Introduction

Multiple organ dysfunction occurs in Mitochondrial diseases (MDs). MDs are sometimes difficult to diagnose, because patients have solitary and/or combination of various symptoms: seizure, myopathy, heart failure and diabetes. Since plasma levels of lactate and pyruvate are not always the perfect biomarker for MDs, there are many pseudo-mitochondrial patients who are suspect for MDs. To minimize the number of specimens from such pseudo-mitochondrial patients is very important issue for diagnostic center. In 2011, Serum fibroblast growth factor 21 (sFGF21) was launched as a biomarker to diagnose muscle-featured MDs. Owing to this, it is now possible to differentiate MDs from similar other diseases using sFGF21.

Purpose

We investigated whether serum growth differentiation factor 15 (sGDF15) can be a more specific biomarker for MDs.

Methods

Blood was extracted from 48 MDs patients, 146 controls, 6 Duchenne muscular dystrophy (DMD), and 8 severe heart failure (HF) patients as disease controls. SFGF21 and sGDF15 was measured using ELISA (Biovendor, Czech and R&D systems, USA). Statistical analysis was performed in Mann-Whitney test, Kruskal Wallis test, and ROC curve analysis using SAS ver.3.3 (SAS institute Inc., USA).

Results

![Graphs showing serum levels of GDF15 and FGF21 in different conditions.](image)

- **A**: SFGF21 level of each mitochondrial disease. SFGF21 level was significantly higher in all mitochondrial diseases (MDs) than in controls. (B) SFGF21 level between all MDs and controls. SFGF21 level was significantly higher in all MDs than in controls. Severe heart failure patients were also elevated expectedly. (C) SGDF15 level of each mitochondrial disease. SGDF15 level was significantly much higher in all MDs than in controls. (D) SGDF15 level between all MDs and controls. SGDF15 level was significantly much higher in all MDs than in controls.

![Graph showing ROC curve for GDF15.](image)

- **E**: ROC curve. GDF15; AUC=0.99, sensitivity=97.9%, specificity=95.2%, cut off value=707.4 pg/mL.

Conlusion

We investigate that sGDF15 is the new and useful biomarker for MDs, which is more advantage for diagnostic tool than that in sFGF21.

References


(This study is in submitting)