Deep Learning in Pan-Cancer Early Detection

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**Introduction**

Morphological Abnormalities

appear later; rely on human skills; hard to detect atypical tumors

Alterations in Gene Expression

appear early; can be automated; comprehensive; next generation sequencing (NGS) techniques have made measurements fast and cheap

Cancer Diagnosis

Early detection of cancer significantly increases the chance of successful treatment. Current state-of-art cancer diagnosis relies on physicians’ experiences to identify morphological abnormalities. Molecular signatures offer an alternative option for early, objective and systematic cancer diagnosis. Deep learning methods are ideal for developing such models as it captures the complicated interactions among different genes.

**Dataset**

The Cancer Genome Atlas (TCGA) Pan-Cancer Transcriptome Profiling

Illumina HiSeq 2000 RNA sequencing platform, log2 transformed RSEM normalized counts. 58,562 transcripts X 10,663 samples (9,887 cancer and 856 normal), 37 types of cancers

**Neural Network**

Architecture

1000 nodes

500 nodes

300 nodes

250 nodes

1 node

Cost Function

\[ J = \frac{1}{m} \sum_{i=1}^{m} y^{(i)} \log(a^{(i)}) \text{pos_weight} + (1 - y^{(i)}) \log(1 - a^{(i)}) \text{pos_weight} \]

**Results**

<table>
<thead>
<tr>
<th>Model</th>
<th>Train</th>
<th>Test</th>
</tr>
</thead>
<tbody>
<tr>
<td>Logistic Regression</td>
<td>7-fold cross validation</td>
<td>Precision</td>
</tr>
<tr>
<td>2 Layer NN</td>
<td>0.83</td>
<td>0.95</td>
</tr>
<tr>
<td>4 Layer NN</td>
<td>0.95</td>
<td>0.93</td>
</tr>
<tr>
<td>8 Layer NN</td>
<td>0.75</td>
<td>0.81</td>
</tr>
</tbody>
</table>

*Hyperparameters: learning_rate = 1e-5, num_epochs = 500, pos_weight = 2

**Feature Reduction**

<table>
<thead>
<tr>
<th>Without Feature Reduction</th>
<th>With Feature Reduction</th>
<th>Accumulated Layer Unit</th>
</tr>
</thead>
<tbody>
<tr>
<td>Prior knowledge (gene sets)</td>
<td>Cell Cycle</td>
<td>Cell Death</td>
</tr>
<tr>
<td>F1 Score</td>
<td>0.90</td>
<td>0.87</td>
</tr>
</tbody>
</table>

**References**