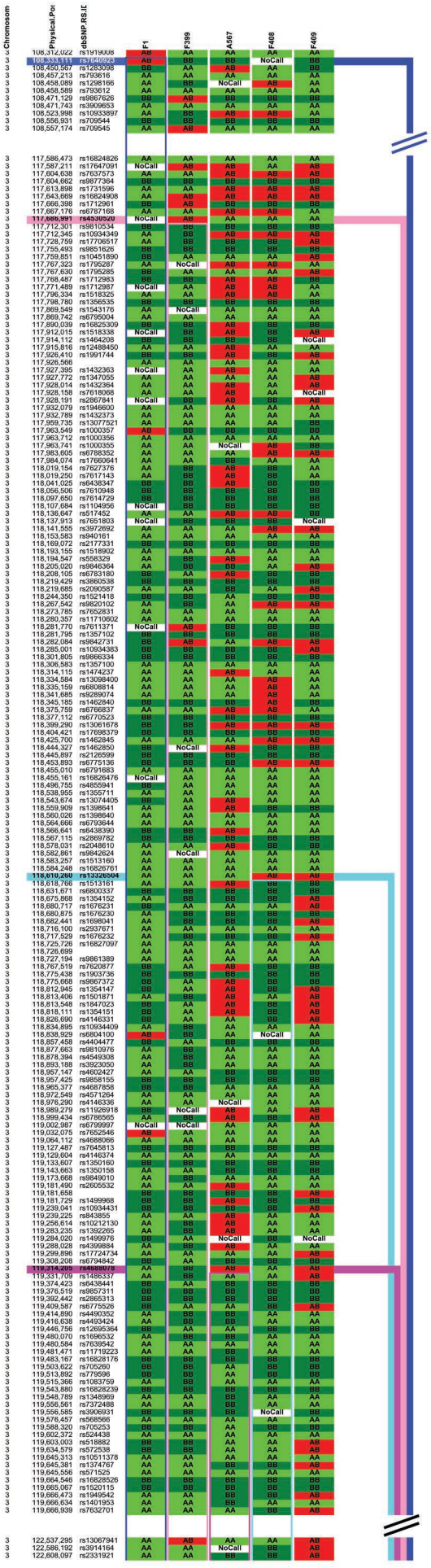
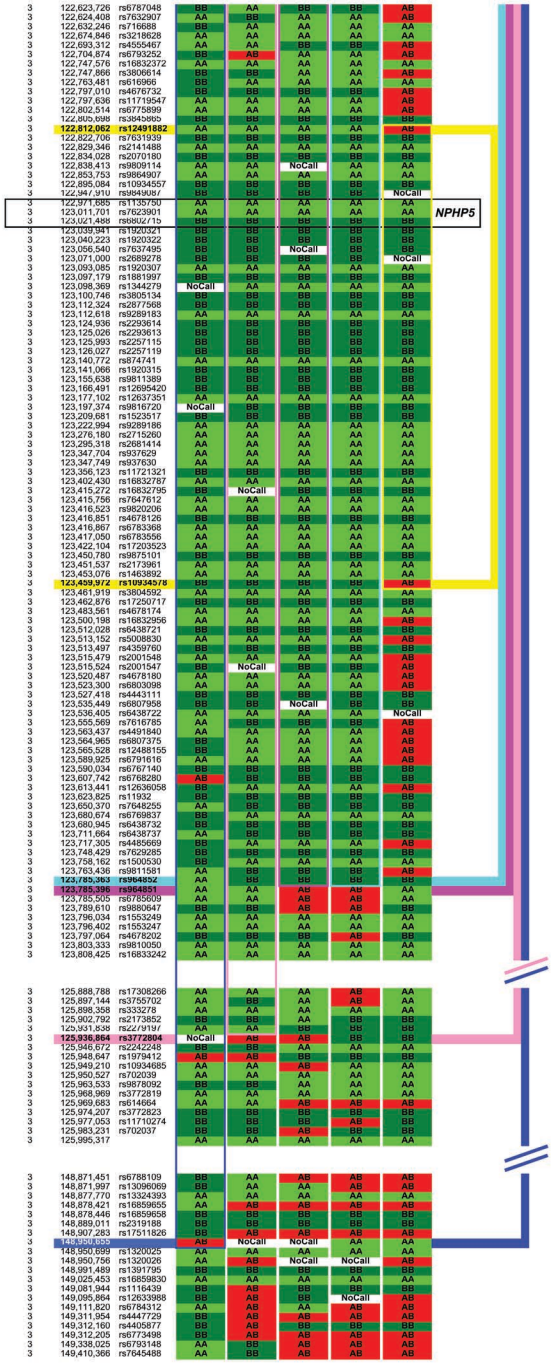

Chromosom
Physical.Pos
dbSNP.RS.ID
F1
F399
A567
F408
F409



Supplementary Figure 3. The *NPHP5* mutation F142fsX147 occurs on a shared haplotype.
Haplotypes from a 250k SNP array of 5 different families with Senior-Loken syndrome due to the homozygous *NPHP5* mutation F142fsX147 are shown. Homozygous alleles are on green background (light green for "AA", dark green for "BB"). Heterozygous alleles are on red background. "No calls" by the SNP evaluation software BRLMM are on white background. Continous segments of homozygosity (that are partially interrupted by rare false heterozygous allele calls) are encased in colored boxes and delimited by same-color brackets on the right with their heterozygous flanking markers indicated in the same color. The position of the *NPHP5* gene is indicated.
Note that all families within the *NPHP5* region share identical homozygous alleles within haplotypes of varying length. In this way we demonstrate that the *NPHP5* mutation F142fsX147 occurs on a shared haplotype and thereby represents a European "founder" mutation by descent from a common ancestor. The homozygous intervals in the three families from Germany measure 40.6 Mb in F1, 8.2 Mb in F399, 4.5 Mb in A567, and in the two families from Switzerland 5.18 Mb in F408, and 0.65 Mb in F409. The short interval of 0.65 Mb was not detectable as a homozygosity peak (see **Table 1** and **Figure 3**).