Supplementary

Figure S1 In-house analytical pipeline.

- FastQ for N samples
- BAM files from a batch
- Sequencing and mapping quality assessment
  - Gender adjustment for chromosome X
  - HMZDeLFinder calling CNV on chrX
- GATK best practise for SNV and INDEL
- CANOES calling CNV on chrX
- Variant classification following ACMG guideline
- Hemizygous CNVs overlapping DMD
- P/LP SNV and INDEL on DMD

Figure S2 The results of Sanger sequencing and multiplex ligation-dependent probe amplification.