

Figure S1: Bait re-balancing improves capture uniformity in the monogenic diabetes targeted capture assay. Relative capture efficiency of target bases was calculated for samples captured using either the Agilent SureSelect Human All Exon v1 (38 Mb) system or the custom monogenic diabetes panel, by dividing the observed depth of coverage at each base by the average coverage of all targeted bases in a given sample; thus, in an ideal system, all bases would be captured with equal efficiency and have a relative value of 1. The average relative capture efficiency was then calculated for a cohort of exome samples (red line) and the monogenic diabetes assay samples (black line), and plotted against base position; examples are shown here for the ABCC8 and HADH genes, and exon positions are indicated on the horizontal axis.