A Details of the network architecture

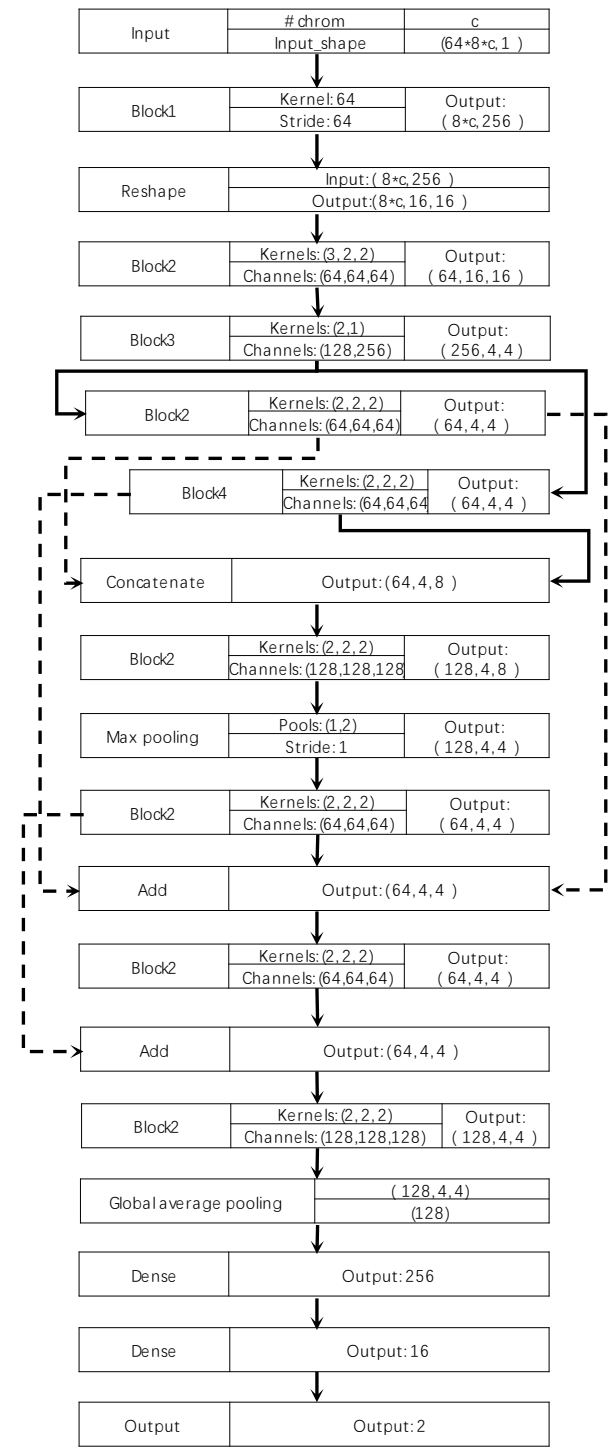

Fig. 6. Architecture of ALS-Net. The details of Block 1, Block 2, Block 3 and Block 4 are provided in Figures 7, 8, 9 and 10, respectively.

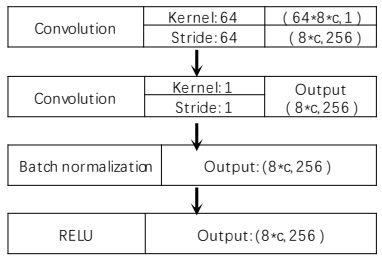

Fig. 7. Network architecture of Block 1. c is the number of chromosomes included in the model.

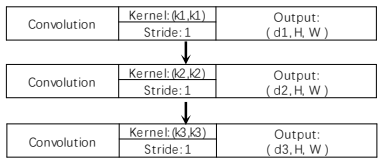

Fig. 8. Network architecture of Block 2. Inputs are given by "Kernels"=$(k1, k2, k3)$ and "Channels"=$(d1, d2, d3)$ in Figure 6, and the shape of the input tensor is (d0,H,W).

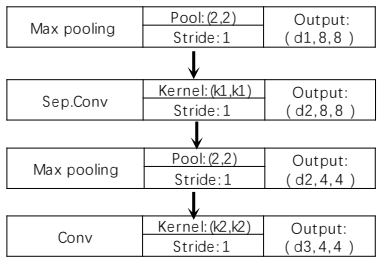

Fig. 9. Network architecture of Block 3. Inputs are given by "Channels"=$(d1, d2, d3)$ in Figure 6. Sep.Conv. = separable convolution layer (Gao et al., 2018).

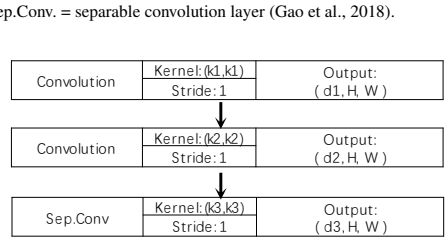

Fig. 10. Network architecture of Block 4. Inputs are given by "Kernels"=$(k1, k2, k3)$ and "Channels"=$(d1, d2, d3)$ in Figure 6, and the shape of the input tensor is (d0,H,W). Sep.Conv. = separable convolution layer (Gao et al., 2018).

B Performance of alternative network architectures

Table 5. The effect of small changes in network architecture on the classification performance for chromosome 7. Acc.=accuracy, Prec.=precision, Sep. Conv.=separable convolution, Conv.=convolution. * the layers have 1024 nodes, 1024×16 nodes and 240 nodes, respectively, with a SELU, a RELU and a SELU activation function, respectively.

Model change	Acc.	Prec.	Recall	F1 score
Block 1: change output size from $(8c, 256)$ to $(8c, 128)$	0.656	0.720	0.510	0.597
Block 4: change Sep. Conv. into Conv.	0.638	0.683	0.515	0.588
First dense layer: 128 nodes	0.662	0.704	0.559	0.623
Set learning rate = 0.005	0.666	0.699	0.583	0.636
Select 4 best promoter regions	0.672	0.670	0.679	0.674
Select 12 best promoter regions	0.681	0.677	0.691	0.684
Three-layer MLP*	0.628	0.711	0.430	0.536

C Classification accuracies of known ALS genes

Table 6. Accuracies for ALS-associated genes of chromosomes 7, 9 and 17 as listed on http://alsod.iop.kcl.ac.uk/ (Abel et al., 2013). The ALS-associated genes from Abel et al. (2013) for chromosome 22 were not kept in our dataset after QC, and hence no results are reported.

Chr	Gene	Accuracy
Chr7	GARS	0.5159
	RAMP3	0.503
	ZNF746	0.524
	DPP6	0.532
Chr9	SUSD1	0.504
	ALAD	0.513
	STEX	0.545
Chr17	MAPT	0.514
	SLC39A11	0.511

D Gene ontology of selected chromosome 7 promoter regions

Table 7. Gene ontology terms of the promoter regions that were selected by Promoter-CNN for chromosome 7.

Gene	Gene ontology class(es)
LAMB4	Basement membrane, cell adhesion
LOC105375113	None - RNA Gene, affiliated with the ncRNA class
LOC105375607	None - RNA Gene, affiliated with the ncRNA class
TRY2P	None - trypsinogen-like pseudogene
TYW1B	tRNA processing, FMN binding, metal ion binding, 4 ion 4 sulfur cluster binding, oxidation-reduction process, tRNA-4-demethylwyosine synthase activity
LOC101929756	None - RNA Gene, affiliated with the ncRNA class
DTX2	None for homo sapiens. For other organisms: metal ion binding, zinc ion binding, protein ubiquitination, notch signaling pathway, cellular component, nucleoplasm