

Bardet–Biedl Syndrome: Delayed Diagnosis in a 14-Year-Old Child with End-Stage Renal Disease

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February 27, 2023

Abstract

Bardet–Biedl syndrome (BBS) is a rare autosomal recessive ciliopathic disorder. Because of its low prevalence and wide spectrum of clinical features, many patients remain undiagnosed. We report a case of a 14-year-old boy with a typical phenotype of BBS who remains undiagnosed until the development of end-stage renal disease.

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BBS_Manuscript_Rasel_Feb_2023.docx available at <https://authorea.com/users/590344/articles/626736-bardet-biedl-syndrome-delayed-diagnosis-in-a-14-year-old-child-with-end-stage-renal-disease>





