Frequency

Benign | Pathogenic

0.1 0.2 0.3 0.4 0.5 0.6 0.7 0.8 0.9 1.0

Pathogenicity score for
Chr12: 103246653 C>T
(PAH gene R>Q)

Global Maxpooling
(1)

(51,1)

BatchNormalization +
Sigmoid+
1D Convolution
(1,1,1)

(51,1)

BatchNormalization +
Relu+
1D Convolution
(1,40,40)

(51,40)

+

6 Layers

+   (51,40)

BatchNormalization +
Relu +
1D Convolution
(5,40,40)

(51,40)

BatchNormalization +
Relu +
1D Convolution
(5,40,40)

(51,40)

○

(51,40)

BatchNormalization +
Relu +
1D Convolution
(5,80,40)

(51,80)

BatchNormalization +
Relu +
1D Convolution
(5,40,40)

(51,40)   Concat   (51,40)

BatchNormalization +
Relu +
1D Convolution
(5,40,40)

(51,40)   (51,40)

(51,40)   Pretrained Solvent
Accessibility Layers   (51,40)

(51,20)

99 Vertebrate
Conservation

(51,40)   Pretrained Secondary
Structure Layers   (51,40)

(51,20)

99 Vertebrate
Conservation

(51,40)   1D Convolution
(1,20,40)   (51,40)

(51,20)

Only Primate
Conservation

(51,40)   1D Convolution
(1,20,40)   (51,40)

(51,20)

Only Mammal
Conservation

1D Convolution
(1,20,40)   (51,40)   +

(51,20)

Reference Sequence

+   (51,40)   1D Convolution
(1,20,40)

(51,20)

Alternative Sequence

(51,40)   1D Convolution
(1,20,40)   (51,40)

(51,20)

Only Vertebrate
Conservation