A Novel Globotriaosylceramide Quantification Assay for the Rapid Diagnosis of Fabry Disease

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RESEARCH HIGHLIGHTS

- Current diagnostic approach for Fabry disease is insensitive, low-throughput, and costly.
- Using rat anti-human Gb₃ monoclonal antibody, a novel Gb₃ quantification assay has been developed in this study.
- Accuracy, precision, and sensitivity of the novel assay was verified using crude cellular lipid extracts.
- The novel assay serves as a cost-effective and rapid approach for the diagnosis of Fabry disease.

1 Introduction

Fabry disease
Systemic accumulation of lipid (Gb₃)¹,²

Genetics
Mutation in GLA gene (Xq22.1)²

Statistics
Affects 1:40,000 males annually³

2 Problem statement

Current diagnosis
Enzyme activity assay & Gb₃ quantification³,⁴

Problems
No early diagnosis & insensitive³,⁵,⁶

3 Research objectives

Assay development
To develop a rapid Gb₃ quantification assay

Method validation
To characterize the novel assay

5 Preliminary findings

Accuracy
85.62 % to 131.22 %
(n = 9)

Sensitivity
90.55 % to 115.70 %
(n = 9)

Response rate
R² = 0.9187
(n = 3)

Gb₃ (green) in Fabry skin fibroblast
Gb₃ (green) in healthy skin fibroblast

CONCLUSIONS

- A novel Gb₃ quantification assay was developed using rat anti-human Gb₃ monoclonal antibody.
- Gb₃ amounts were successfully quantified in three human cell lines.
- Further studies can be done on quantifying Gb₃ using human biological samples.

REFERENCES