Diagnosing the oil drop: A case report and review of the literature

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Galactosemia is a rare autosomal recessive condition caused by one of the three defective enzymes, galactose 1 phosphate uridyl transferase (GALT), galactokinase and/or epimerase, involved in galactose metabolism. Cataract formation is consequent to accumulation of galactitol in the crystalline lens.[1] In classic galactosemia, galactitol also accumulates in liver and brain causing hepatomegaly, liver failure, lactose intolerance, hypoglycemia, sepsis, and mental retardation. Duarte variant with partial enzyme activity is a milder form of galactosemia and is often undiagnosed.

Although a rare cause of cataract, timely recognition is important as dietary restriction of lactose-containing foods in early stages can reverse cataract formation and minimize adverse effects of galactitol on liver and brain. Diagnosis of galactosemia rests on the presence of galactose metabolites in urine and blood, enzymatic assay, and mutation analysis. Gold standard is GALT activity measurement in red blood cells.[2] In our case, the typical appearance of cataract with history of chronic diarrhea prompted the diagnosis and was confirmed by cataractous lens protein analysis.

A 3-year-old male child was brought by parents with complaints of recurrent episodic abdominal pain, chronic diarrhea, and failure to thrive since early childhood. Elsewhere, the child was diagnosed to have malabsorption syndrome and was thoroughly investigated including jejunal biopsy to rule out celiac sprue. Lately, parents had noticed a white reflex in both eyes which kept on progressing. On general examination, the child was drowsy, apathetic with generalised wasting. Mental functions seemed age appropriate. The lens showed a central typical oil droplet like amber-colored opacity within the substance of nucleus in both eyes [Fig. 1]. Fundus was normal. Digestive history and distinctive lenticular opacities were highly suggestive of galactosemic cataract.

Urine for reducing sugars was negative. Enzyme assay was advised but parents were not willing for any additional tests. Child's general condition improved significantly after galactose-free diet; child gaining 2 kg in 3 weeks. Lenticular opacities did not show any regression after dietary restriction and child underwent bilateral cataract extraction with intraocular lens implantation. Lens aspirate sample thus obtained and galactose solution of 1 mg/mL was reconstituted, processed similarly and subjected to electophoresis analysis at Institute of Chemical Technology. Galactose peak was observed at 21.16 min in both sample and galactose solution [Fig. 2] establishing the diagnosis of galactosemia.

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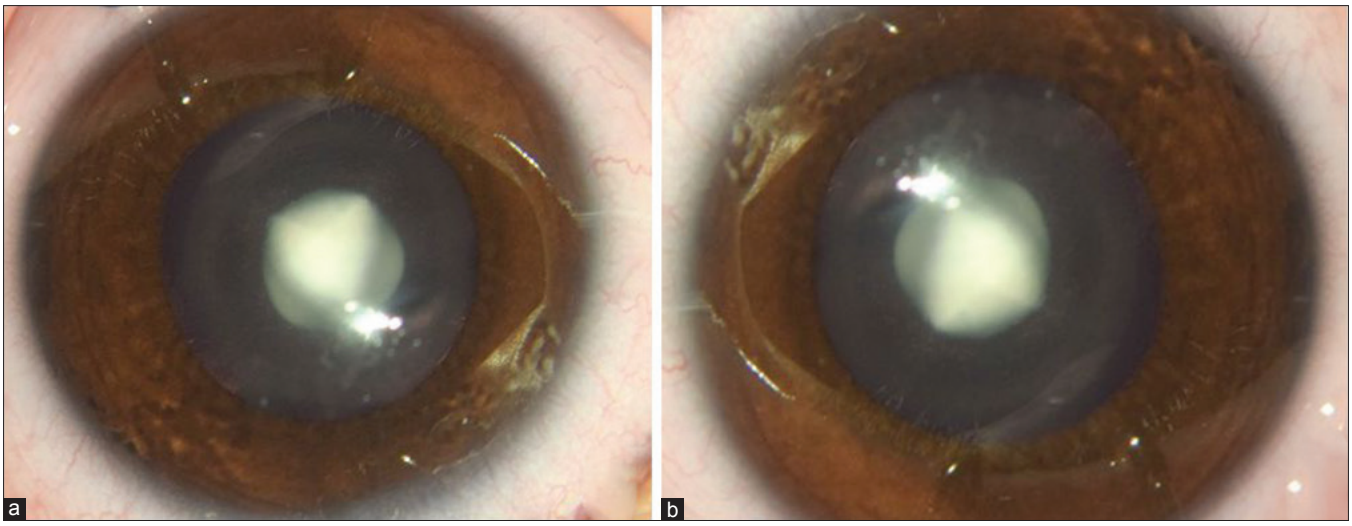

Figure 1: Digital photograph of right eye (a) and left eye (b) of the patient showing classical oil droplet cataract

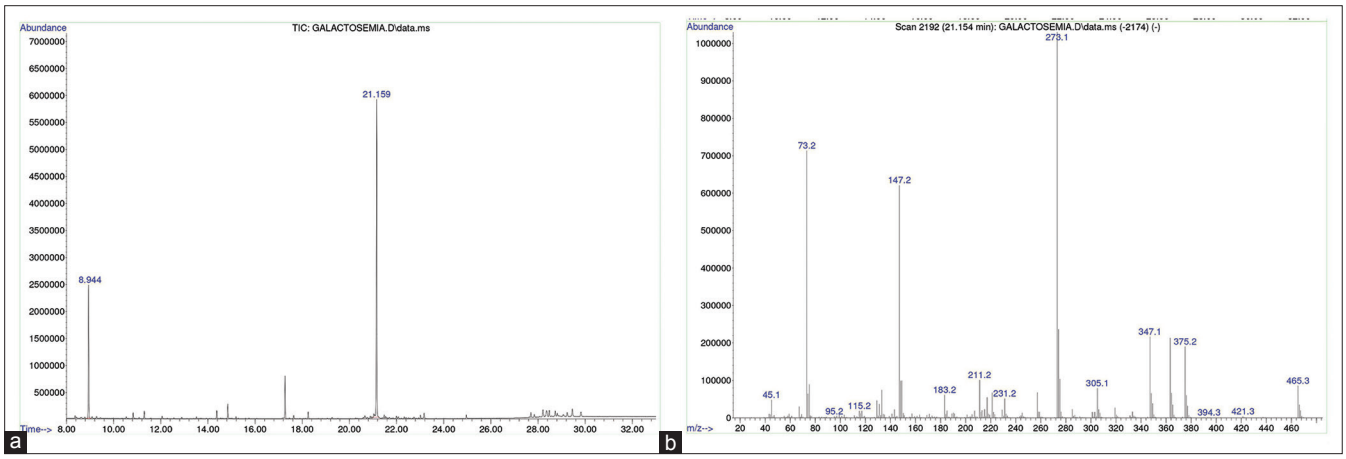

Figure 2: Galactose peak was observed at 21.16 min present both in the standard galactose (a) and galactosemia sample (b)

In conclusion, diagnosis of galactosemia can be established by cataractous lens protein analysis where other investigations are not possible.

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Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

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