

## The Role of Vitamin D in the Assessment of Treatment Response in Children with Gaucher Disease

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### ABSTRACT

Gaucher disease (GD) is a hereditary autosomal recessive disorder. Numerous biomarkers that have a role in the pathophysiology and etiology of GD are used for the diagnosis and prognosis of this disorder in pediatric populations. The study sought to examine the significance of vitamin D levels in assessing the treatment response of individuals with GD receiving enzyme replacement therapy (ERT) during follow-up evaluations. Case-control research was conducted including 70 children (both male and female) aged 1 to 13 years diagnosed with Gaucher disease, recruited from the Pediatric Department and the Unit of Rare Diseases. The vitamin D levels were assessed in samples from Gaucher patients classified as newly diagnosed GD patients that didn't receive treatment (n=7), those undergoing ERT for 3-6 months (n=20), 6-12 months (n=20), and patients received ERT for over one year (n=23), and these levels were compared with those of twenty gender- and age-matched control subjects. The investigation was conducted from December 2023 to May 2024. The vitamin D levels are evaluated using an enzyme-linked immunosorbent test (ELISA) kit. The data revealed that levels of vitamin D in patients were considerably lower ( $p<0.05$ ) compared to age-matched controls. It was concluded that Vitamin D showed to have a diagnostic value in newly diagnosed GD patients that didn't receive treatment with a poor monitoring role as it elevated slowly with the treatment progress.

**Keywords:** Enzyme replacement therapy, Gaucher disease, imiglucerase, lysosomal storage disorder, Vitamin D.

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