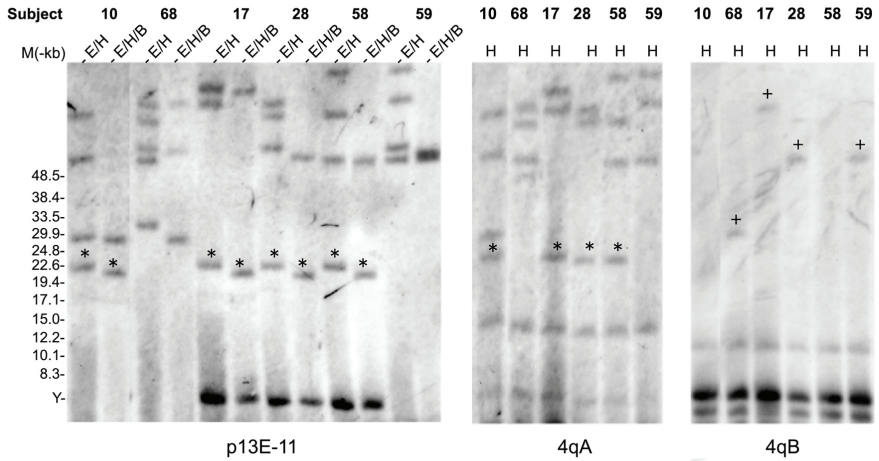
FSHD1 Diagnostic Deletion Testing Summary

FSHD1 testing: Southern blotting vs Molecular Combing vs Single-Molecule Optical Mapping

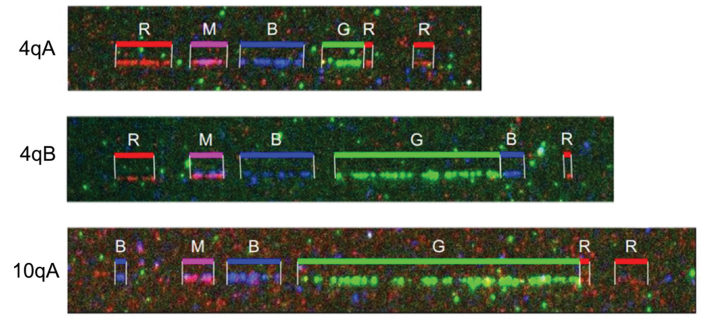
All FSHD1 deletion testing requires high molecular weight (HMW) DNA, typically from a blood draw. All techniques provide the size of the chromosome 4 D4Z4 arrays and if FSHD permissive.

A Southern blot hybridization



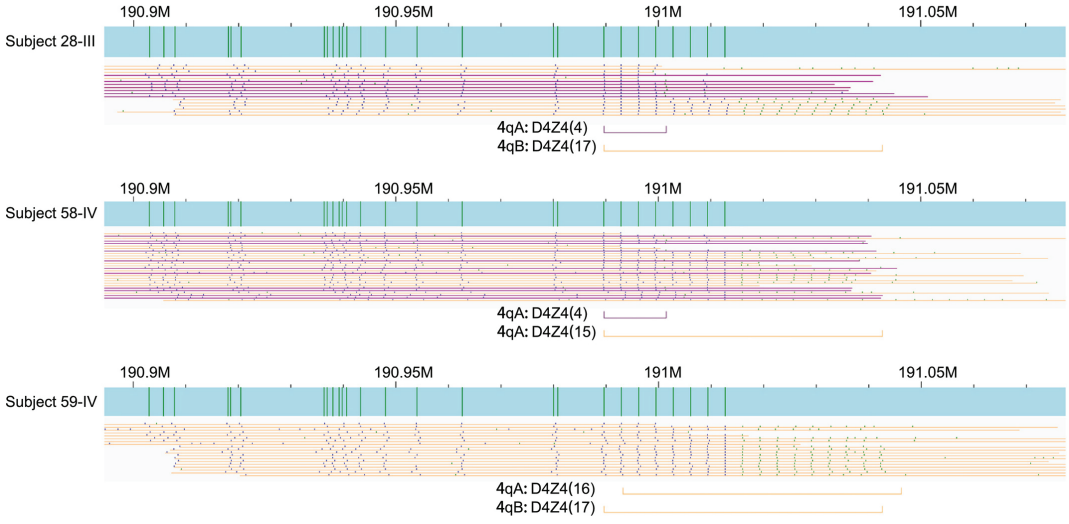
Labor intensive: Multiple Southern blots using different restriction enzyme digestions are probed with p13E-11 to determine D4Z4 array sizes and their 4A or 4B designations.

B FISH combing



Fully automated: Chromosome spreads are hybridized with fluorescent probes to simultaneously detect and determine the sizes of the chromosome 4 and 10 D4Z4 arrays and their 4A or 4B designations.

C SMOM analysis



Fully automated: Chromosome preparations are nicked with enzymes and fluorescently labeled. The sizes of the D4Z4 arrays on chromosome 4 and 10 and the 4A or 4B designations are determined by the patterns of the labels.

Figure from: Q. Zhang *et al*., (2019) *Molecular Genetics & Genomic Medicine* 7:e565.