

## Case Report

# Optic Nerve Cyst Associated with Optic Disc Pit in a Child with Multi-Cystic Kidney

Verlyn Yang<sup>1\*</sup>, Sandeep Bhuta<sup>2</sup>, and Aditi Kirpalani<sup>1</sup><sup>1</sup>Department of Ophthalmology, Gold Coast University Hospital, Australia<sup>2</sup>Department of Radiology, Gold Coast University Hospital, Australia**\*Corresponding author**

Verlyn Yang, Department of Ophthalmology, Gold Coast University Hospital, Australia, Email: yangverlyn@gmail.com

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- Optic nerve cyst
- Optic disc pit
- Coloboma
- ODP-M

**Abstract**

In this case report, we present an unusual case of a 7-year-old female who came to the attention of the Ophthalmology department for reduced vision in the right eye 6/18 (LogMAR 0.4) with normal vision in the left eye. Fundoscopic examination revealed signs of an optic disc coloboma associated with an optic disc pit with adjacent sub-retinal fluid extending to the macula. She was born with a left multi-cystic kidney. Further work-up in the form of MRI and USG Orbits demonstrated a well-defined optic pit at the optic nerve head and a tubular channel extending to a 6-7mm multi-cystic lesion.

**ABBREVIATIONS**

Optic disc pit maculopathy (ODP-M); Magnetic Resonance Imaging (MRI)

**INTRODUCTION**

Congenital optic disc pit is an uncommon congenital condition occurring in about 1 in 11,000 people with no known risk factors [1]. There has been suggestion of possible autosomal dominant inheritance but no genes have been identified as of yet [2]. Reported incidence of bilateral optic disc pits is 10-15% [3]. Often, the optic disc pit is detected in an asymptomatic patient undergoing routine ophthalmic fundus examination. The pits are generally found infero-temporally within the nerve or centrally. They appear to be a round or oval depression differing in pigmentation from the nerve [3]. Patients become symptomatic with visual deterioration when optic disc pit maculopathy (ODP-M) develops including the collection of intra- and sub-retinal fluid and retinal pigment changes [4]. ODP-M develops in 25-75% of patients with ODP often in their 3<sup>rd</sup> and 4<sup>th</sup> decades of life [5] but cases of paediatric ODP-M have been reported [6]. The association of a cyst adjacent to the ODP is highly unusual and has only been isolated to case reports [7,8].

**CLINICAL PRESENTATION**

A 7-year-old girl was referred to the Ophthalmology department for reduced vision in her right eye. She was diagnosed with a left multi-cystic kidney at birth. This had regressed effectively, leaving her with a solitary functioning kidney. Parents reported that she had been struggling at school and was diagnosed with learning difficulties. She was also being treated for Attention deficit hyperactivity disorder (ADHD). Her

mother had been diagnosed with Stragardt's disease. There were no other systemic, familial or genetic conditions of note.

On examination, her visual acuity was noted to be right eye: 6/18 (LogMAR 0.4), left eye: 6/9 (LogMAR 0.1), with no refractive error. Significantly, she was found to have an anomalous optic nerve appearance in the right eye on fundus examination. There were signs of an optic disc coloboma with an ODP visualized. Left eye was unremarkable.

**INVESTIGATIONS/IMAGING FINDINGS**

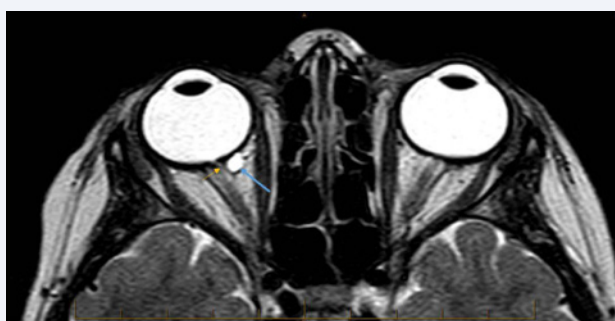
MRI of the brain and orbits was organized to exclude structural abnormalities of the brain and to better visualize optic nerve abnormalities. MRI brain was normal and the optic nerve pit was well-visualized. However, the pit extends in a tubular fashion to form a peripapillary cyst as illustrated in Figure 1. The appearance is distinct from that expected of a morning glory syndrome. The cyst appears to be of 6-7mm in size.

Ultrasound imaging of the orbit, further characterized the ODP measuring at 1.1mm (Figure 2a). It also shows a linear hypoechoic tract extending from the pit into an irregular hypoechoic cyst (Figure 2b).

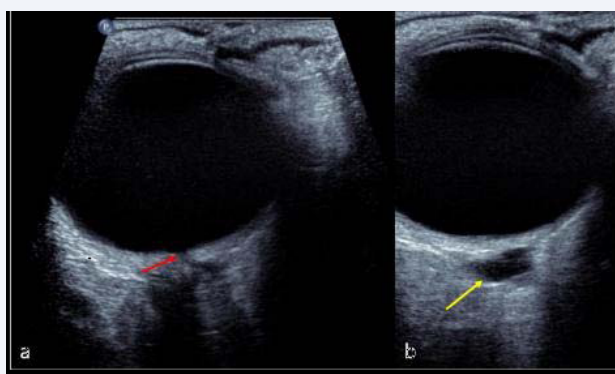
Color fundus photograph shows an ODP on the temporal aspect of the disc and a coloboma, as illustrated in Figure 3.

**DIFFERENTIAL DIAGNOSES**

The presence of an ODP could be a variant of an optic disc coloboma and an isolated finding in a well-child but it has also been associated with congenital anomalies such as renal hypoplasia, Aicardi syndrome amongst others<sup>4</sup>. In this patient, we seek out a possible unifying diagnosis with differential diagnoses



**Figure 1** T2 axial image of the Right optic nerve pit, tubular extension (orange arrow) into a cystic structure which enhances with contrast (blue arrow).



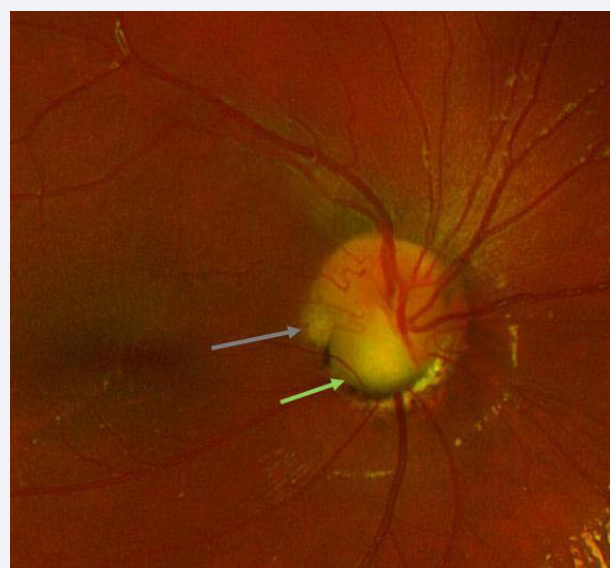
**Figure 2** Ultrasound orbit. (a) ODP measuring 1.1mm and linear hypoechoic tract. (b) hypoechoic cyst.

including PAX2-related disorder (or synonym: Papillorenal syndrome), PAX6-related disorder, CHARGE syndrome and Cat-eye syndrome, amongst others<sup>9</sup>. The patient has been worked up from a genetic point of view. However, there have been no significant genetic causative mutations identified.

Another differential diagnosis is the morning glory disc but that is easily ruled out clinically.

The ODP is typically found in an asymptomatic individual but can cause visual problems when the macula is involved, termed Optic Disc Pit – Maculopathy (ODP-M). In this case report, the patient had sub-retinal fluid associated with the ODP and intra-retinal fluid in the perifoveal region, which is the likely cause of her reduced vision in the right eye.

Multiple hypotheses exist to explain the source of fluid in ODP-M. The most common theory relates to vitreous associated with vitreous liquefaction, supported by the observation of ODP-M occurring commonly between ages 30-40 [10,11] and small studies showing spontaneous resolution of ODP-M with completion of posterior vitreous detachment (PVD) [12]. Other hypotheses include cerebrospinal fluid (CSF) communication between the subarachnoid and subretinal spaces [13]. Other theories relate to the concept of blood vessel leakage at the ODP and Bruch's membrane but have been disproved respectively [14,15]. The most popular theory relating to vitreous traction does not hold in the paediatric population as PVD does not occur in this age group [16].



**Figure 3** Colour fundus photograph demonstrating ODP on temporal aspect of disc (grey arrow) and coloboma inferiorly (green arrow).

## TREATMENT

Many treatment options have been discussed in literature including peripapillary laser, pneumatic displacement or pars planar vitrectomy with limited efficacy and drawbacks<sup>17, 18</sup>. Our child remained clinically stable over the last 12 months, with no significant change of visual acuity and findings on OCT-M, with no intervention. We will continue to observe her at regular intervals.

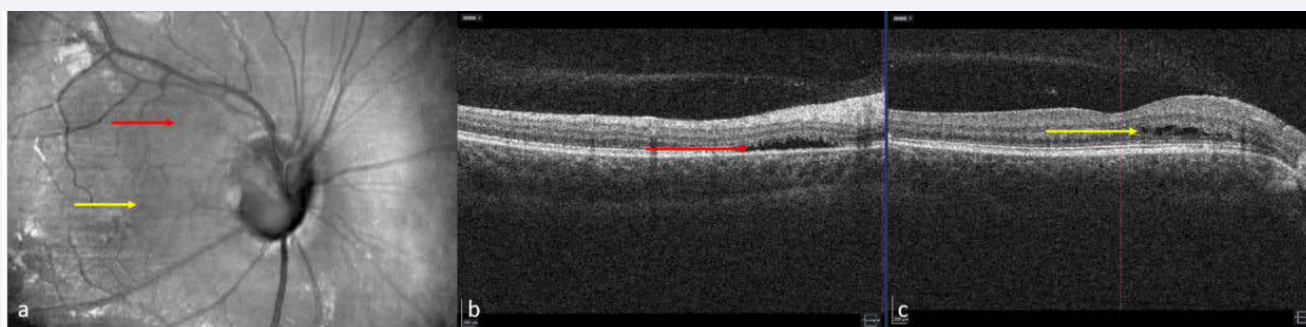
The presence of an optic nerve cyst in association with an ODP is extremely rare and has only been mentioned in two case reports in literature till date<sup>7,8</sup>. In the report by Lee et al.<sup>7</sup>, the patient had compressive optic neuropathy with electrophysiology changes and visual field defect to complement resulting from a 6x6mm optic nerve cyst within the sheath. In our case, the ODP cyst was not found to be within the sheath. Our child was too young to perform a reliable visual field test and the likelihood of positive findings in her are low. However, that would be attempted when she gets older.

## CONCLUSION

In conclusion, we report an unusual case of a young child with ODP-M with co-existing congenital renal abnormalities for which a unifying diagnosis is sought. The incidental radiological findings of a cyst in this child will prompt ongoing monitoring to ensure the cyst does not grow to a size that would cause compressive optic neuropathy. Neuroimaging should be considered for patients with atypical optic nerve head appearances if they have visual field defects.

## LEARNING POINTS

- Congenital optic disc pit is an uncommon congenital condition which may be hereditary but the genes have not yet been identified.
- Optic nerve cysts are a rare entity.



**Figure 4** OCT imaging. (a) OCT Disc demonstrates the ODP and coloboma. (b) cross section of the retina demonstrating sub-retinal fluid (red arrow), corresponding to area marked with red arrow in (a), associated with the ODP; (c) cross section of the retina at the fovea demonstrating perifoveal intra-retinal fluid (yellow arrow), corresponding to area marked with yellow arrow in (a).

- The clinical significance is the potential for compressive optic neuropathy; hence, it should be identified if present.
- Optic disc pits can have maculopathies associated with it and cause reduction in vision.
- Patients with such findings should be referred to an Ophthalmologist for clinical correlation of imaging findings.

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## Cite this article

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