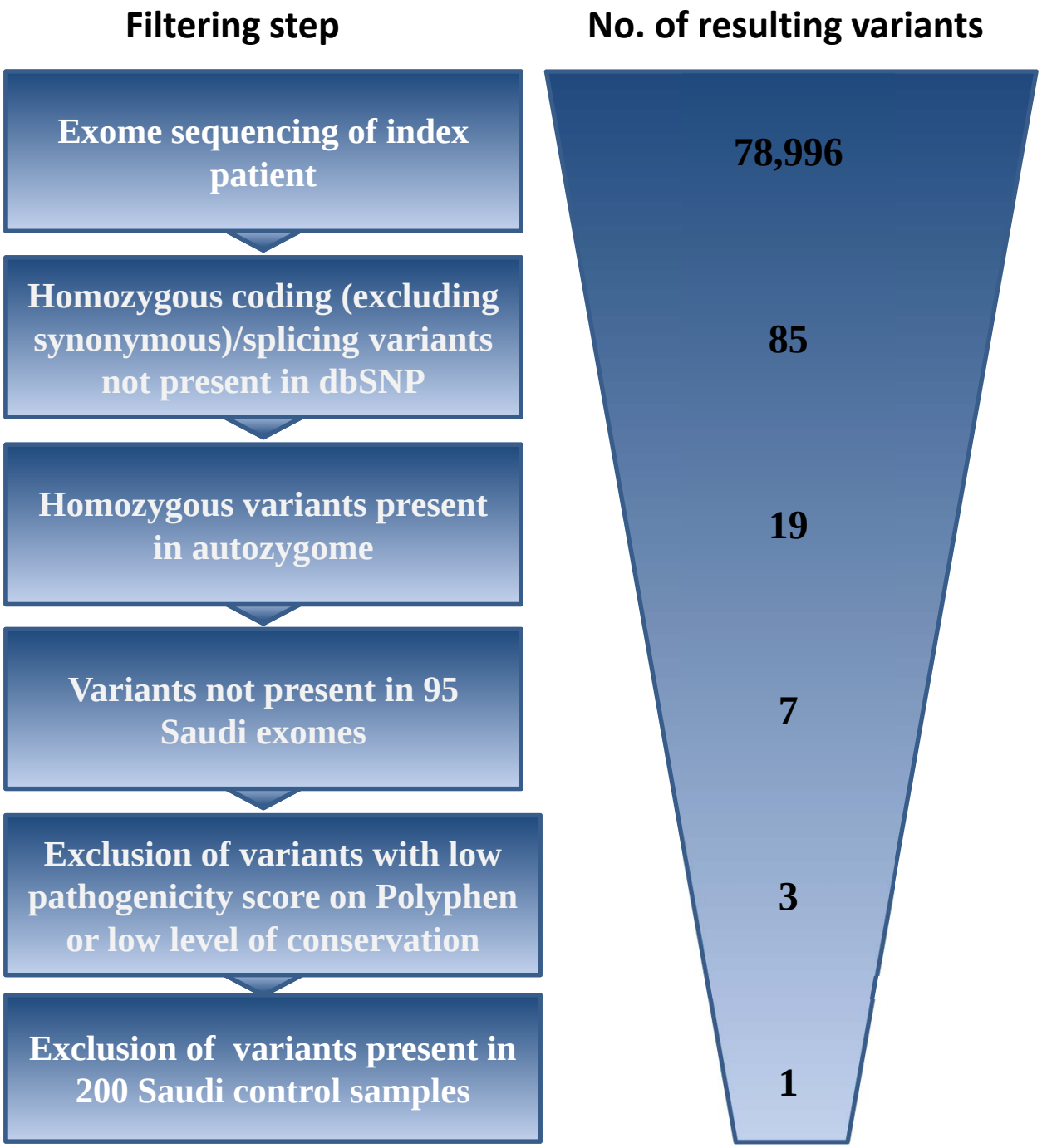
Figure S2


Filtering step
No. of resulting variants
Exome sequencing of index patient
78,996
Homozygous coding (excluding synonymous)/splicing variants not present in dbSNP
85
Homozygous variants present in autozygome
19
Variants not present in 95 Saudi exomes
7
Exclusion of variants with low pathogenicity score on Polyphen or low level of conservation
3
Exclusion of variants present in 200 Saudi control samples
1