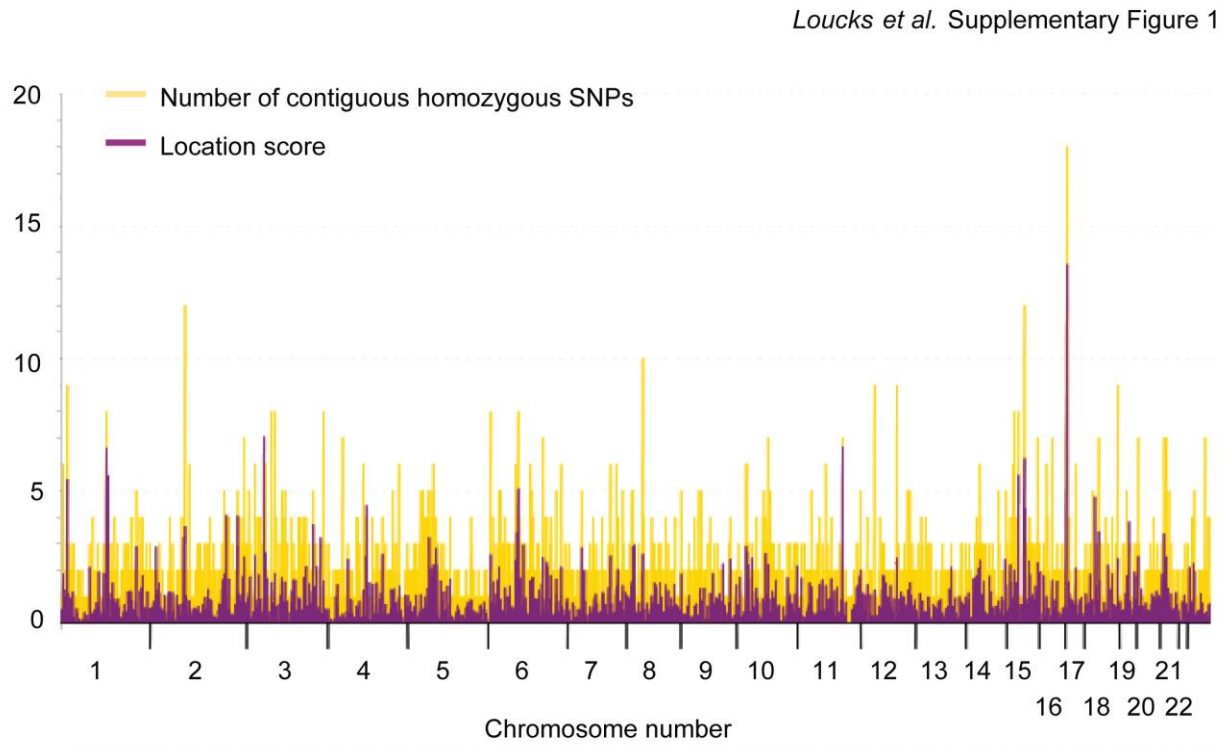
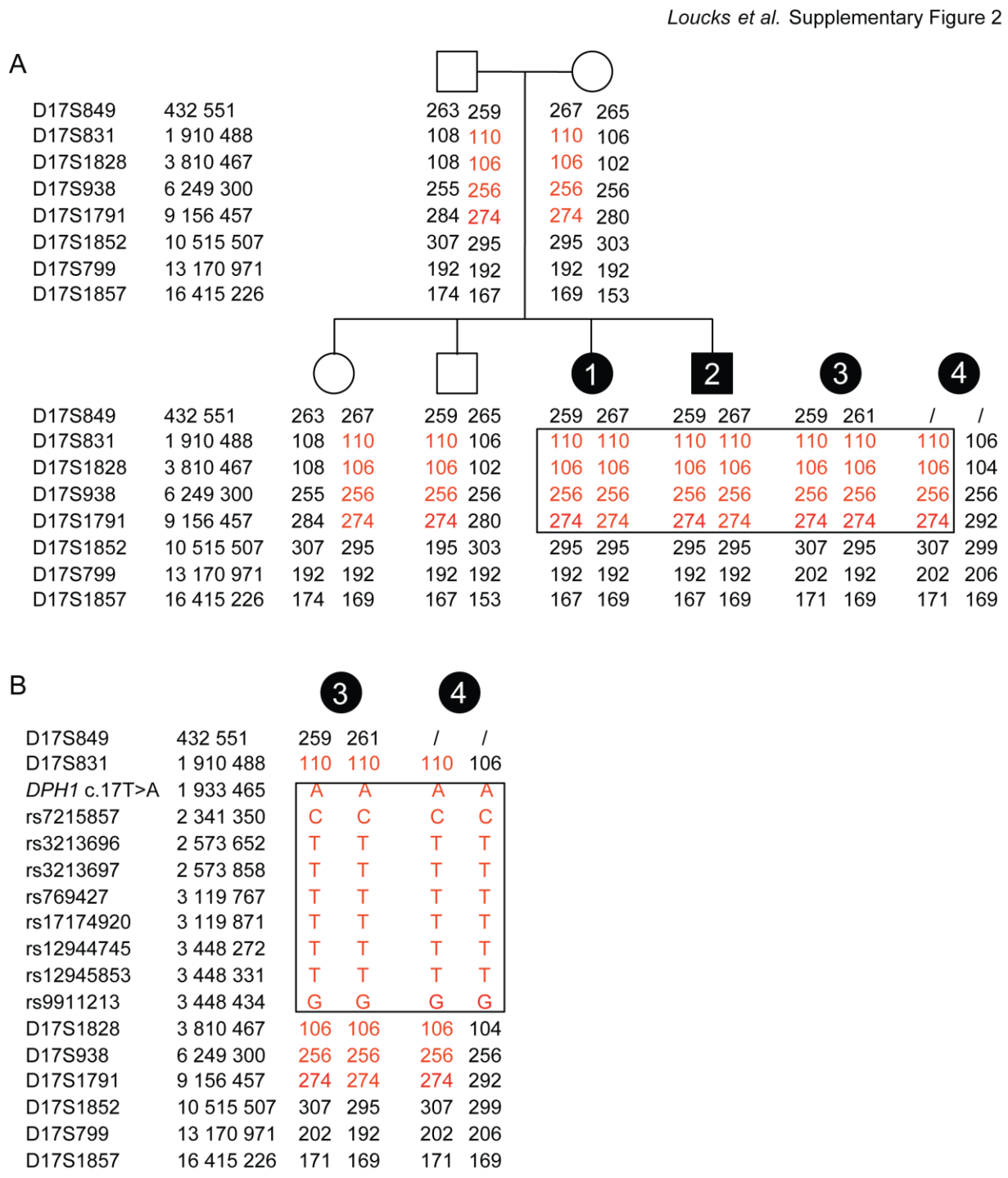


Supp. Figure S1. A genome-wide 10K SNP array of patients 1, 2 and 3 from the North American genetic isolate showed a homozygous region ~9.8 Mb on chromosome 17 shared by all three patients. The yellow indicates the number of contiguous homozygous SNPs in each chromosomal region, and the purple indicates the location score, which is a measure of the frequency of particular SNPs in the general population.



Supp. Figure S2. Microsatellite mapping confirmed the homozygous region found on chromosome 17 in patients 1-3 and targeted SNP genotyping revealed that only a small ~1.9 Mb region containing the c.17T>A *DPH1* mutation is shared by all four patients from the North American isolate **A:** Microsatellite mapping confirming the homozygous region on chromosome 17 found from the 10K SNP array. The marker names are indicated on the left, followed by their

Mb positions according to NCBI build 37.1. For each individual, both microsatellite alleles are shown as the sequence length in base pairs. Markers in red represent regions that were homozygous as determined by the 10K SNP array on patients 1 and 2 their unaffected siblings. These homozygous regions were shared by patient 3 but not by patient 4. **B:** Targeted SNP genotyping revealed a small ~1.9 Mb homozygous region shared by all four patients between the *DPH1* c.17T>A variant and the D17S1828 microsatellite marker.