Monitoring of Fabry Disease Treated by Migalastat in one Family with Amenable Mutations

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Biochemical Tests of Amenable Cases in One Family with pD33Y mutation (Fig. 1)
1) α-galactosidase activities in DBS, lymphocytes, and plasma lysoGb3, and urine Gb3 ; DBS is useful for amenable measurement and there are interfamilial differences of α-Gal activities. Plasma α-Gal was no changes (Fig. 2-4)
2) Plasma LysoGb3 measurement is most useful both in male and female patients. Male patients with this mutation showed elevated plasma LysoGb3 after Migalastat treatment. (Fig. 5).
3) Urinary Gb3 was increased in two male patients (Fig. 6).

Long Term Clinical Efficacy of Migalastat (Fig. 7)
1) Cardiac Echo and ECG findings were maintained.
2) Kidney Function eGFR and creatinine were not changed.
3) Laboratory findings: Most of lab tests were not changed. But, plasma BNP in four female patients showed decreased tendency by migalastat treatment.
4) Subjective symptoms: no GI signs, rather less chest pain and tireless, better stair climbing etc female patients, headache and depression in male patients.

1. Amenable Mutation with pD33Y; In male patients with inadequately elevated enzyme activity in DBS and WBC, and increase plasma lysoGb3 which indicated that male patients may not be amenable whereas four female patients with same gene mutation showed 3-4 increased activities. DBS enzyme measurement of enzyme activities were useful for Amenable tests.
2. There are some intrafamilial different responses of α-Gal activities in DBS and lymphocytes.
3. Clinical findings: Cardiac echo and eGFR in these patients were maintained and BNP decreased tendency by Migalastat in female patients. Patients with amenable mutation was amenable to the efficacy of Migalastat treatment as shown in Figure 8.