi H, Kawasaki R, **Nishida K**; Research Group of Severe Contact Lens-associated

Ocular Complications. Severe Ocular Complications Associated With Wearing of Contact Lens in Japan. Eye Contact Lens. 2022 Feb 1;48(2):63-68. doi: 10.1097/ICL.0000000000000870.PMID: 34860721

9. Shiraki A, Sakimoto S, Eguchi M, Kanai M, Hara C, Fukushima Y, Nishida K, Kawasaki R, Sakaguchi H, Nishida K., Analysis of Progressive Neovascularization in Diabetic Retinopathy Using Widefield OCT Angiography. Ophthalmol Retina. 2022 Feb;6(2):153-160. doi: 10.1016/j.oret.2021.05.011. Epub 2021 May 26.PMID: 34051418
10. Koh S, Inoue R, Maeda N, Oie Y, Jhanji V, Miki A, Nishida K., Corneal tomographic changes during corneal rigid gas-permeable contact lens wear in keratoconic eyes. Br J Ophthalmol. 2022 Feb;106(2):197-202. doi: 10.1136/bjophthalmol-2020-317057. Epub 2020 Nov 10.PMID: 33172864
11. Miki A, Okazaki T, Weinreb RN, Morota M, Tanimura A, Kawashima R, Usui S, Matsushita K, Nishida K., Evaluating Visual Field Progression in Advanced Glaucoma Using Trend Analysis of Targeted Mean Total Deviation. J Glaucoma. 2022 Jan 12. doi: 10.1097/IJG.0000000000001985. Online ahead of print.PMID: 35019876
12. Maruyama K, Mei S, Sakaguchi H, Hara C, Miki A, Mao Z, Kawasaki R, Wang Z, Sakimoto S, Hashida N, Quantock AJ, Chan K, Nishida K., Diagnosis of Choroidal Disease With Deep Learning-Based Image Enhancement and Volumetric Quantification of Optical Coherence Tomography. Transl Vis Sci Technol. 2022 Jan 3;11(1):22. doi: 10.1167/tvst.11.1.22. PMID: 35029631
13. Matsushita K, Kawashima R, Hashida N, Hamano Y, Harada K, Higashisaka K, Baba K, Sato S, Huang W, Matsumoto H, Hamanaka T, Quantock AJ, Nishida K., Barium-induced toxic anterior segment syndrome. Eur J Ophthalmol. 2021 Dec 30:11206721211069223. doi: 10.1177/11206721211069223. Online ahead of print.PMID: 34967228
14. Kawashima R, Matsushita K, Hashida N, Kuniyoshi K, Fujikado T, Nishida K., Complete Visual Recovery From Severe Outer Retinitis After Tonsillitis. J Neuroophthalmol. 2021 Dec 1;41(4):e578-e583. doi: 10.1097/WNO.0000000000001073.PMID: 32956228
15. Uematsu S, Sakaguchi H, Sayanagi K, Ikuno Y, Yokoyama A, Asai T, Fukushima Y, Hara C, Sakimoto S, Nishida K., Association between choriocapillaris flow deficit and choroidal neovascularization activity in

eyes with myopic choroidal neovascularization. Sci Rep. 2021 Nov 9;11(1):21947. doi: 10.1038/s41598-021-01557-z.PMID: 34754047

16. Takahashi S, Usui S, Hashida N, Kubota H, Nishida K, Sakaguchi H, **Nishida K.,** Multimodal imaging of indapamide-induced bilateral choroidal effusion: a case report. BMC Ophthalmol. 2021 Nov 4;21(1):386. doi: 10.1186/s12886-021-02147-3.PMID: 34736431
17. Matsushita K, Kawashima R, Kawasaki R, **Nishida K.,** Prognostic factors for successful Baerveldt glaucoma implant surgery for refractory glaucoma after multiple surgeries. Jpn J Ophthalmol. 2021 Nov;65(6):820-826. doi: 10.1007/s10384-021-00864-2. Epub 2021 Aug 10.PMID: 34374907
18. Yasukura Y, Miki A, Maeda N, Koh S, **Nishida K.,** Effect of Prostaglandin Analogues on Corneal Biomechanical Parameters Measured With a Dynamic Scheimpflug Analyzer. J Glaucoma. 2021 Nov 1;30(11):996-1000. doi: 10.1097/IJG.0000000000001895.PMID: 34086611
19. Hara C, Sawa M, Gomi F, **Nishida K.,** Questionnaire Survey on Driving among Patients with Age-Related Macular Degeneration in Japan. J Clin Med. 2021 Oct 21;10(21):4845. doi: 10.3390/jcm10214845.PMID: 34768365
20. Koh S, Chalmers R, Yamasaki K, Kawasaki R, **Nishida K.,** Factors influencing the 8-item contact lens dry eye questionnaire score and comparison of translations in Japanese soft contact lens wearers. Cont Lens Anterior Eye. 2021 Oct 4:101519. doi: 10.1016/j.clae.2021.101519. Online ahead of print.PMID: 34620551
21. Hara C, Kamei M, Sakaguchi H, Matsumura N, Sakimoto S, Suzuki M, Nishida K, Fukushima Y, **Nishida K.,** Long-term outcomes of intravitreal activated protein C injection for ischemic central retinal vein occlusion: an extension trial. Graefes Arch Clin Exp Ophthalmol. 2021 Oct;259(10):2919-2927. doi: 10.1007/s00417-021-05072-9. Epub 2021 Apr 24.PMID: 33893866
22. Kanai M, Shiozaki D, Sakimoto S, Shiraki A, Hara C, Kawasaki R, Sakaguchi H, **Nishida K.,** Association of disorganization of retinal inner layers with optical coherence tomography angiography features in branch retinal vein occlusion. Graefes Arch Clin Exp Ophthalmol. 2021 Oct;259(10):2897-2903. doi: 10.1007/s00417-021-05168-2. Epub 2021 Apr 16.PMID: 33861366

23. Shugyo A, Koh S, Inoue R, Ambrósio R Jr, Miki A, Maeda N, **Nishida K.**, Optical Quality in Keratoconus Is Associated With Corneal Biomechanics. Cornea. 2021 Oct 1;40(10):1276-1281. doi: 10.1097/ICO.0000000000002631.PMID: 33332893
24. Shiraki A, Sakimoto S, **Nishida K.**, Reconnection of the Severed Vein in Proliferative Diabetic Retinopathy. Ophthalmol Retina. 2021 Sep;5(9):887. doi: 10.1016/j.oret.2021.06.004.PMID: 34503759
25. Wakabayashi T, Kamuro R, Hashida N, Shiraki N, Sakaguchi H, Ohguro N, **Nishida K.**, Acute endophthalmitis and hyphema mimicking pink hypopyon associated with ocular toxocariasis: A case report. Am J Ophthalmol Case Rep. 2021 Aug 10;23:101188. doi: 10.1016/j.ajoc.2021.101188. eCollection 2021 Sep.PMID: 34430757
26. Nakao G, Koh S, Inoue R, Maeno S, Maeda N, **Nishida K.**, The Characteristics and Risk Factors of Very Asymmetric Keratoconus. Eye Contact Lens. 2021 Sep 1;47(9):511-514. doi: 10.1097/ICL.0000000000000830.PMID: 34424224
27. Yasukura Y, Oie Y, Kawasaki R, Maeda N, Jhanji V, **Nishida K.**, New severity grading system for Fuchs endothelial corneal dystrophy using anterior segment optical coherence tomography. Acta Ophthalmol. 2021 Sep;99(6):e914-e921. doi: 10.1111/aos.14690. Epub 2020 Nov 30.PMID: 33258212
28. Kawashima R, Baba K, Matsushita K, Soma T, Kurashige M, Umeda D, Nakamura M, Morii E, **Nishida K.**, Intraocular Endoscopy Resolved Tube Occlusion of an Ahmed Glaucoma Valve. Case Rep Ophthalmol. 2021 Aug 24;12(2):706-711. doi: 10.1159/000518286. eCollection 2021 May-Aug.PMID: 34594208
29. Suzaki A, Koh S, Maeda N, Asonuma S, Santodomingo-Rubido J, Oie Y, Soma T, Fujikado T, **Nishida K.**, Optimizing correction of coma aberration in keratoconus with a novel soft contact lens. Cont Lens Anterior Eye. 2021 Aug;44(4):101405. doi: 10.1016/j.clae.2020.12.071. Epub 2021 Jan 10.PMID: 33436159
30. Takahashi S, Nishida K, Sakaguchi H, **Nishida K.**, A Case of Idiopathic Dense Vitreous Hemorrhage: Suspected Rupture of a Large Retinal Arterial Macroaneurysm on the Optic Disc. Case Rep Ophthalmol. 2021 Jul 16;12(2):634-639. doi: 10.1159/000511110. eCollection 2021 May-Aug.PMID: 34413754
31. Eguchi M, Sakaguchi H, **Nishida K.**, Giant Proliferative Membrane

Triggering Re-retinal Detachment after Vitrectomy. Ophthalmol Retina. 2021 Jul;5(7):655. doi: 10.1016/j.oret.2021.01.010.PMID: 34243969

32. Chen ZJ, Ma L, Brelen ME, Chen H, Tsujikawa M, Lai TY, Ho M, Sayanagi K, Hara C, Hashida N, Tam PO, Young AL, **Nishida K**, Tham CC, Pang CP, Chen LJ., Identification of *TIE2* as a susceptibility gene for neovascular age-related macular degeneration and polypoidal choroidal vasculopathy. Br J Ophthalmol. 2021 Jul;105(7):1035-1040. doi: 10.1136/bjophthalmol-2019-315746. Epub 2020 Mar 9.PMID: 32152144
33. Watanabe S, Hayashi R, Sasamoto Y, Tsujikawa M, Ksander BR, Frank MH, Quantock AJ, Frank NY, **Nishida K.**, Human iPS cells engender corneal epithelial stem cells with holoclone-forming capabilities. iScience. 2021 Jun 5;24(6):102688. doi: 10.1016/j.isci.2021.102688. eCollection 2021 Jun 25.PMID: 34195566
34. Koh S, Maeda N, **Nishida K.**, Trick of the Light Through the Cornea. Ophthalmology. 2021 Jun;128(6):836. doi: 10.1016/j.ophtha.2020.12.026.PMID: 34023051
35. Yasukura Y, Wakabayashi T, Sakaguchi H, **Nishida K.**, Simultaneous bilateral choroidal neovascularization associated with *Staphylococcus aureus* infective endocarditis: A case report. Am J Ophthalmol Case Rep. 2021 Feb 18;22:101037. doi: 10.1016/j.ajoc.2021.101037. eCollection 2021 Jun.PMID: 33681530
36. Mitsuoka Y, Soma T, Maruyama K, **Nishida K.**, Fusarium infection complicating rheumatic keratitis that acutely progressed to endophthalmitis during regular infusion of tocilizumab: a case report. BMC Ophthalmol. 2021 May 19;21(1):224. doi: 10.1186/s12886-021-01981-9.PMID: 34011297
37. Nishida K, Takahashi S, Sakaguchi H, Sato S, Kanai M, Shiraki A, Wakabayashi T, Hara C, Fukushima Y, Sakimoto S, Sayanagi K, Kawasaki R, **Nishida K.**, Impact of photoreceptor density in a 3D simulation of panretinal laser photocoagulation. BMC Ophthalmol. 2021 May 7;21(1):200. doi: 10.1186/s12886-021-01945-z.PMID: 33962561
38. Hieda O, Hiraoka T, Fujikado T, Ishiko S, Hasebe S, Torii H, Takahashi H, Nakamura Y, Sotozono C, Oshika T, Morimoto T, **Nishida K**, Nishikawa N, Song YS, Tokutake T, Nishi Y, Shigeno Y, Kurihara T, Negishi K, Tsubota K, Ono M, Nakai T, Tan D, Tanaka S, Kinoshita S; ATOM-J. Study Group., Efficacy and safety of 0.01% atropine for

prevention of childhood myopia in a 2-year randomized placebo-controlled study. Jpn J Ophthalmol. 2021 May;65(3):315-325. doi: 10.1007/s10384-021-00822-y. Epub 2021 Feb 14.PMID: 33586090

39. Ando S, Hashida N, Yamashita D, Kawabata T, Asao K, Kawasaki S, Sakurai K, Yoshimori T, **Nishida K.**, Rubicon regulates A2E-induced autophagy impairment in the retinal pigment epithelium implicated in the pathology of age-related macular degeneration. Biochem Biophys Res Commun. 2021 Apr 30;551:148-154. doi: 10.1016/j.bbrc.2021.02.148. Epub 2021 Mar 16.
40. **西田 幸二**, 村上 晶, 東 範行, 島崎 潤, 宮田 和典, 山田 昌和, 外園 千恵, 白石 敦, 榛村 重人ら. 前眼部形成異常の診療ガイドライン. 日本眼科学会雑誌 125 巻 6 号 Page605-629, 2021.
41. **西田 幸二**, 東 範行, 阿曽沼 早苗, 石井 一葉, 臼井 智彦, 大家 義則, 春日 俊光, 川崎 良, 倉上 弘幸, 河本 晋平ら. 無虹彩症の診療ガイドライン. 日本眼科学会雑誌 125 巻 1 号 Page38-76, 2021.
42. 岡崎 智之, 河嶋 瑠美, 松下 賢治, 相馬 剛至, 臼井 審一, 三木 篤也, **西田 幸二**. 人工角膜移植後続発緑内障に対して二期的にバルベルトインプラント挿入術を施行した一例. 眼科臨床紀要 14 巻 9 号 Page562-567, 2021
43. 伊藤 潤, 原 千佳子, 若林 卓, 白木 彰彦, 坂口 裕和, **西田 幸二**. 光干渉断層血管撮影にて血流改善が観察できた網膜中心動脈閉塞症による一過性網膜虚血に伴う paracentral acute middle maculopathy の 1 例. 日本眼科学会雑誌 125 巻 8 号 Page732-737, 2021
44. 満岡 友祐, 森本 壮, **西田 幸二**. 多局所 ERG で中心部網膜の反応が Supernormal を示したオカルト黄斑ジストロフィ(OMD)の長期経過. 眼科臨床紀要 14 巻 8 号 Page501-505, 2021
45. 渡邊 怜美, 相馬 剛至, 大家 義則, 高 静花, 川崎 諭, 辻川 元一, 前田 直之, **西田 幸二**. 視力予後不良の水疱性角膜症に対する QOL 改善を目的とした角膜内皮移植の 4 例. 眼科臨床紀要 14 巻 2 号 Page73-76, 2021.
46. 雲井 美帆, 三木 篤也, 松下 賢治, 前田 直之, **西田 幸二**. コンタクトレンズセンサーによる緑内障患者の 24 時間眼圧測定. 眼科臨床紀要 14 巻 1 号 Page6-10, 2021

2. 学会発表

1. Development of stem cell-based therapy for limbal stem cell deficiency
   -from tissue stem cell to iPS cell, **Kohji Nishida**, The 7th Asia Cornea Society Biennial Scientific Meeting, 2021/4/28, International (Web), Oral
2. Development of iPS cell-based therapy for limbal stem cell deficiency, **Kohji Nishida**, The 7th Asia Cornea Society Biennial

Scientific Meeting, 2021/4/29, International (Web), Oral

3. iPS 細胞を用いた角膜再生医療, 西田幸二, 第 122 回日本耳鼻咽喉科学会総会・学術講演会, 2021/5/14, 国内 (Web), 口頭
4. iPS を用いた角膜移植, 西田幸二, 第 48 回日本毒性学会, 2021/7/8, 国内 (神戸国際会議場), 口頭
5. 眼の再生医療, 西田幸二, 第 93 回筑後眼科研究会, 2021/7/17, 国内 (Web), 口頭
6. Ocular Surface Reconstruction Using iPSC-derived Corneal Epithelial Cell, Kohji Nishida, The 36th Asia-Pacific Academy of Ophthalmology Congress, 2021/9/10, International (Web), Oral
7. 角膜の再生医療, 西田幸二, 第 52 回大阪医科薬科大学眼科セミナー, 2021/9/18, 国内 (ホテルグランヴィア大阪), 口頭
8. 再生医療で視力を回復させる, 西田幸二, 第 152 回広島県眼科医会講習会, 2021/10/3, 国内 (Web), 口頭
9. iPS 細胞を用いた角膜再生医療, 西田幸二, 第 35 回千葉県眼科手術懇話会, 2021/10/23, 国内 (Web), 口頭
10. 細胞シートを用いた角膜再生医療, 西田幸二, 第 2 回細胞シート工学イノベーションフォーラム, 2021/11/1, 国内 (Web), 口頭
11. Induced Pluripotent Stem Cell-Derived Limbal Stem Cells, Kohji Nishida, AAO 2021 - American Academy of Ophthalmology Annual Meeting -, 2021/11/13, International (Web), Oral
12. 保健承認された角膜再生医療, 西田幸二, 第 20 回眼科診療アップデートセミナー2022 in Kyoto, 2022/3/13, 国内 (Web), 口頭
13. アカデミアから見た再生医療産業化の現状と課題, 西田幸二, 第 21 回日本再生医療学会, 2021/3/18, 国内 (Web), 口頭

G. 知的財産権の出願・登録状況

1. 特許取得
   該当なし
2. 実用新案登録
   該当なし
3. その他
   該当なし

Clinical Practice Guideline for Aniridia

Research on rare and intractable diseases, Health, Labour and Welfare Sciences Research Grants
Clinical Practice Guideline Development Committee† of the research group of "Intractable corneal diseases on establishing standardized diagnosis and treatment"

Introduction

Aniridia is an intractable eye disease with varying degrees of iris hypoplasia and is complicated by keratopathy, cataract, glaucoma, macular hypoplasia, and nystagmus. The responsible gene is *PAX6*, which is known as the master gene in ocular development, and aniridia develops when the amount of functional gene is halved (haploinsufficiency) because of a loss of function of one allele.

The Act on Medical Care for Patients with Intractable Diseases (Intractable/Rare Disease Act) designates aniridia as an intractable disease, and the diagnostic criteria and severity classification of aniridia were defined by the research group of "Intractable corneal diseases on establishing standardized diagnosis and treatment." We have now created this clinical practice guideline in accordance with the Medical Information Network Distribution Service (Minds) method to ensure that high-quality medical care is provided to patients with aniridia. Minds is part of the Promotion Project for Evidence-Based Medicine, which is operated by the Japan Council for Quality Health Care and is commissioned by the Ministry of Health, Labour and Welfare.

Minds defines clinical practice guidelines as "Documents presenting the optimum recommendation to support the decision-making of patients and healthcare professionals by taking into consideration systematic reviews of the evidence, their overall assessment, and the balance between benefits and harms, etc., for medical practices of high clinical importance."

Therefore, rather than creating guidelines for important clinical issues in the clinical management of aniridia by aggregating opinions from experts, we systematically collected evidence through systematic reviews, evaluated and summarized all the evidence and then, on the basis of this evaluation, summarized the recommendations for clinical questions on important clinical issues.

In this clinical practice guideline, we summarize the evidence for 6 clinical questions and 3 background questions that we consider to be important for clinical practice and make recommendations for the clinical questions. Randomized controlled trials and other high-evidence studies have not been conducted on aniridia because it is a rare disease, so strong recommendations could not be made for any of the clinical questions. However, as specified as a goal of Minds, we hope that this clinical practice guideline will help patients and healthcare professionals to discuss information on treatment options that are considered scientifically appropriate and help them to agree on and select the best approach under consideration of the patients' wishes and beliefs, the healthcare professionals' ethics, and social constraints.

†: Research on rare and intractable diseases, Health, Labour and Welfare Sciences Research Grants; the research group of "Intractable corneal diseases on establishing standardized diagnosis and treatment"
Chairperson: Kohji Nishida (Department of Ophthalmology, Osaka University Graduate School of Medicine)
Supervisory committee: Akira Murakami (Department of Ophthalmology, Juntendo University Graduate School of Medicine)
Noriyuki Azuma (Department of Ophthalmology and Laboratory for Visual Science, National Center for Child Health and Development)
Jun Shimazaki (Department of Ophthalmology, Tokyo Dental College, Ichikawa General Hospital)
Kazunori Miyata (Miyata Eye Hospital, Medical Corporation Meiwakai)
Masakazu Yamada (Department of Ophthalmology, Kyorin University School of Medicine)
Chie Sotozono (Department of Ophthalmology, Kyoto Prefectural University of Medicine)
Atsushi Shiraishi (Department of Ophthalmology, Ehime University School of Medicine)
Shigeto Shimmura (Department of Ophthalmology, Keio University School of Medicine)
Tomohiko Usui (Department of Ophthalmology, University of Tokyo Hospital)
Yoshinori Oie (Department of Ophthalmology, Osaka University Graduate School of Medicine)
Committee members: Please refer to the main text
Contact: Kohji Nishida, Department of Ophthalmology, Osaka University Graduate School of Medicine
2-2 Yamadagaoka, Suita, Osaka, 565-0871, Japan
Conflicts of interest: Please refer to the main text

Research on rare and intractable diseases, Health, Labour and Welfare Sciences Research Grants
The research group of “Intractable corneal diseases on establishing standardized diagnosis and treatment”
Kohji Nishida, Principal Investigator

List of Authors

● Chairperson

Kohji Nishida	Department of Ophthalmology, Osaka University Graduate School of Medicine

● Committee members (In alphabetical order)

Akira Matsuda	Department of Ophthalmology, Juntendo University Graduate School of Medicine
Akira Murakami	Department of Ophthalmology, Juntendo University Graduate School of Medicine
Atsushi Shiraishi	Department of Ophthalmology, Ehime University School of Medicine
Daisuke Tomida	Department of Ophthalmology, Tokyo Dental College, Ichikawa General Hospital
Hiroyuki Kurakami	Department of Medical Innovation, Osaka University Hospital
Hitoha Ishii	Department of Ophthalmology, University of Tokyo Hospital
Jun Shimazaki	Department of Ophthalmology, Tokyo Dental College, Ichikawa General Hospital
Junko Yoshida	Department of Ophthalmology, University of Tokyo Hospital
Kanji Hori	Department of Ophthalmology, Juntendo University Graduate School of Medicine
Kenji Matsushita	Department of Ophthalmology, Osaka University Graduate School of Medicine
Mayumi Sainohira	Department of Ophthalmology, Kagoshima University Faculty of Medicine
Motokazu Tsujikawa	Department of Biomedical Informatics, Division of Health Sciences, Osaka University Graduate School of Medicine
Noriyuki Azuma	Department of Ophthalmology and Laboratory for Visual Science, National Center for Child Health and Development
Ryo Kawasaki	Department of Vision Informatics, Osaka University Graduate School of Medicine
Sanae Asonuma	Department of Ophthalmology, Osaka University Graduate School of Medicine
Shimpei Komoto	Department of Ophthalmology, Yodogawa Christian Hospital
Takahiro Minami	Department of Ophthalmology, University of Tokyo Hospital
Takashi Miyai	Department of Ophthalmology, University of Tokyo Hospital
Takefumi Yamaguchi	Department of Ophthalmology, Tokyo Dental College, Ichikawa General Hospital
Tomohiko Usui	Department of Ophthalmology, University of Tokyo Hospital
Tomomi Yamada	Department of Medical Innovation, Osaka University Hospital
Toshimitsu Kasuga	Department of Ophthalmology, Juntendo University Graduate School of Medicine
Yasuhito Hayashi	Department of Ophthalmology, Ehime University School of Medicine
Yoshinori Oie	Department of Ophthalmology, Osaka University Graduate School of Medicine
Yuko Hara	Department of Ophthalmology, Ehime University School of Medicine
Yumi Hashimoto	Department of Ophthalmology, University of Tokyo Hospital

● External review committee members

Yuichi Hori	Department of Ophthalmology, Toho University Omori Medical Center
Toshiyuki Ojima	Department of Community Health and Preventive Medicine, Hamamatsu University School of Medicine

● Approved by

Japanese Ophthalmological Society
Japan Cornea Society
Japanese Association of Pediatric Ophthalmology

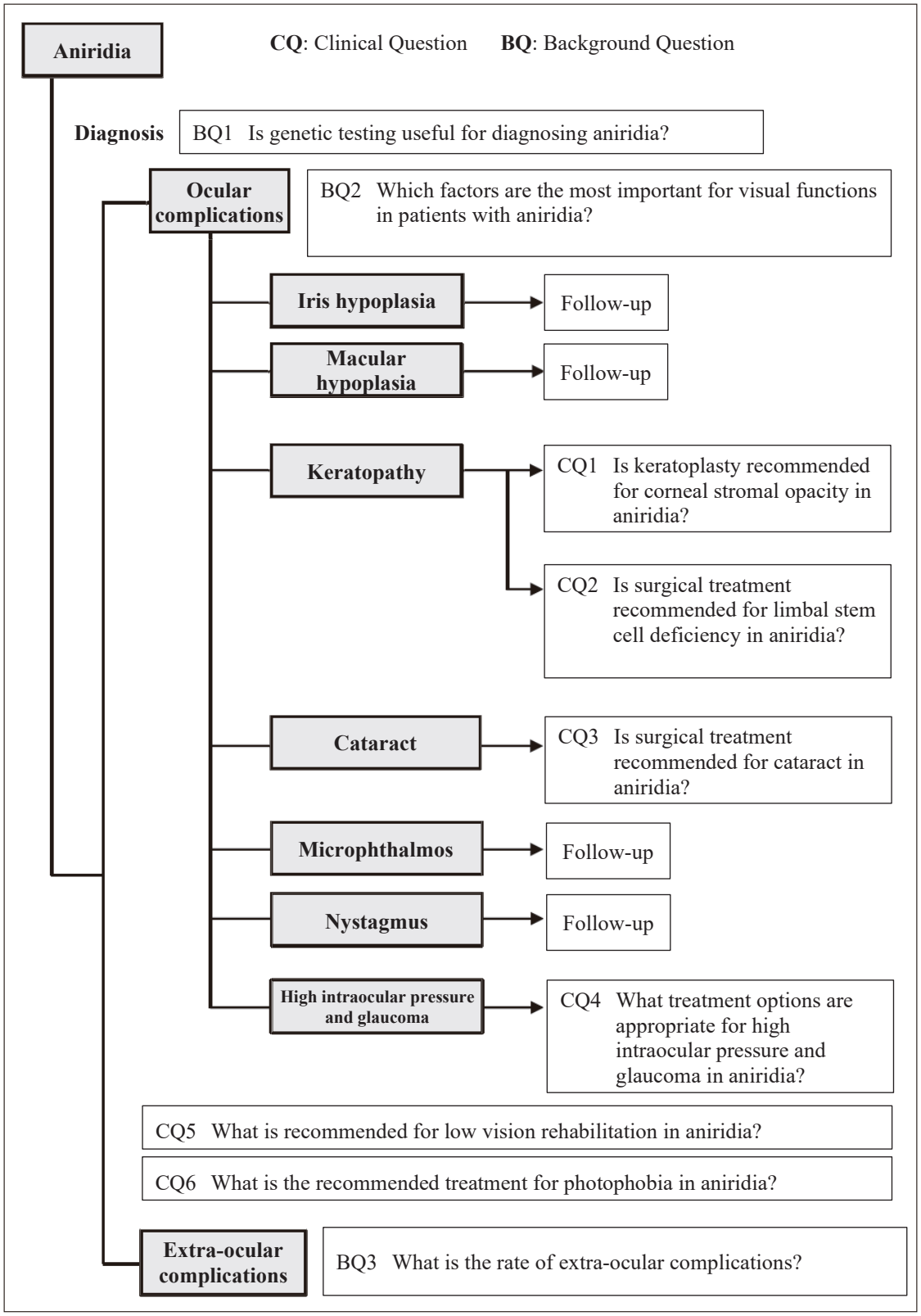
● Collaborators

Noriaki Akai	Life Sciences Library, Osaka University Library
Nozomi Nishida	Department of Ophthalmology, Osaka University Graduate School of Medicine

Guideline Summary

CQ No.	CQ	Summary and recommendation	Level of recommendation
1	Is keratoplasty recommended for corneal stromal opacity in aniridia?	Weak recommendation not to perform keratoplasty for corneal stromal opacity in aniridia. The improvement in visual function obtained by a corneal transplant is limited because of the comorbidities of aniridia. In addition, the long-term prognosis for visual acuity is often poor because of glaucoma and graft dysfunction over time.	Weakly recommend “not to implement”
2	Is surgical treatment recommended for limbal stem cell deficiency in aniridia?	Weak recommendation to perform surgical treatment for limbal stem cell deficiency in aniridia. Specifically, a certain rate of successful ocular surface reconstruction can be expected by performing allogeneic limbal transplantation or cultivated oral mucosal epithelial transplantation. In addition, when corneal stromal opacity is present, combination with a corneal transplant is often useful for improving visual acuity.	Weakly recommend “to implement”
3	Is surgical treatment recommended for cataract in aniridia?	Although some patients with aniridia may experience an improvement in visual acuity with cataract surgery in aniridia, surgery is difficult because of the fragility associated with the capsule and the Zonule of Zinn; in addition, there is a high risk of possible worsening of postoperative glaucoma, anterior fibrosis syndrome, and bullous keratopathy. Therefore, it is recommended to consider the risks associated with surgery and to provide sufficient information to the patient in advance.	Weakly recommend “to implement”
4	What treatment options are appropriate for high intraocular pressure and glaucoma in aniridia?	To lower intraocular pressure, the following procedures should be performed: (1) intraocular pressure-lowering therapy with drugs such as eye drops and oral drugs, (2) angle surgery (goniotomy, trabeculotomy), (3) filtration surgery (mainly trabeculectomy), (4) glaucoma implant surgery, and (5) ciliary body coagulation. When selecting treatment, first consider drug therapy, such as eye drops and oral drugs, and thereby pay attention to adverse effects; if treatment is ineffective, consider angle surgery. If angle surgery is difficult or unsuccessful, trabeculectomy or glaucoma implant surgery should be considered; when deciding which treatment to perform, it is recommended to consider factors such as the condition of the affected eye, the surgeon’s level of experience, and whether or not the facility has been certified for glaucoma implant surgery. If these treatments are unsuccessful, ciliary body coagulation may be performed, but only if the benefits outweigh the risk of complications with poor visual prognosis, such as phthisis bulbi.	Strongly recommend “to implement”
5	What is recommended for low vision rehabilitation in aniridia?	The basis for low vision rehabilitation is refraction correction for ametropia, which aims to improve visual function in aniridia. It is also recommended to use additional low vision devices, such as magnifiers, tinted lenses, low vision glasses, closed-circuit television (CCTV), and Iris Lens.	Strongly recommend “to implement”
6	What is the recommended treatment for photophobia in aniridia?	Tinted lenses and Iris Lens are recommended as treatments for photophobia in aniridia.	Strongly recommend “to implement”

Medical Diagram



Chapter 1 Scope

I. Clinical characteristics

Aniridia is a disease caused by haploinsufficiency of the *PAX6* gene, the master gene of ocular development, due to a loss-of-function mutation of 1 allele[1]. The *PAX6* gene is expressed in various tissues of the eyeball during development, and therefore a diverse range of ocular complications can occur. In addition to iris hypoplasia with varying degrees of severity, keratopathy, cataract, glaucoma, macular hypoplasia, and nystagmus syndrome can occur[2)-4)]. Background question (BQ) 2 summarizes the ocular complications that are considered to be important factors in determining visual function. In addition, extra-ocular complications such as callosal agenesis, epilepsy, higher brain dysfunction, anosmia, diabetes, and Wilms' tumor are also known to occur, and the incidences of these extra-ocular complications are summarized in BQ3[5)6)].

II. Epidemiological characteristics

Aniridia is a rare disease, and the prevalence is assumed to be 1/64,000 to 1/96,000.[7)8)] There is no difference between men and women. Aniridia is a hereditary disease with an autosomal dominant inheritance pattern. Approximately 2/3 of cases are familial, and the rest (1/3) are sporadic.

III. Flow of medical care and treatment

1. Diagnosis and severity

The research group "Intractable corneal diseases on establishing standardized diagnosis and treatment" has established the following diagnostic criteria and severity grading for aniridia[9)]. The usefulness of genetic testing is summarized in BQ1.

1) Diagnostic criteria

A. Symptoms
   1. Bilateral visual impairment (Note 1)
   2. Photophobia (Note 2)

B. Test findings
   1. Slit-lamp examination shows iris hypoplasia with varying degrees of severity, ranging from partial iris atrophy to complete iris deficiency (Note 3).
   2. Hypoplasia of the macula is observed by fundus examination and optical coherence tomography (OCT) (Note 4).
   3. Slit-lamp examination reveals corneal lesions such as limbal stem cell deficiency and corneal opacity (Note 5).
   4. Slit-lamp examination findings indicate a cataract (Note 6).
   5. Ultrasonography, magnetic resonance imaging (MRI), and computed tomography (CT) show microphthalmia.
   6. Nystagmus is observed.
   7. Glaucoma is shown by intraocular pressure test (Note 7).

C. Differential diagnosis
   1. Iris atrophy due to past infection with the herpesvirus family
   2. Iris defect due to trauma or intraocular surgery
   3. Iris coloboma associated with closure of the optic fissure
   4. Rieger anomaly
   5. Iridocorneal endothelial syndrome

D. Extra-ocular complications
   Abnormalities associated with *PAX6* gene mutation (Note 8)

E. Genetic tests
   A pathogenic gene mutation or deletion of the 11p13 region of the *PAX6* gene is identified.

F. Other findings
   Other family members are affected (Note 9).

<Diagnosis category>

Definite : Any item under A and both B1 and E are observed, and all items under C are excluded.

Probable : (1) Any item under A and both B1 and F are observed, and all items under C are excluded.
(2) Any item under A and both B1 and B2 are observed, and all items under C are excluded.
(3) Any item under A and both B1 and B3 are observed, and all items under C are excluded.

Possible : Any item under A and also B1 are observed, but the items under C cannot be completely excluded.

Note 1. Visual impairment is caused by ocular complications such as macular hypoplasia, cataract, glaucoma, and limbal stem cell deficiency.

Note 2. Photophobia can be an accompanying symptom, depending on the degree of the iris defect.

Note 3. 60%–90% are bilateral.

Note 4. The macula pigment, foveal depression, and foveal avascular area of the macula become obscured.

Note 5. Depending on the disease stage, corneal lesions of varying degrees can occur, ranging from hypoplasia of the palisades of Vogt to invasion of conjunctival tissue with blood vessels and keratinization of the epithelium.

Note 6. This complication occurs in about 80% of cases.

Note 7. This complication occurs in 50% to 75% of cases due to dysplasia of the angle.

Note 8. The *PAX6* gene is expressed not only in ocular tissues but also in the central nervous system, islands of Langerhans (in the pancreas), and olfactory epithelium. Hypoplasia of these tissues causes callosal agenesis, epilepsy, higher brain dysfunction, anosmia, and glucose intolerance. Thus, *PAX6* gene mutation may be accompanied by various extra-ocular complications.

Note 9. Familial (autosomal dominant inheritance) aniridia accounts for 2/3 of cases, and the rest are sporadic.

2) Severity classification

Grade I : One eye is affected, and the fellow eye (the other eye) is healthy.
Grade II : Both eyes are affected, and the corrected visual acuity of the better eye is ≥0.3.
Grade III : Both eyes are affected, and the corrected visual acuity of the better eye is ≥0.1 and <0.3.
Grade IV : Both eyes are affected, and the corrected visual acuity of the better eye is <0.1.

Note 1 : "Healthy" is a condition in which the corrected visual acuity is ≥1.0, no visual field abnormality is observed, and no organic abnormality is observed in the eye.
Note 2 : In grades I to III, if secondary glaucoma is accompanied by a narrowing of the visual field of the better eye, the severity classification is moved up by 1 level.
Note 3 : Narrowed vision indicates that the residual visual field at the center is within 20 degrees with the Goldmann perimeter I/4 optotype.
Note 4 : If visual acuity cannot be measured in patients, e.g., infants, the severity classification should be determined comprehensively from ophthalmological findings, etc.

2. Treatment

Although various ocular complications are observed in aniridia, follow-up is required for iris hypoplasia, macular hypoplasia, microphthalmia, and nystagmus because, in principle, there is no treatment for these conditions (refer to the Medical Diagram). The ocular complications keratopathy (of which there are 2 types: corneal stromal opacity and limbal stem cell deficiency), cataract, and high intraocular pressure and glaucoma may be treated with keratoplasty, cataract surgery, and glaucoma eye drops or surgery, respectively[10)-12)]. Specific details are described under Clinical Question (CQ) 1 to CQ4, respectively. Refer to CQ5 for low vision care and CQ6 for the treatment of photophobia, which is a frequent patient complaint.

IV. Information described in this clinical practice guideline

1. Title

Clinical practice guideline for aniridia

2. Purpose

This clinical practice guideline aims to improve the following outcomes:

- Diagnosis of aniridia
- Visual function in aniridia
- Keratopathy as an ocular complication
- Cataract as an ocular complication
- Glaucoma as an ocular complication
- Diagnosis of extraocular complications

3. Topic

Diagnosis of aniridia and clinical management of ocular complications

4. Expected users and facilities, and medical sites where the guideline may be applicable

Physicians at ophthalmology departments of university hospitals and regional core hospitals, practitioners at eye clinics, and patients

5. Relationship with existing guidelines

There are no existing clinical practice guidelines in Japan.

6. Important clinical issues

1) Genetic tests for aniridia

Genetic testing for pathogenic mutations in the *PAX6* gene or deletion of the 11p13 region has been performed to diagnose aniridia. However, its usefulness is debatable.

2) Visual function in aniridia

Ocular complications of aniridia include iris hypoplasia, macular hypoplasia, keratopathy, cataract, microphthalmia, nystagmus, and glaucoma. However, it is unclear to what extent each ocular complication affects visual function.

3) Treatment options for keratopathy (corneal stromal opacity)

Keratopathy, one of the ocular complications of aniridia, includes corneal stromal opacity and limbal stem cell deficiency. Penetrating keratoplasty may be performed as treatment for corneal stromal opacity. However, it has not been determined which of the various treatment options is optimal.

4) Treatment options for keratopathy (limbal stem cell deficiency)

Keratopathy, one of the ocular complications of aniridia, includes corneal stromal opacity and limbal stem cell deficiency. Limbal transplantation and cultivated epithelial transplantation may be performed as treatments for limbal stem cell deficiency. However, it has not been determined which treatment is optimal.

5) Treatment options for cataract

In aniridia, cataract surgery is often more difficult than usual because of corneal opacity and a shallow anterior chamber. Keratopathy may also progress as a result of surgical invasion. Therefore, it is necessary to clarify whether cataract surgery or follow-up should be selected.

6) Treatment options for glaucoma

Treatment of glaucoma, one of the ocular complications of aniridia, includes eye drops, oral drugs, and surgical treatment. Surgical treatment is performed when there is a lack of response to eye drops and oral drugs. Surgical treatment includes trabeculotomy, trabeculectomy, and implant surgery and is selected according to the patient's age, residual visual field, intraocular pressure, and background factors. Each type of surgery has its own adverse effects and complications, and it is not clear which treatment is most appropriate. Therefore, clarification is required.

7) Treatment options for low vision
Patients with aniridia often complain of low vision. It has not been determined what type of low vision care is appropriate.

8) Treatment options for photophobia
Patients with aniridia often complain of photophobia. It has not been determined what type of photophobic care is appropriate.

9) Extra-ocular complications in aniridia
Aniridia may be associated with extra-ocular complications such as callosal agenesis, epilepsy, higher brain dysfunction, anosmia, glucose intolerance, and Wilms' tumor, but the frequency is unknown.

7. Scope covered by the guideline
Patients diagnosed with aniridia

8. Clinical Questions list
CQ1: Is keratoplasty recommended for corneal stromal opacity in aniridia?
CQ2: Is surgical treatment recommended for limbal stem cell deficiency in aniridia?
CQ3: Is surgical treatment recommended for cataract in aniridia?
CQ4: What treatment options are appropriate for high intraocular pressure and glaucoma in aniridia?
CQ5: What is recommended as low vision rehabilitation in aniridia?
CQ6: What is the recommended treatment for photophobia in aniridia?

V. Information regarding systematic review

1. Search schedule
Literature search: November – December 2018
Literature screening: December 2018 – June 2019
Evaluation of overall evidence and summary:
July – September 2019

2. Search for evidence

1) Evidence types
The search included existing clinical practice guidelines, systematic review (SR)/meta-analysis articles, and individual research articles, in that order of priority. Randomized controlled trials (RCT), non-randomized controlled trials, observational studies, and case series were included as individual research articles.

2) Database
The search was conducted in Medline (OvidSP), The Cochrane Library, and Ichushi-Web. In addition, articles that were not stored in these databases were included if cited.

3) Basic search strategy
To fully cover existing guidelines and SR/meta-analysis articles, etc., and to prevent articles from being omitted from the search, initially a general search was conducted, and then an individual search was conducted for each CQ. For all databases, the entire recording period of the database was searched unless otherwise specified. The literature search included articles in English and Japanese.

3. Inclusion and exclusion criteria for literature
If there were existing guidelines and SR articles that met the inclusion criteria, they were given priority. If there were no existing guidelines or SR articles that met the criteria, an SR was conducted independently for individual research articles (*de novo* SR). In the *de novo* SR, priority was given to RCTs that met the inclusion criteria. If no RCT met the criteria, observational studies were included. Depending on the CQ, case series and case reports were also included.

4. Evaluation method and summary of evidence
The assessment of the overall strength of evidence followed the method described in the "Minds Handbook for Clinical Practice Guide Development 2017." The integration of the overall evidence was qualitative and, where appropriate, quantitative.

VI. From preparation of the recommendations through finalization and release

1. Basic policy for the preparation of recommendations
Recommendation decisions were based on the deliberations of the guideline development group. If no consensus was reached, a vote was made. In addition to the "strength of evidence" and "balance between benefits and harms," the "diversity of patient values" and "economic perspective" were also taken into consideration for determining recommendations and their strength.

2. Finalization
An external review was conducted. Public comments were solicited, and the results are reflected in the final version.

3. Specific method of the external review
The external review committee members submitted comments individually. The guideline development group discussed whether the clinical practice guideline needed to be modified for each comment and decided on the action to be taken. Similarly, for public comments, the guideline development group discussed the need to modify the clinical practice guideline for each comment and decided on the action to be taken.

4. Plan for release
After the external review was complete and the public comments were processed, the guidelines supervisory committee decided on the final release. The release method was decided after discussions between the guideline development group and guideline supervisory committee.

(Yoshinori Oie)

References

1) **Brauner SC**, **Walton DS**, **Chen TC**: Aniridia. Int Ophthalmol Clin 48: 79−85, 2008.
2) **Lim HT, Kim DH, Kim H**: *PAX6* aniridia syndrome : clinics, genetics, and therapeutics. Curr Opin Ophthalmol 28: 436-447, 2017.
3) **Edén U, Riise R, Tornqvist K**: Corneal involvement in congenital aniridia. Cornea 29: 1096-1102, 2010.

4) **Nishida K, Kinoshita S, Ohashi Y, Kuwayama Y, Yamamoto S**: Ocular surface abnormalities in aniridia. Am J Ophthalmol 120: 368-375, 1995.
5) **Netland PA, Scott ML, Boyle JW 4th, Lauderdale JD**: Ocular and systemic findings in a survey of aniridia subjects. J AAPOS 15: 562-566, 2011.
6) **Fischbach BV, Trout KL, Lewis J, Luis CA, Sika M**: WAGR syndrome: a clinical review of 54 cases. Pediatrics 116: 984-988, 2005.
7) **Nelson LB, Spaeth GL, Nowinski TS, Margo CE, Jackson L**: Aniridia. A review. Surv Ophthalmol 28: 621-642, 1984.
8) **Edén U, Iggman D, Riise R, Tornqvist K**: Epidemiology of aniridia in Sweden and Norway. Acta Ophthalmol 86: 727-729, 2008.
9) **Oie Y, Kawasaki S, Nishida N, Kinoshita S, Sotozono C, Ohashi Y, et al.**: Research on rare and intractable diseases, Health, Labour and Welfare Sciences Research Grants; the epidemiologic research group of "Rare intractable corneal disease"; the research group of "Intractable corneal diseases on establishing standardized diagnosis and treatment"; Diagnostic criteria and severity grading for aniridia. Journal of Japanese Ophthalmological Society 124: 83-88, 2020.
10) **Neuhann IM, Neuhann TF**: Cataract surgery and aniridia. Curr Opin Ophthalmol 21: 60-64, 2010.
11) **Wang JD, Zhang JS, Xiong Y, Li J, Li XX, Liu X, et al**: Congenital aniridia with cataract: case series. BMC Ophthalmol 17: 115, 2017.
12) **Wiggins RE, Jr, Tomey KF**: The results of glaucoma surgery in aniridia. Arch Ophthalmol 110: 503-505, 1992.

無虹彩症の診療ガイドライン評価のためのアンケート調査

質問 1. あなたの眼科医としての経験年数を教えてください。

_____ 年

質問 2. あなたのご所属を教えてください。（例：大阪大学医学部附属病院）

_____

質問 3. 前眼部形成異常患者の診療にどの程度関与していますか？

年間約 _____ 例

質問 4. 貴施設にて、これまでに無虹彩症の難病申請をしたのは何例ですか？

_____ 例

質問 5. 無虹彩症の診療ガイドラインについてご存知ですか？

☐ 知っている　　☐ 知らない

質問 6. 無虹彩症の診療において、診療ガイドラインをどの程度参照していますか？

☐ かなり参考にしている　　☐ 概ね参考にしている
☐ まあまあ参考にしている　　☐ ほとんど参考にしていない
☐ 全く参考にしていない　　☐ 見たことがない

質問 7. 貴施設の無虹彩症患者の何%くらいで本診療ガイドラインに準じた診療が行われていますか？

☐ 0%　☐ 25%　☐ 50%　☐ 75%　☐ 100%

質問 8. 本診療ガイドラインに準じた診療が行われない理由は何ですか？（複数回答可）

☐ 独自の治療指針があるため　　☐ 患者側の要望のため
☐ ガイドラインに賛同できない、あるいは分からないため
☐ その他：具体的にご記載ください

質問 9. ガイドラインの使用目的は何ですか？（複数回答可）

☐ 施設内の治療の標準化　　☐ 学生・研修医・看護師などへの教育
☐ 自身の臨床疑問の解決　　☐ 研究のアイディアを探すため
☐ その他：具体的にご記載ください

質問 10. 本診療ガイドラインの以下の内容はどの程度評価できますか（役に立ちますか）？

・ＣＱの数

☐ 多い　☐ やや多い　☐ 適当　☐ やや少ない　☐ 少ない

・ＣＱが臨床現場に即している

☐ そう思う　☐ どちらともいえない　☐ 思わない

・推奨の分かりやすさ

☐ とても分かりやすい　☐ 分かりやすい　☐ どちらともいえない
☐ 分かりにくい　☐ とても分かりにくい

・解説の内容

☐ とても役に立つ　☐ 少し役に立つ　☐ どちらともいえない
☐ あまり役に立たない　☐ 全く役に立たない

・本邦の現状を加味している

☐ そう思う　☐ どちらともいえない　☐ 思わない

質問 11. 日本の無虹彩症診療において、本診療ガイドラインはどのように役に立つと思いますか？（複数回答可）

☐ 無虹彩症認知度の向上　☐ 診療の標準化　☐ アウトカムの向上
☐ 教育の向上　☐ 役に立たない　☐ 分からない

質問 12. その他、本診療ガイドラインに関してご要望などがあればお書きください。