Red cataract in propionic acidaemia

B Staniszewski[1], S Patel[1], S Burgess[2]

1. Department of Ophthalmology, Ninewells Hospital and Medical School, Dundee
2. Department of Ophthalmology, NHS Forth Valley, Falkirk Community Hospital, Falkirk

Systemic and Ophthalmic Manifestations of Propionic Acidaemia

Propionic acidaemia is a rare autosomal recessive disorder (incidence, less than 1 in 100,000) that causes chronic metabolic decompensation with paroxysmal ketoacidosis, failure to thrive and developmental delay. The condition is caused by deficiency in propionyl-CoA carboxylase which plays a role in the normal breakdown of proteins, fat and cholesterol. As a result, a substance called propionyl-CoA and other potentially harmful compounds accumulate in mitochondria, causing bone marrow suppression and metabolic acidosis.

With timely clinical intervention and improved management, patient survival time has lengthened considerably in recent years.

Several studies have previously demonstrated an association between propionic acidaemia and optic nerve atrophy [1,2]. There have been no reports of lens opacities in patients with propionic acidaemia described in literature.

Aim

To report an unusual case of red cataracts in association with propionic acidaemia and discuss its management.

Case Presentation

26 year old male with a diagnosis of propionic acidemia first presented to the eye clinic for visual assessment at the age of 10. His visual acuities (VA) were 6/9.5(OD) and 6/38(OS). Fundal examination showed mild temporal pallor of the left optic nerve head. He subsequently developed progressive optic nerve atrophy in both eyes. Due to patient's learning difficulties, VA assessment has been challenging and variable VA have been documented during his clinic visits. The patient has received multiple sessions of hyperbaric oxygen therapy with reported improvement to wellbeing and quality of vision by patient's mother. The patient has gradually developed bilateral, symmetrical lenticular opacities of unusual morphology. Both lenses had a distinct red/amber discolouration. Following a discussion of risks and benefits, we have proceeded with left cataract surgery.

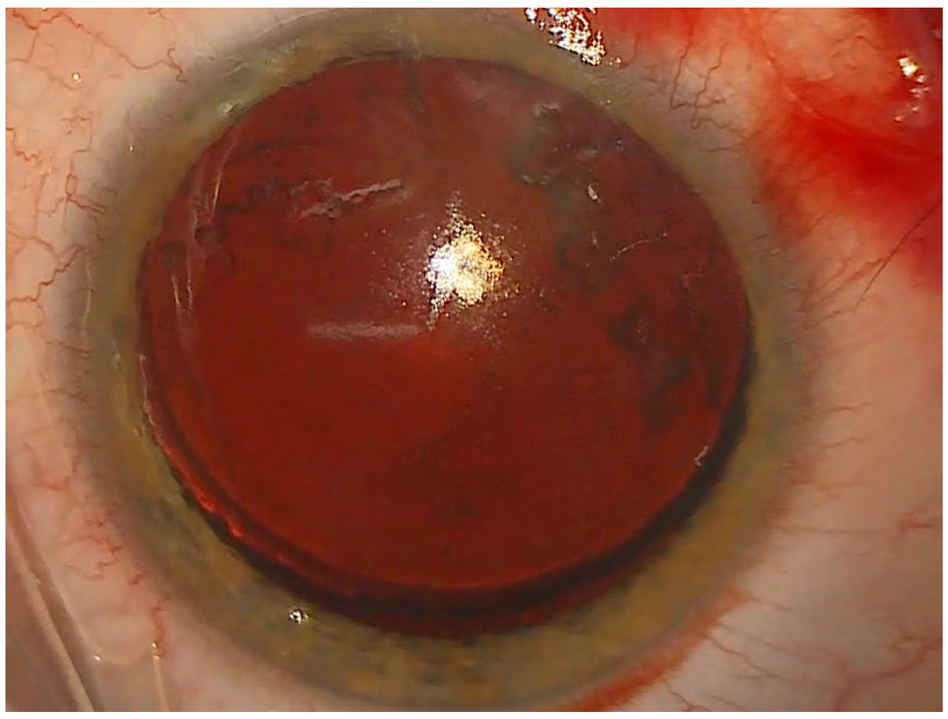

Fig1. Left cataract at the beginning of the operation

Cataract Surgery

Left phacoemulsification and IOL insertion was performed under sub-Tenon's local anaesthesia. The surgery resembled a paediatric cataract extraction. The anterior capsule was elastic. Vision blue and high viscosity cohesive viscoelastic (Microvisc plus) were used. The crystalline lens was soft and removed with aspiration only.

There were no intraoperative complications and VA(OS) documented preoperatively as 60 ETDRS letters at 2m improved to 0.05 Sonksen LogMAR visual acuity at 1 week postoperatively.

More importantly, both the patient and his mother have reported a significant improvement to patient's quality of vision and visual alertness.

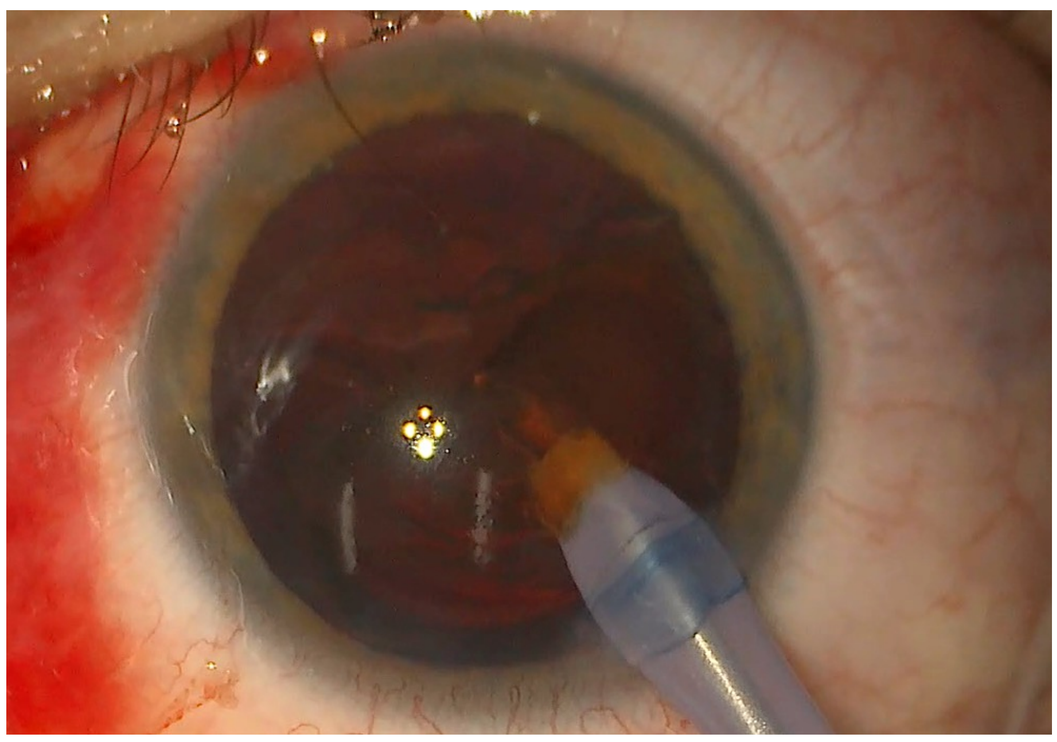

Fig 2. Aspiration of lens material

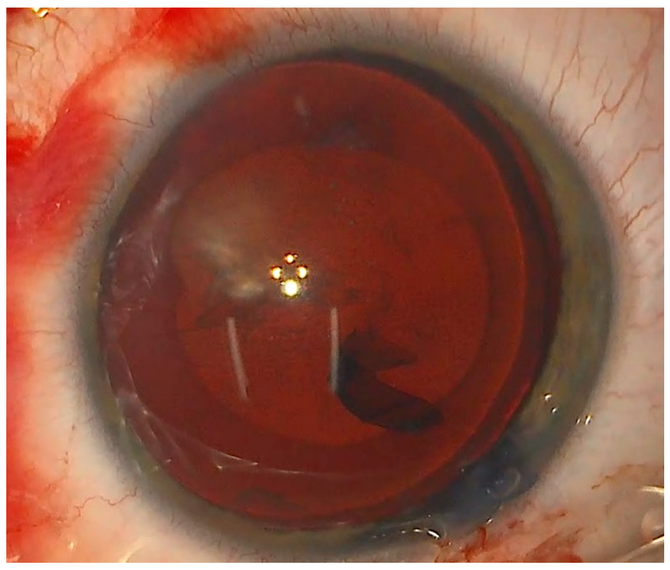

Fig 3. Capsulorrhexis

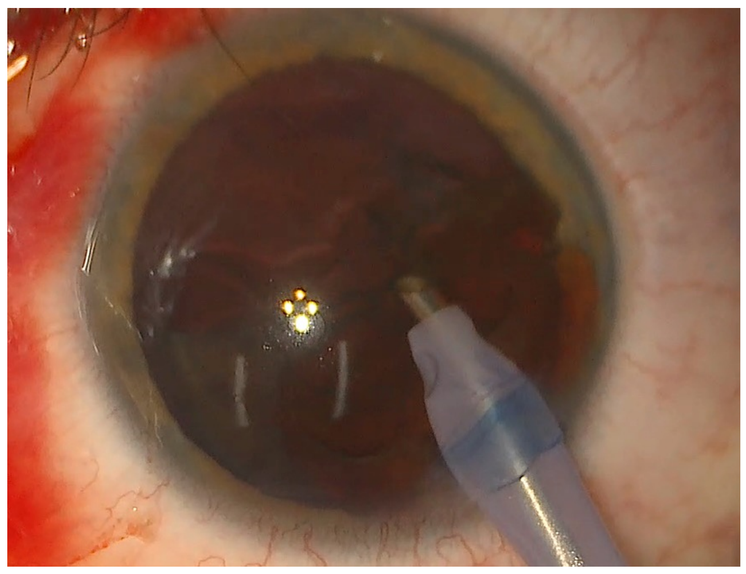

Fig 4. Aspiration of lens material

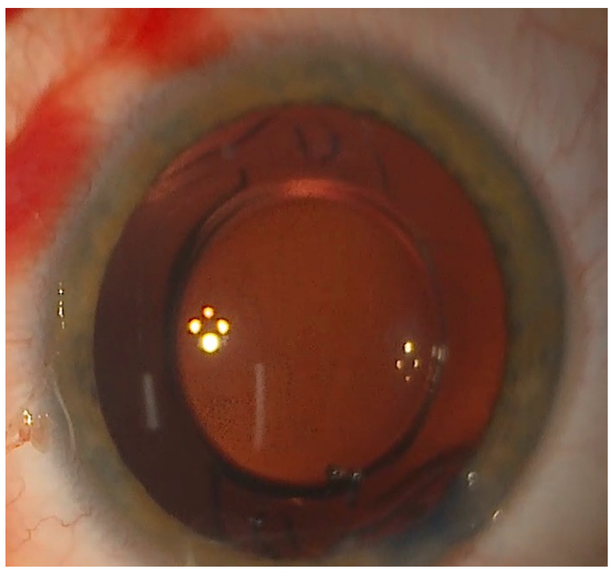

Fig 5. Posterior chamber lens implant

References:

1. Ianchulev T, Kolin T, Moseley K, Sadun A. Optic nerve atrophy in propionic acidemia. Ophthalmology. 2003 Sep;110(9):1850-4.
2. Martinez Alvarez L, Jameson E, et al. Optic neuropathy in methylmalonic acidemia and propionic acidemia. Br J Ophthalmol. 2016 Jan;100(1):98-104.