Why should I get gene mutation analysis?

Getting a mutational analysis only requires 15-20 mls of blood from the patient. It is only recommended after the initial diagnosis of dysferlin deficiency is already made by either a biopsy or a monocyte dysferlin assay.

There are two main reasons for getting a gene mutation analysis (gene sequencing) of your two copies of the dysferlin gene:

1. The gold-standard for diagnosis in genetic disorders is the identification of the defect (mutation) in the gene causing the disease. There are several forms of muscular dystrophy and LGMDs with very similar clinical symptoms, and analysis at the genetic level is the only way to confirm your diagnosis of LGMD2B. A deficiency of dysferlin by a biopsy or a monocyte dysferlin assay points towards LGMD2B, but only the identification of the mutations will confirm that diagnosis.

2. If you know the nature and position of your mutations you will be able to benefit in the near future from different therapies that target specific mutations. Examples of two such therapies are given below:
    a. Stop-codon skip: This therapy is currently in clinical trials and will potentially benefit only those patients who have a specific kind of mutation, called a "nonsense" or "stop" mutation.
    b. Exon skipping: This therapy is also in clinical trials and will potentially benefit a subset of patients with mutation in specific areas of the dysferlin gene.