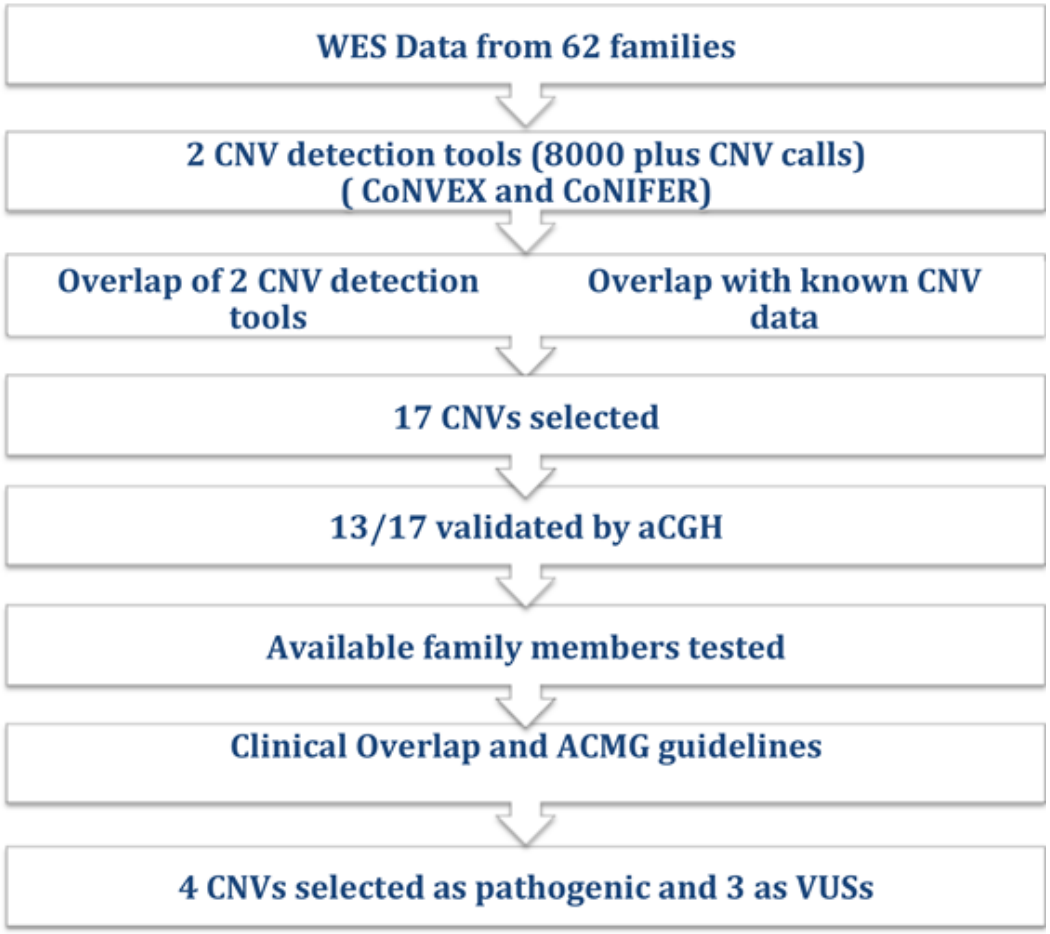
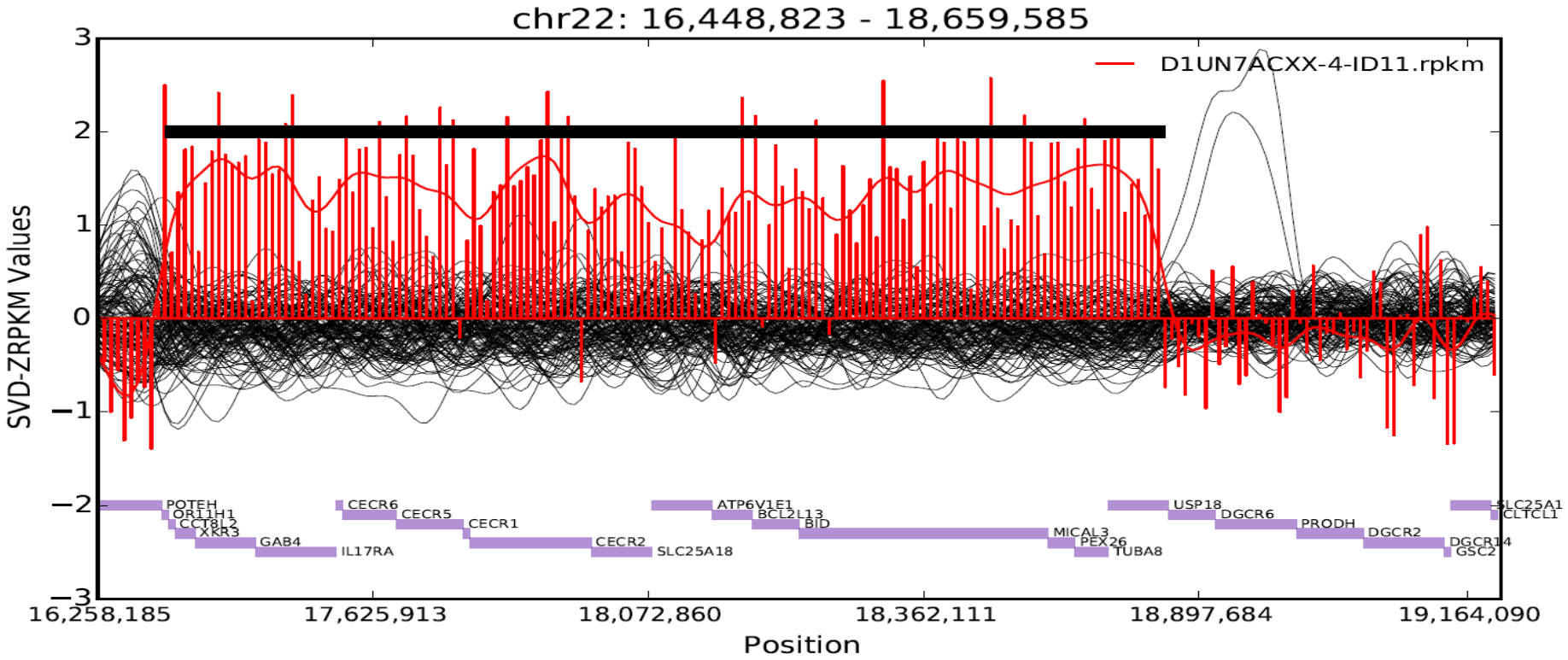
Figure S1. Copy-number variants (CNVs) inference from WES data, (a) flowchart showing the steps of CNV discovery, (b) a representative pathogenic CNV is shown here. This is a triplication at 22q11 as shown in (b), aCGH data of the proband and parents is depicted in (c) Rests of the pathogenic CNVs are not shown here.

(a)



(b)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SVD-ZRPKM Values
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2
1
0
−1
−2
−3
POTEH
OR11H1
CCT8L2
XKR3
GAB4
IL17RA
CECR6
CECR5
CECR1
CECR2
SLC25A18
ATP6V1E1
BCL2L13
BID
MICAL3
PEX26
TUBA8
USP18
DGCR6
PRODH
DGCR2
DGCR14
GSC2
SLC25A1
CLTCL1
16,258,185
17,625,913
18,072,860
18,362,111
18,897,684
19,164,090
Position

(c)