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Supplemental information

A spectrum of recessiveness among Mendelian disease variants in UK Biobank

Alison R. Barton, Margaux L.A. Hujoel, Ronen E. Mukamel, Maxwell A. Sherman, and Po-Ru Loh

Figure S1. Carriers of recessive Mendelian disease variants display quantitative phenotypes across many loci. Mendelian diseases and their associated genes are listed to the left in each column, and effect size is plotted on the right for each associated quantitative trait in (error bars, 95% CIs). Units are standard deviations with the exceptions of pigmentation traits (which used consecutive integers to code levels defined by UK Biobank) and age of menarche (years). Positive-effect variants are shown in red, negative-effect variants in blue, and variants not Bonferroni-significant for one of the displayed traits in gray. Marker shapes correspond to effects on the gene and gene product as reported in ClinVar.

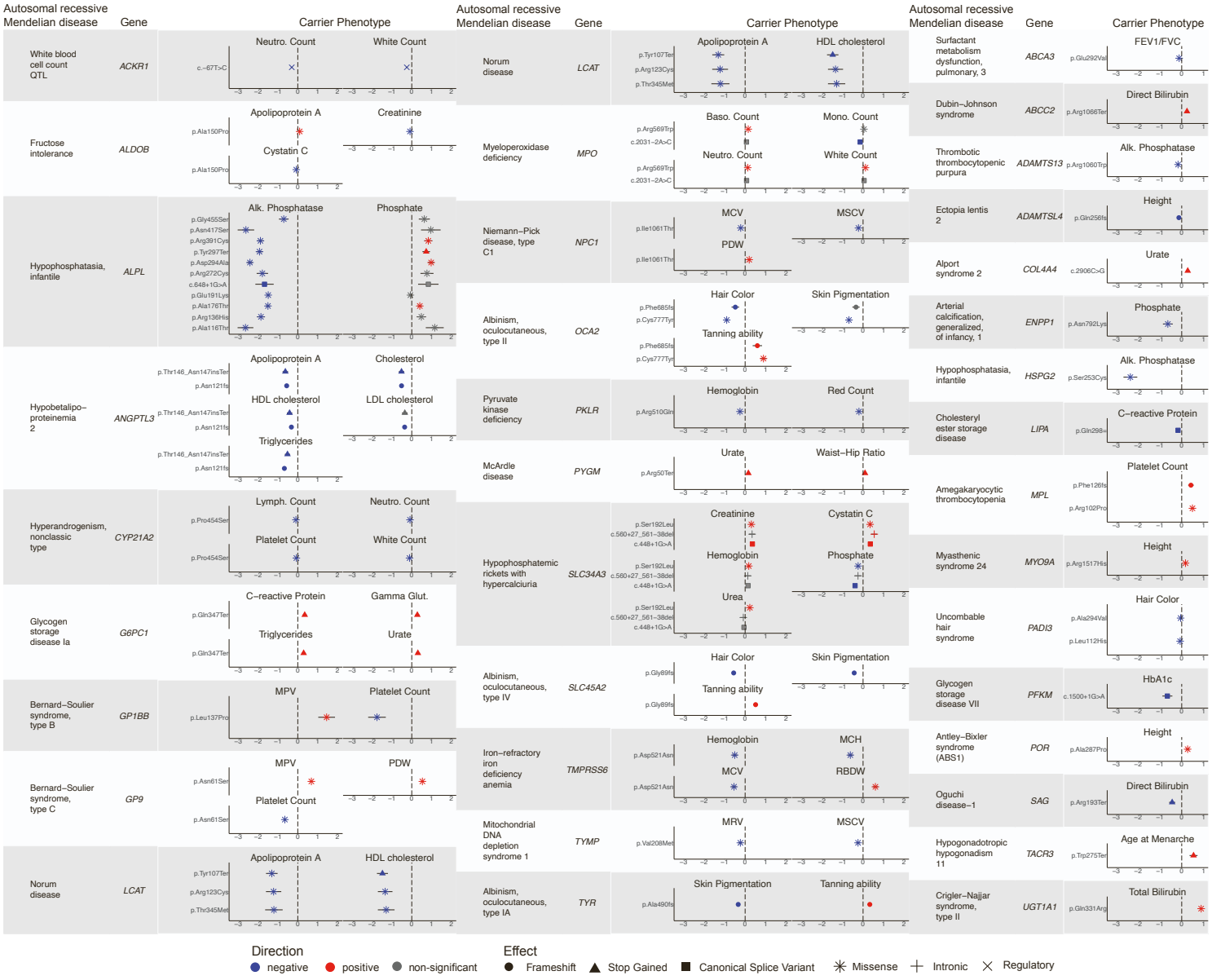

Figure S2. Comparison between odds ratios for mitigated disease phenotypes in CF carriers in UK Biobank versus Miller *et al*. (2020). Results for this analysis of UK Biobank are shown as red circles; results from Miller *et al*. are shown as black triangles (data points, odds ratio; error bars, 95% CIs).