Lecture Title and Date:

Deep Learning Fundamentals II [second part], 26th Feb 2025

Objectives of the Lecture:

- Deep Learning in Image Feature Detection
- Using CNN for DNA sequences
- Extending CNN for Long-Range dependencies
- Deep learning & Motif enrichment

Key Concepts and Definitions

- Kernel Filter (Convolutional Feature) : A small matrix that slides over pixels to detect patterns such as edges in the images.
- Parameter Sharing: Applying the same weight to different spatial position, thus making CNN scalable and efficient
- Feature Map: Output of each subsequent layer in CNN
- Stride: The step size by which filter moves across the input consequently affecting the output size
- Pooling: Reducing the dimensionality of feature maps.
- Hierarchical Feature Learning: CNN learns features progressively from low level to high level.
- STARR seq: Parallel reporter assay to identify transcriptional enhancers based on their activity directly.
- Cis regulatory elements: They are DNA sequences that regulate the transcription of genes.

Main Content/Topics

1. **Convolutional Neural Network (CNN)**:

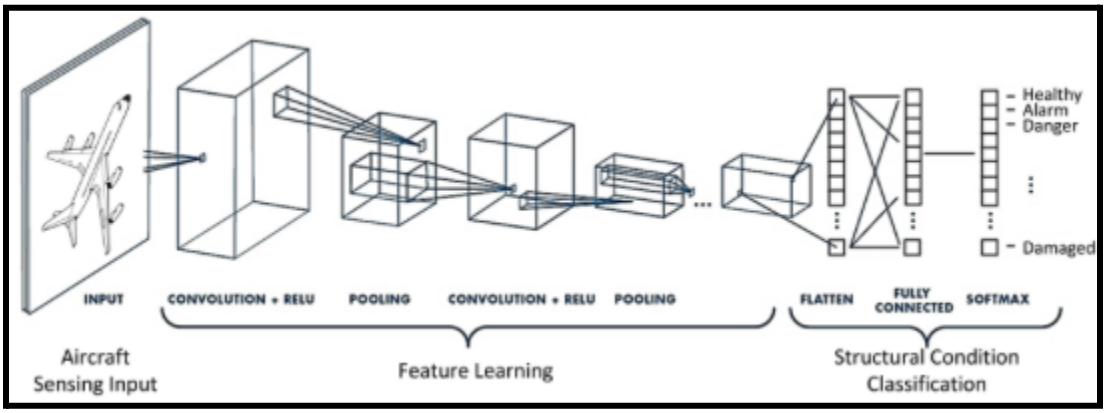
   Introduction:

   i. CNN is a class of deep learning models which are designed for analyzing structured data such as images.
   ii. They are good in feature extraction by learning hierarchical patterns through convolutional operations.

   Working:

   iii. CNN uses number concepts and mathematical functions such as:

1. Convolution
2. RELU
3. Pooling
4. Flattening
5. Softmax



iv. Detecting Features via Kernel Filters
    1. CNN uses kernels to scan the images in small patches
    2. Each filter detect specific patterns like: edges or textures
    3. Same filter is applied across all locations

v. After applying multiple filters, CNNs generate multiple feature map and each captures different features.

vi. As we stack more convolutional layers, it will learn more features, initially there are low level features are extracted but as we go deep in the layers, more high level features are extracted
    1. Low level features: Edges, corners
    2. Mid level features: Shapes, textures
    3. High level features: Objects, faces

vii. Stride: The number of pixels a filter moves across the image.
    1. It affects the resolution of the feature map and the models' computational efficiency.
    2. Larger stride value results in smaller output dimensions..

viii. Pooling: It helps to reduce the spatial size of the feature maps, making efficient computation.
    1. Max pooling: Takes the maximum value in a region
    2. Average pooling: Computes the average value in the window

2. Application of CNN in regulatory genomics

a. CNN are also used in genomics to learn features from DNA sequences instead of images
    i. Input: One-hot encoded DNA sequences
    ii. Convolutional Filters detect transcriptional factor binding sites (TF motifs)
    iii. Output: Prediction of enhancer activity or regulatory function
b. Example (Deep Learning for Enhancer Activity Prediction - STARR seq):
    i. CNN model takes a 4kb genomic window as input.
    ii. Convolutional layers extract sequence motifs related to enhancer activity.
    iii. Final layer predicts STARR-seq peaks (active enhancer regions)
c. BASSET (a deep learning model for Binding and Sequence-based Sequence Enhancer Transcription prediction) is a model that applies Convolutional Neural Networks (CNNs) to genomic sequence data to predict the activity of enhancers, which are regulatory DNA regions that increase gene expression. The model takes a one-hot encoded DNA sequence as input, applies multiple convolutional layers to detect motifs and local interactions, and outputs predictions about enhancer activity, such as the likelihood of enhancer regions identified through experiments like STARR-seq. BASSET's strength lies in its ability to learn sequence motifs and their interactions, helping to identify transcription factor binding sites and other regulatory sequences that control gene expression.

Discussion/Comments

Overall, CNN is the powerful feature extraction in both image processing and genomics as they learn throughout the hierarchical representation from raw data. Dimensional reduction features such pooling can improve efficiency but at the cost of losing some information which is a limitation in tasks requiring long-range dependencies, such as enhancer-promoter interactions in gene regulation.

Additional suggested Reference

1. https://towardsdatascience.com/convolutional-neural-networks-explained-9cc5188c4939/
2. https://towardsdatascience.com/the-math-behind-convolutional-neural-networks-6aed775df076/

Both of the articles provide intuitive explanations of CNN, their architecture, feature extraction and application in DL. Also the second article mostly covers the mathematical foundation of CNNs.