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BILATERAL NUCLEAR CATARACTS IN FANCONI-BICKEL SYNDROME

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Fanconi-Bickel syndrome (FBS) or glycogenosis type XI, is a rare inherited metabolic disorder with autosomal recessive inheritance. It is caused by a defect of GLUT2, a facilitative glucose transporter expressed in liver and kidney. Its clinical features are those of hepatic glycogenosis together with a Fanconi-type nephropathy. Although defective hepatic uptake of galactose results in hypergalactosemia, cataracts are not a common feature. They have only been reported anecdotically in 3 patients without any clinical details. Here, we present a female patient with FBS who had a positive screening test for galactosemia and who was started on a galactose free-diet at 20 days of age. Classical causes for galactosemia were ruled out. At the same age, bilateral nuclear cataracts were first detected. The diagnosis of FBS was first made clinically and recently confirmed by molecular analysis of the GLUT2 gene (G318R/G318R). The patient is now 17 year old and bilateral cataracts have not changed. Among some other genetic conditions, hypergalactosemia is well known to cause nuclear cataracts early after birth. In contrast to classical galactosemia patients, cataracts in this case have not reversed on a galactose-free diet. The underlying pathogenetic mechanisms leading to irreversible cataracts only in few patients with FBS still need further elucidation.

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