Shanghai Jieyi
Biotechnology Co., LTD.
Sample number: GPD-
B2307586

MLPA Molecular Detection Report

surname and personal name: Date of birth: 2020-02-28 Sample Number: GPD-B2307586 Sample type: peripheral blood

sex: Male Sample date: 2023-12-01 Sample collection date: 2023-12-04 Sample trait: normal

Bed number: Hospitalization number: - test hospital: - test doctor: -

Clinical complaint:

F Hx:

surveillance project: **Detection of large muscular dystrophin gene in pseudofertilizer-multiplex ligation probe amplification technique (mu ltiplex ligation-dependent probe amplification, MLPA)**

Test content: **This test is only for the copy number variants (deletions / duplications) of the DMD gene, and it cannot be tested for the point mutation, minor deletion, minor insertion and so on.**

Note: The above information is subject to the information provided by the subject person.

Conclusion: No exonic deletions or duplications of DMD gene were found.

Detection result:

Genes (transcripts)	Exons (Exon)	Copy number	Missing / duplication	Pathogenic grading	Related diseases
DMD	-	-	No deletions or duplications were detected	-	Pseudotrophic large muscular dystrophy

Result note:

No copy number variants (exon deletions or duplications) were detected on the DMD gene, as shown in the diagram of the report. This test is limited to other forms of variation (punctions, minor insertions or minor deletions) in the DMD gene.

The main causal gene for developing muscular dystrophy is the DMD gene, whose mutation causes a large pseudomuscular dystrophy, following an X-linked recessive inheritance. According to the severity of the disease can be divided into duchenne muscular dystrophy (DMD) or muscular dystrophy (BMD), DMD children with bilateral gastrocnemius muscle gradually false hypertrophy, tendon reflex weakened or disappear, proximal muscle weakness lead to walking difficulties, with the aggravation, before about 12, died of respiratory failure or heart failure. The clinical presentation of BMD patients is similar to DMD but mild DMD, and most patients are able to walk freely after age 20 years. The disease is mostly diagnosed in men, and women are mostly asymptomatic carriers. 8% -22% of female carriers can show different degrees of muscle weakness, called symptomatic carriers.

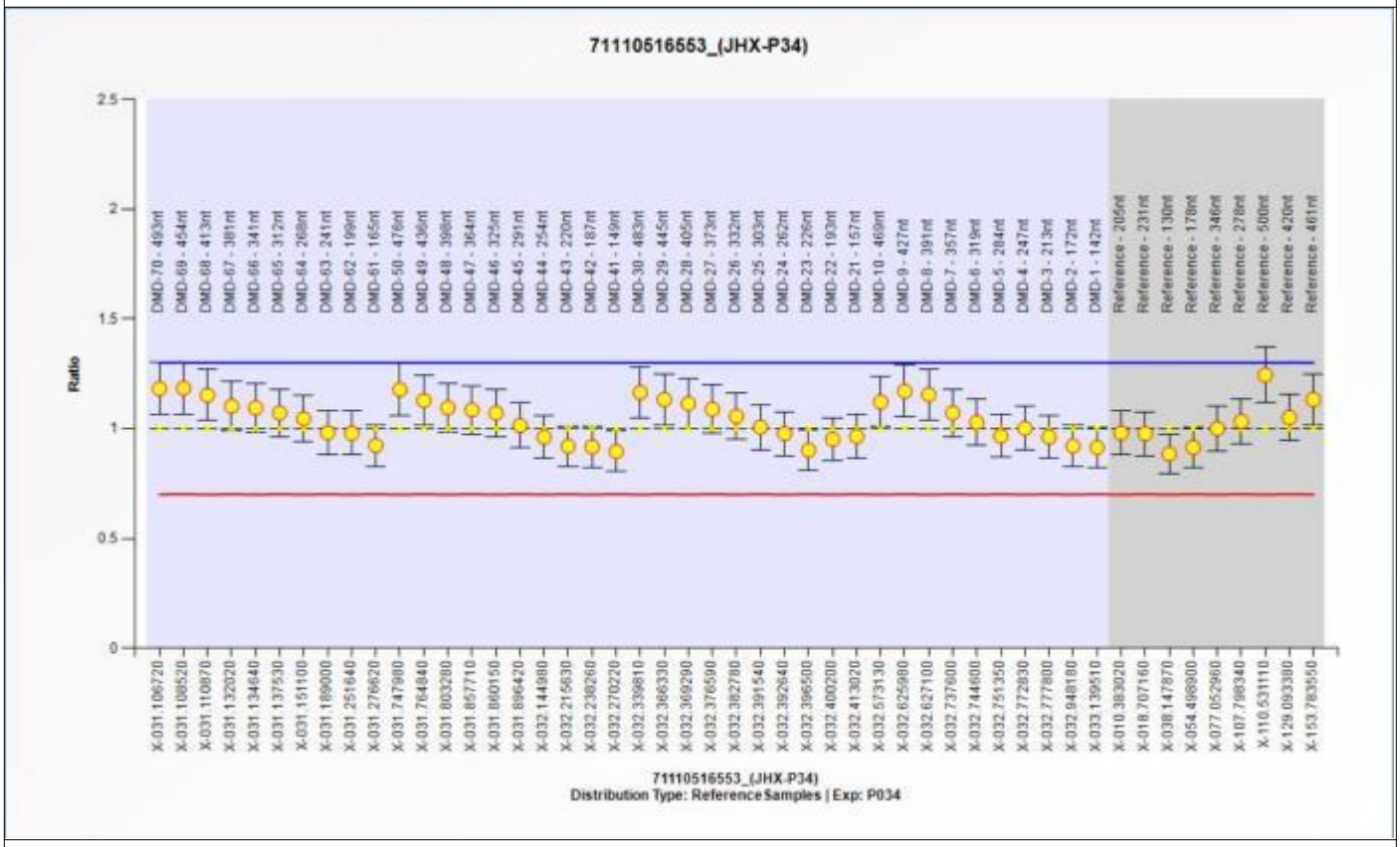
The current reported exonic deletions or repeated variants in the DMD gene account for 2 / 3 of all the mutation types in the gene, and another 1 / 3 of the gene variants are caused by point mutations, small deletions or minor insertions.

Suggestion: It is suggested to consider the second-generation sequencing if necessary.

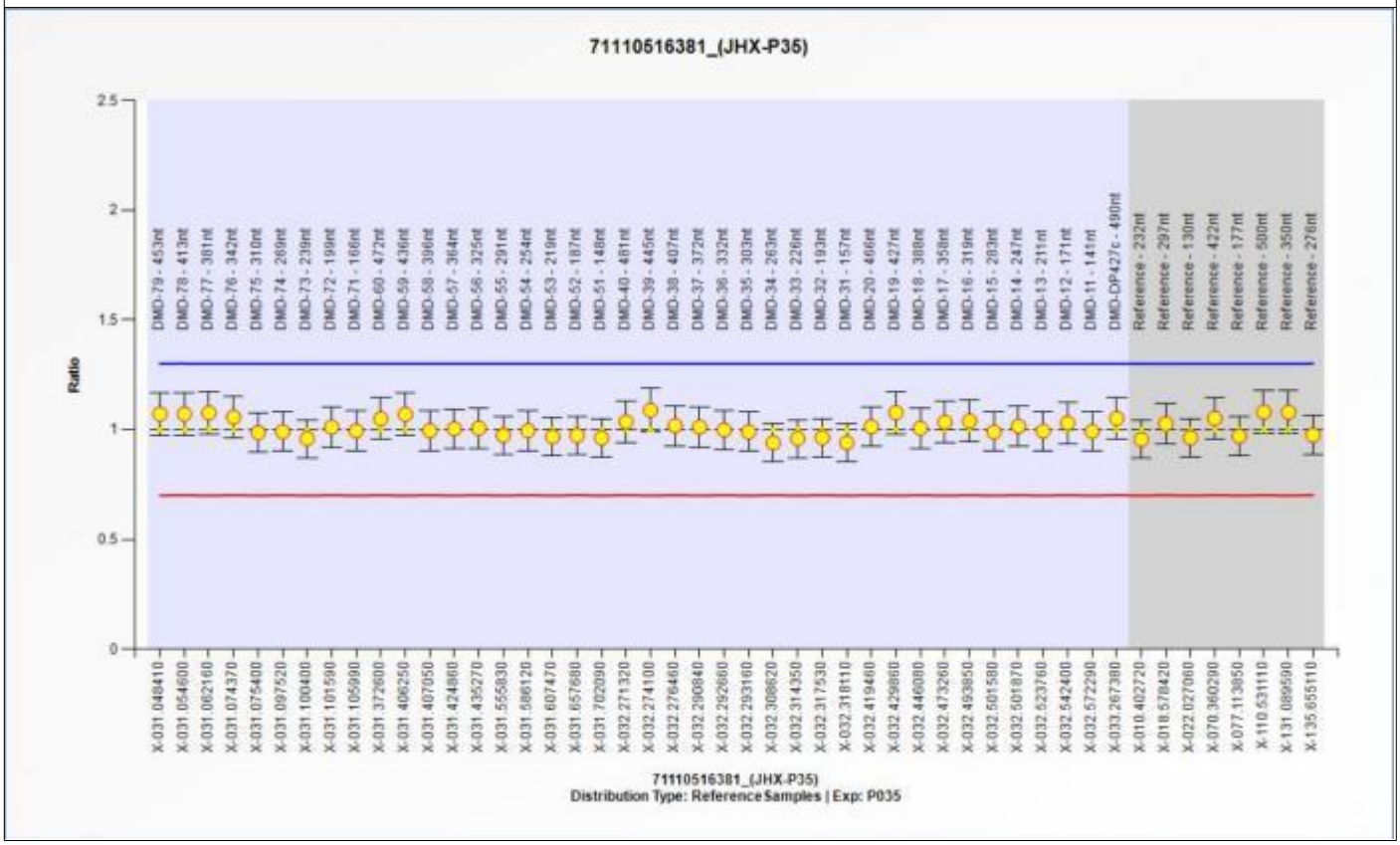
Inspection time: 2023-12-10 Report time: 2023-12-11

Appendix-Gene test results

DMD -P034-MLPA



DMD -P035-MLPA



Note: The fluorescence signal intensity is between 0.7 and 1.33, and the copy number is usually considered normal.