



A rare case of bilateral vitreoretinopathy of Aicardi syndrome

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ABSTRACT

Purpose: To report a rare case of Aicardi syndrome presenting with concurrent peripheral retina nonperfusion with 360-degree neovascularization in the right eye and stalk tissue with a peripapillary fibrovascular membrane and tractional retinal detachment (TRD) in the left eye.

Observations: A one-month-old girl was referred for an ophthalmic evaluation to confirm the diagnosis of Aicardi syndrome due to abnormal brain magnetic resonance imaging. A 360-degree circumferential peripheral avascular retina with extensive neovascularization was present in the right eye. Stalk tissue with fibrovascular proliferation causing TRD was found in the left fundus. The retina of the right eye became quiescent after completing peripheral laser photocoagulation. The detached retina in the left eye was flattened, and the peripapillary chorioretinal lacunae became visible one year after surgical removal of the traction. In addition, the axial length growth of the left eye regained.

Conclusion and importance: This is a rare case of Aicardi syndrome with concurrent peripapillary fibrovascular traction in one eye and peripheral retina nonperfusion in the other eye. Surgical intervention is vital not only for removing the traction and flattening the retina but also for promoting continual growth of the eyeball.

1. Introduction

Aicardi syndrome is a rare genetic disorder seen almost exclusively in females, with presumed X-linked dominant inheritance.¹ It is characterized by a triad of infantile spasm, complete or partial agenesis of the corpus callosum, and chorioretinal lacunae.² Other major or supporting ophthalmic features that form part of the diagnostic criteria include optic nerve head hypoplasia, optic disc coloboma, and microphthalmia. Anterior segment ophthalmic features that have been reported are a persistent pupillary membrane,³ posterior synechia,⁴ an iris cyst,⁵ and aniridia.⁶ The published posterior segment ophthalmic findings comprise persistent fetal vasculature,⁴ optic nerve aplasia,⁷ a morning glory disc,⁸ and posterior scleral ectasia.⁹

Here, we present a rare case of an infant diagnosed with who was found to have a stalk-like fibrovascular membrane with focal tractional retinal detachment (TRD) and successfully underwent ophthalmic surgical intervention with improved anatomical outcomes.

2. Case report

A girl was born at 38 weeks of gestation with a birth weight of 2580 g via elective cesarean section. At one month old, she presented with frequent eye blinking seizure-like attacks with evidence of left-sided focal epileptic discharge on the electroencephalogram (EEG). Brain magnetic resonance imaging (MRI) showed dysgenesis and thinning of the corpus callosum (Fig. 1), dilation of the ventricles, cystic lesions at bilateral thalamostriatal grooves, hypoplastic bilateral hippocampal formation, cystic change at bilateral anterior temporal lobes and inferior frontal lobes, and loss of normal myelinated tracts at posterior capsules and dorsal brain stem. She was referred for an ophthalmic evaluation based on the impression of Aicardi syndrome. To confirm the diagnosis of Aicardi syndrome, genetic testing was performed and demonstrated a homozygous TREX1 gene mutation (c.292_293insA and p. (Cys99Metfs)).

The ophthalmic examination under anesthesia (EUA) at one month of age revealed poorly developed retinal vasculature that was mildly

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Fig. 1. Magnetic resonance imaging (MRI) of the patient. Sagittal T1-weighted MRI shows thinning of the corpus callosum (white arrows).

tortuous with no significant dilatation, the presence of an inferior preretinal hemorrhage, and a peripheral 360-degree avascular zone in the right eye (Fig. 2A). The left eye featured stalk tissue and focal TRD involving the superior peripapillary area (Fig. 2B). Fundus fluorescein angiography (FA) demonstrated extensive 360-degree neovascularization with late hyperfluorescence over the retina vascular-avascular border in the right eye and focal hyperfluorescence over the peripapillary stalk tissue in the left eye (Fig. 2C and D). Her right eye underwent indirect peripheral laser photocoagulation over the avascular retina, and the left eye simultaneously underwent 23G-system pars planar vitrectomy to release the peripapillary fibrovascular traction.

In the follow-up EUA performed at 13 months of age, the right retina was quiescent post photocoagulation, and there was no recurrent fibrous formation over the left retina, with reattachment of the affected retina. The most significant postoperative finding after removal of the overlying

fibrovascular membrane and flattening of the underlying retina was the identification of peripapillary chorioretinal lacunae in the left eye (Fig. 3A) within the corresponding region. From the subsequent EUA performed at 26 months of age, her right retina remained stationary, while peripapillary chorioretinal lacunae in the left eye increased in size and number (Fig. 3B).

Prior to vitrectomy at one month of age, the axial length of the left eye was 1.03 mm shorter than that of the right eye. EUA at 13 months and 26 months of age showed axial length development, and the spherical equivalent of the left eye was almost comparable to that of the right eye (Table 1). Intraocular pressures were within normal ranges bilaterally. Over the last two and a half years, her fix-and-follow vision became bilateral, and her ambulatory ability developed independently.

3. Discussion

In our case report of Aicardi syndrome, we emphasized the rare ophthalmic finding of a peripapillary stalk with focal TRD and successful attachment of the retina with continual development of the eye following vitrectomy. The structure that protrudes forward into the vitreous could be a persistent fibrovascular stalk due to incomplete regression of the hyaloid artery. With time, this broad-based stalk causes traction on the retinal layer, leading to the formation of retinal folds and retinal tenting, as seen in our patient. From the literature review, only a single case report has documented the presence of unilateral epipapillary and peripapillary gliotic tufts, where it was described as persistent fetal vasculature.⁴ In contrast to our patient, the patient described in the previous report was treated conservatively.

At the age of 1 month, our patient underwent 25G vitrectomy to release the fibrous traction. The aim of surgery was to regain the normal anatomical structure of the patient's left eye. After the surgery, the affected retina was flattened, with no recurrence of fibrous membrane formation. The recovery of the retinal structure even contributed to unmasking the clinical features of peripapillary chorioretinal lacunae (Fig. 3), which helped to confirm the diagnosis of Aicardi syndrome in this child by fulfilling the classical triad of this disorder.¹⁰ Although the patient's eyes were not microphthalmia at birth, the axial length in the left eye was shorter than the right eye. Removal of the traction also

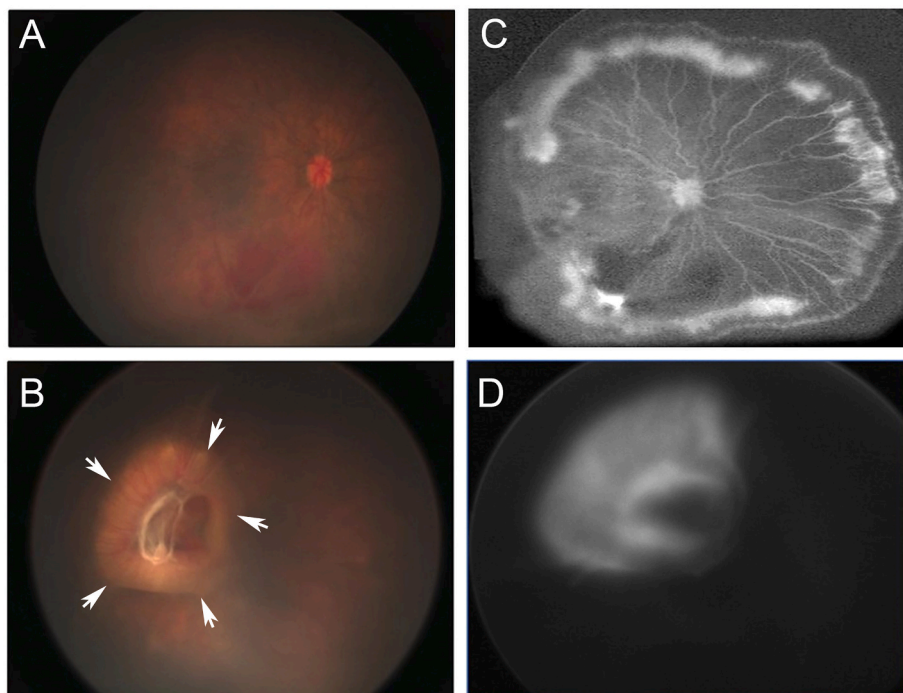


Fig. 2. Fundus photographs and fluorescein angiography of the patient at 1 month old: (A) Tortuosity of the retinal vessels and inferior preretinal hemorrhage were noted in the right eye with 360-degree peripheral retina avascularity. (B) A peculiar looking stalk with fibrovascular proliferation (white arrows) and abnormal vitreoretinal traction accompanied by tractional retinal detachment was noted in the left eye. The macula could not be identified, and retinal vessels were not apparent. (C) Tortuosity of the retinal vessels, absence of the foveal avascular zone, and extensive neovascularization near the junction of the vascular and avascular regions were seen in the right eye. (D) Hyperfluorescence of the fibrovascular stalk and absence of the normal retinal vasculature were noted in the left eye.

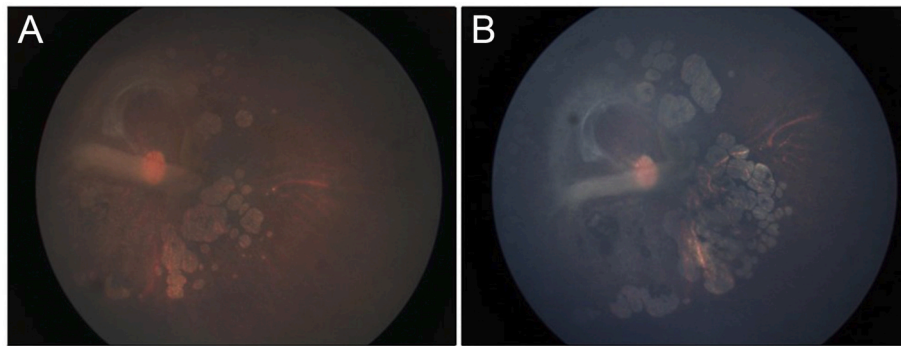


Fig. 3. Follow-up left fundus photographs of the patient (A) at 13 months old: showing classic peripapillary chorioretinal lacunae, and (B) at 26 months old: showing the progression of the morphology of the peripapillary chorioretinal lacunae in terms of size and number.

Table 1

Profile of the axial length and spherical equivalent at different ages.

Age (Months)	Axial length (mm)		Spherical equivalent (diopters)	
	OD	OS	OD	OS
1	18.42	17.45	-6.50	-6.75
13	21.64	22.44	-12.75	-14.50
26	24.84	24.41	-11.50	-8.75

allowed the left eye to grow in parallel with the right eye, as evidenced by continual elongation of the axial length (Table 1), thus preventing the sequelae of microphthalmia and possible phthisis bulbi.

Unilateral peripheral retinal nonperfusion and anomalous retinal vasculature in the right eye, as observed in this child (Fig. 2A), have been reported in the previous publication.⁶ However, the two patients had a poor visual prognosis (no perception of light [NPL]) due to the sequelae of chronic retinal detachment, with one of the patients eventually developed phthisis bulbi.⁶ In contrast, in our patient, the abnormalities could be identified very early (during EUA at one month of age), and peripheral laser photocoagulation was completed after the FA demonstrated late hyperfluorescence due to neovascularization (Fig. 3A). As a result, the right eye fundus remained quiescent, and a bilateral visual acuity of at least fixed-and-follow vision was documented at the age of two. Loss of peripheral retinal vascularization was associated with congenital optic anomalies, and retinal laser photocoagulation of the avascular retina could prevent further proliferative retinopathy.¹¹

Concerning the ophthalmic findings in Aicardi syndrome, the two most common features are chorioretinal lacunae (which was present in our case) and optic nerve coloboma.³ Both unilateral and bilateral chorioretinal lacunae have been reported in many case series and case reports.^{3, 6} These features are commonly found within the peripapillary area and are characterized by retinal pigment epithelium excavation through the choroid layer into the bare scleral layer.³ In our patient, from one year to two years of age, the morphology of her left unilateral peripapillary chorioretinal lacunae changed in terms of size and number (Fig. 3A and Fig. 3B), with no clinically associated retinal edema or subretinal fluid.³

For the past 20 years, advancements in imaging techniques and improved histopathological evaluations have led to the accumulation of other phenotypic variances in Aicardi syndrome.³ For example, our patient was found to have thinning of the corpus callosum instead of the agenesis corpus callosum.⁶ Aicardi syndrome is commonly reported to be associated with multiple seizure types.¹² In our patient, the eye blinking episodes were likely an atypical absence seizure, which has been reported in the form of staring spells.¹ In addition, spasticity was also noted at 1 year old during clinical follow-up.

4. Conclusion

In conclusion, we highlighted a rare case of Aicardi syndrome with two different rare phenotypic variances in two different eyes: a peripapillary fibrovascular stalk with focal TRD and peripheral retinal nonperfusion with anomalous retinal vasculature. In addition, treatments performed on both eyes allowed complete attachment of the retina, favorable axial length development, and ambulatory vision for the patient. Without prompt evaluation and treatment, the retinal pathologies may provide a major source of morbidity.

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Authorship

All authors attest that they fulfil the current ICMJE criteria for authorship.

Patient consent

Informed consent was obtained from the parents of the patient.

Declaration of competing interest

None.

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None.

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