Integrating sequencing technologies in personal genomics: optimal low cost reconstruction of structural variants

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Figure S1

MM values and worst case reconstruction examples of a 10Kb novel insertion.

(A) Mapability values for all the 30mers of a ~ 10Kb novel insertion (Variant ID in Huref: 1104685256488, with 1000 flanking sequences): $MM(flanking_{1000bp}(Ins), G_{hg18}, 30, 0)$. The insertion region is shown in blue.

(B) and (C) show the simulation results in reconstructing this region with a same total budget of \sim \$7. The solid blue lines are the assembled contigs that can be localized back to this insertion, with solid red lines for the parts that do not match due to mis-assembly. The dotted blue lines are the contigs that cannot be localized back to this insertion, with the dotted red lines representing the parts that do not match.

(B) Typical worst-case reconstruction result with $\sim 0x$ long reads, $\sim 7x$ medium reads, and $\sim 17.5x$ short reads.

(C) Typical worst-case reconstruction result with ~ 0.05x long reads, ~ 7x medium reads, and ~ 10x short reads.



A) MM(10Kb Insertion + 1000bp up/down-stream, hg18, 30mer, 0 mismatch)

Figure S1: MM values and worst case reconstruction examples of a 10Kb novel insertion.