

ANIRIDIA

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[Early phenotypic features of aniridia-associated keratopathy and association with PAX6 coding mutations.](#)

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[Management of Congenital Aniridia-Associated Keratopathy: Long-Term Outcomes from a Tertiary Referral Center.](#)

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[Recurrent heterozygous PAX6 missense variants cause severe bilateral microphthalmia via predictable effects on DNA-protein interaction.](#)

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[Long-term outcomes of donor eyes after conjunctival limbal autograft and allograft harvesting.](#)

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[\[WAGR syndrome by heterozygous deletion of the WT1 gene. Pediatric case report\].](#)

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[Anterior chamber angle in aniridia with and without glaucoma.](#)

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[Novel PITX2 Mutations including a Mutation Causing an Unusual Ophthalmic Phenotype of Axenfeld-Rieger Syndrome.](#)

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[The Cone Photoreceptor Mosaic in Aniridia: Within-Family Phenotype-Genotype Discordance.](#)

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[A Novel PAX6 Heterozygous Mutation Found in a Chinese Family with Congenital Aniridia.](#)

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[\[Identification of a novel PAX6 mutation in a sporadic case with congenital **aniridia**\].](#)

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[A family with a mild form of congenital nystagmus and optic disc coloboma caused by a novel PAX6 mutation.](#)

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[NOVEL APPROACH TO SCLERAL FIXATION OF A REPER INTRAOCULAR LENS AND ARTIFICIAL IRIS COMPLEX FOLLOWING PARS PLANA LENSECTOMY AND](#)

[VITRECTOMY FOR ECTOPIA LENTIS AND CATARACT IN A PATIENT WITH ANIRIDIA AND NYSTAGMUS.](#)

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[Haploinsufficiency of the brain-derived neurotrophic factor gene is associated with reduced pain sensitivity.](#)

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[Transscleral Fixation of Black Diaphragm Intraocular Lens in Complete Aniridia and Aphakia Due to Posttraumatic Eye Rupture: A Pilot Study.](#)

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[Functional reassessment of PAX6 single nucleotide variants by in vitro splicing assay.](#)

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WAGR

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[\[WAGR syndrome by heterozygous deletion of the WT1 gene. Pediatric case report\].](#)

Galvis-Blanco SJ, Arias-Flórez JS, Contreras-García GA.

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[In vivo epigenetic editing of Sema6a promoter reverses transcallosal dysconnectivity caused by C11orf46/Ar14ep risk gene.](#)

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[The genetic architecture of aniridia and Gillespie syndrome.](#)

Hall HN, Williamson KA, FitzPatrick DR.

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WAGRO syndrome: a rare genetic condition associated with aniridia and additional ophthalmologic abnormalities.

Ferreira MAT¹, Almeida Júnior IG¹, Kuratani DK¹, Rosa RFM^{1,2}, Gonzales JFO¹, Telles LEB³, Ferrão YA⁴, Zen PRG^{1,2}.

Author information

Abstract

Aniridia is a congenital eye disorder with a variable degree of hypoplasia or absence of iris tissue. It is caused by loss of function of the PAX6 gene and may be an isolated ocular abnormality or part of a syndrome. WAGRO refers to a rare genetic condition leading to Wilms tumor, aniridia, genitourinary anomalies, mental retardation, and obesity and is caused by a deletion of the short arm of chromosome 11 (11p), where the PAX6 gene is located. Here, we report on an 8-year-old boy with aniridia, polar cataract, and lens subluxation along with neuropsychomotor and speech delays. Karyotype evaluation showed an interstitial deletion including region 11p13-p14, confirming the diagnosis of WAGRO syndrome. In cases of aniridia, a diagnosis of WAGRO syndrome should be considered.

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[A C1976Y missense mutation in the mouse *Ip3r1* gene leads to short-term mydriasis and unfolded protein response in the iris constrictor muscles.](#)

Chen B, Qi CY, Chen L, Dai MJ, Miao YY, Chen R, Wei WE, Yang S, Wang HL, Duan XG, Gong MW, Wang Y, Xue ZF.

Exp Anim. 2020 Jan 29;69(1):45-53. doi: 10.1538/expanim.19-0007. Epub 2019 Aug 8.

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PETERS ANOMALY

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Alkatan HM, Al Dhaheri H, Al Harby M.
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Reis LM, Houssin NS, Zamora C, Abdul-Rahman O, Kalish JM, Zackai EH, Plageman TF Jr, Semina EV.

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Shah C, Sen P, Mohan A, Chandra K, Jain E.

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[Sostdc1 is expressed in all major compartments of developing and adult mammalian eyes.](#)

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[Anterior chamber angle in aniridia with and without glaucoma.](#)

Bajwa A, Burstein E, Grainger RM, Netland PA.

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Khodjaev NS, Bessarabov AN, Sobolev NP, Shkandina YV.

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Ferreira MAT, Almeida Júnior IG, Kuratani DK, Rosa RFM, Gonzales JFO, Telles LEB, Ferrão YA, Zen PRG.

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Jiang S, Baig K, Kalevar A, Choudhry N, Gupta RR.

Retin Cases Brief Rep. 2019 Mar 14. doi: 10.1097/ICB.0000000000000863. [Epub ahead of print]

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ARTIFICIAL IRIS IMPLANTS WITH ANIRIDIA

Eye (Lond). 2019 Mar;33(3):358-362. doi: 10.1038/s41433-018-0202-4. Epub 2018 Sep 12.

A new knotless technique for combined transscleral fixation of artificial iris, posterior chamber intraocular lens, and penetrating keratoplasty.

Yoeruek E¹, Bartz-Schmidt KU².

Author information

Abstract

PURPOSE:

To present a new surgical technique for treating corneal opacity and aniridia with aphakia and the results in a small consecutive case series.

METHODS:

A three-piece acrylic intraocular lens (IOL) was attached to a customized silicone iris prosthesis and fixed with three 10-0 polypropylene sutures in a knotless technique using Z-sutures after trephination of the recipient cornea. The medical records of all consecutive patients who had received a keratoplasty and an implantation of an artificial iris and IOL were reviewed.

RESULTS:

Five eyes of five patients were included in the analysis. The mean age of the patients was 46.2 years and the mean follow-up was 24.6 months. The mean best-corrected visual acuity improved from 1.36 logMAR before surgery to 0.78 logMAR after surgery during the follow-up. At the last follow-up visit, the artificial iris-IOL complex was well centered with good positioning in all cases.

CONCLUSIONS:

Management of post-traumatic aniridia combined with aphakia and corneal scars or graft failure by haptic fixation of a foldable IOL on an artificial iris combined with a simultaneous keratoplasty appears to be a promising approach, which allows to correct a complex lesion with a less traumatic and faster procedure.

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30209266

PUBLICATIONS 2019

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TRAUMATIC ANIRIDIA

[\[Clinical and theoretical substantiation of geometrical, optical and functional features of the pupil of the artificial iris\].](#)

Khodjaev NS, Bessarabov AN, Sobolev NP, Shkandina YV.
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Al-Rashidi SH.
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