Comments to the Author

This manuscript details the distribution of genetic mutations of *DMD/BMD* in Chinese Han population which are well-characterized for structural and functional changes of dystrophin induced by the *DMD* gene. In this study, they preferentially included *DMD* patients with five criteria, and 1,051 unrelated Chinese families with probands affected to *DMD/BMD* were recruited. With peripheral blood samples, they used the TIANamp DNA Kit for genomic DNA isolation and the Qubit dsDNA HS Assay kit for assessing DNA. They performed MLPA analysis to detect some structural mutations, and they also performed next-generation sequencing and Sanger sequencing for small mutations. Using *DMD* gene mutations, 97.91 % of 1,051 probands were identified, and distribution of deletions and small mutations in the *DMD* gene were summarized in various ways. They also found novel small mutations such as missense, frameshift, nonsense and splice site. To assess inheritance rate of the mutations in the *DMD* gene, mothers of 471 alived probands were included in carrier screening, and 60.93% of mothers have inherited mutations to their offspring.

Major points:

1) The authors should clarify the general diagnostic criteria for diagnosis of DMD both clinically and genetically. Because the authors demonstrated how well the proposed diagnosis strategy detect mutations in the *DMD* gene for the pre-diagnosed patients, the authors should make sure that the patients have had a genetic diagnosis in advance.

2) It is unclear whether relatives of 1,051 probands were used for the analysis. In the manuscript, the authors used the term “families”, but it seems that only probands were used for the analysis except carrier screening analysis. The authors should clearly describe how the relatives of probands were used for analysis if they have been used. However, if mothers of 471 probands are only relatives used for the analysis, then I don’t recommend using the word “family”.

3) Although the sex ratio is unbalanced (Male : 1,022 vs Female : 29), it was not considered in the manuscript. Is there any possibility of selection bias? In particular, *DMD* gene is located on X chromosome. The authors should outline the sex effect in this disease and how selection bias can be corrected.

4) The authors found mutations in *DMD* gene including 53 novel mutations. However, it is unclear how they determine their findings are real mutations. The authors should provide details of detecting mutations, and measures for assessing quality of sequencing should be provided for novel mutations if possible.

5) Please provide the definition of sporadic patient.

6) Please provide the demographic characteristic of subjects in table.

Minor points

1) Full name of DMD and BMD should be provided before using their alias in the manuscript.

2) There are several spacing errors in the manuscript.

3) Numbers of subjects in introduction and the other sections are different (1053 vs 1051).

4) Softwares and tools used in this study should be cited. For example, PROVEAN, polyphen-2.