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Abstract Topic:- Clinical Genetics

Abstract Title:- Significance of diagnostic biomarker (lyso-Gb1) in Gaucher disease of Indian population.

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Aims:-To check the levels of Lyso-GB1 in treated, untreated and controls in serum sample of Gaucher disease patient using human glucosyl sphingosine (lyso-Gb1) ELISA kit.

Methods:- To check the levels of lyso GB1 in treated, untreated and controls in serum sample of GD patient human glucosyl sphingosine (lyso GB1) ELISA kit is used. Whole blood (2ml) was collected and centrifuged at 3000rpm for 10 min at 4 degree Celsius. Supernatant (serum) aliquoted & stored in -80 degree Celsius. Confirmation was done with human glucosyl sphingosine ELISA kit. OD is measured spectrophotometrically at 450nm to calculate concentration of lyso GB1. Statistical analysis was done. The ELISA kit uses sandwich ELISA as the method. The microelisa stripplate provided in this kit has been precoated with antibody specific to lyso G1. The optical density is measured spectrophotometrically at a wave length at 450 nm. The OD was found proportional to the concentration of Lyso gb1

Results:-

The levels of lyso GB1 is highly elevated in untreated patients and modest elevation was seen in treated patients. There was very little concentration of Lyso gb1 in control sample.

Conclusions:- lyso-Gb1 is a specific and sensitive biomarker at diagnosis.

The need for a more reliable biomarker for GD and disease progression led to identifying the deacylated form of accumulated glucosylceramide, lyso-Gb1. Lyso-Gb1 is a direct metabolite of GBA and plays an essential role in disease-related pathology.

Thus, we can use lyso-Gb1 as a potential biomarker for mass screening to detect Gaucher disease in developing country like India.

Keywords:- Gaucher disease, biomarker, glucosyl sphingosine, lyso-GB1, lysosomal storage disorder