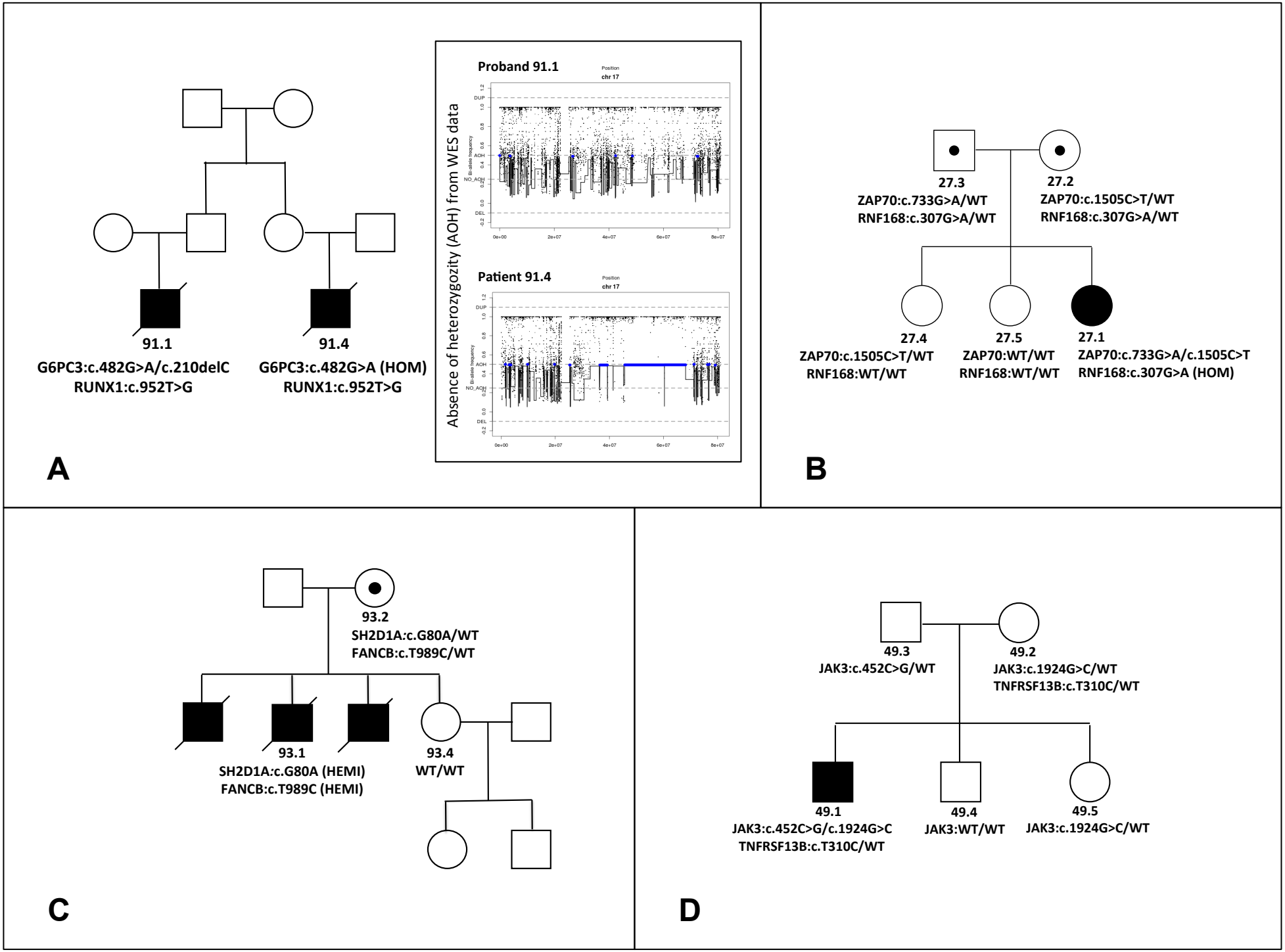
91.1
G6PC3:c.482G>A/c.210delC
RUNX1:c.952T>G
91.4
G6PC3:c.482G>A (HOM)
RUNX1:c.952T>G
Absence of heterozygozity (AOH) from WES data
Proband 91.1
Patient 91.4
A
27.3
ZAP70:c.733G>A/WT
RNF168:c.307G>A/WT
27.2
ZAP70:c.1505C>T/WT
RNF168:c.307G>A/WT
27.4
ZAP70:c.1505C>T/WT
RNF168:WT/WT
27.5
ZAP70:WT/WT
RNF168:WT/WT
27.1
ZAP70:c.733G>A/c.1505C>T
RNF168:c.307G>A (HOM)
B
93.2
SH2D1A:c.G80A/WT
FANCB:c.T989C/WT
93.1
SH2D1A:c.G80A (HEMI)
FANCB:c.T989C (HEMI)
93.4
WT/WT
C
49.3
JAK3:c.452C>G/WT
49.2
JAK3:c.1924G>C/WT
TNFRSF13B:c.T310C/WT
49.1
JAK3:c.452C>G/c.1924G>C
TNFRSF13B:c.T310C/WT
49.4
JAK3:WT/WT
49.5
JAK3:c.1924G>C/WT
D

Figure E5.