

Introduction to Machine Learning

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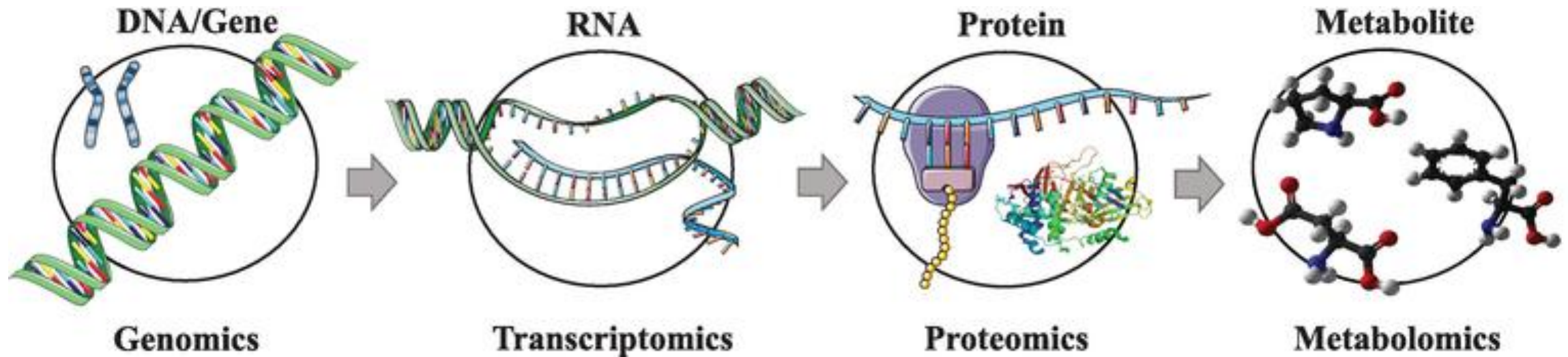
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Content

- What are data?
- What do we attend this course for?
- What are we going to learn?
- How are we going to learn?

Central Dogma

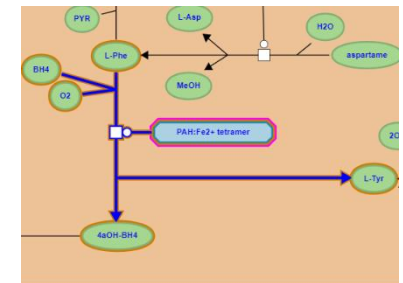
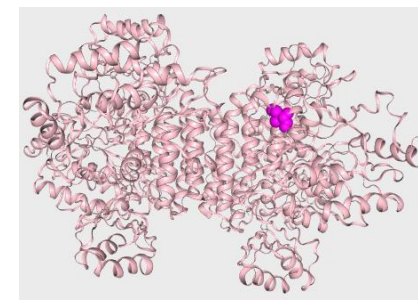
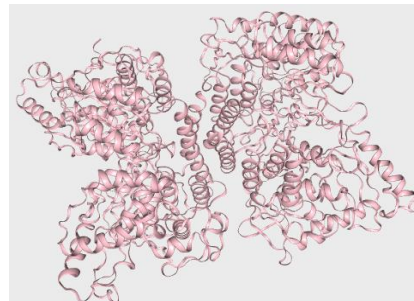


PAH gene
 Ref ...A**T**CGAT...
 P1 ...A**A**CGAT...
 NM_000277.3(PAH):c.971T>A

PAH mRNA
 Ref ...A**U**CGAU...
 P1 ...A**A**CGAU...
 NM_000277.3(PAH):c.971T>A

PAH protein
 Ref ...**Ile**-Asp...
 P1 ...**Asn**-Asp...
 NM_000277.3(PAH):p.Ile324Asn

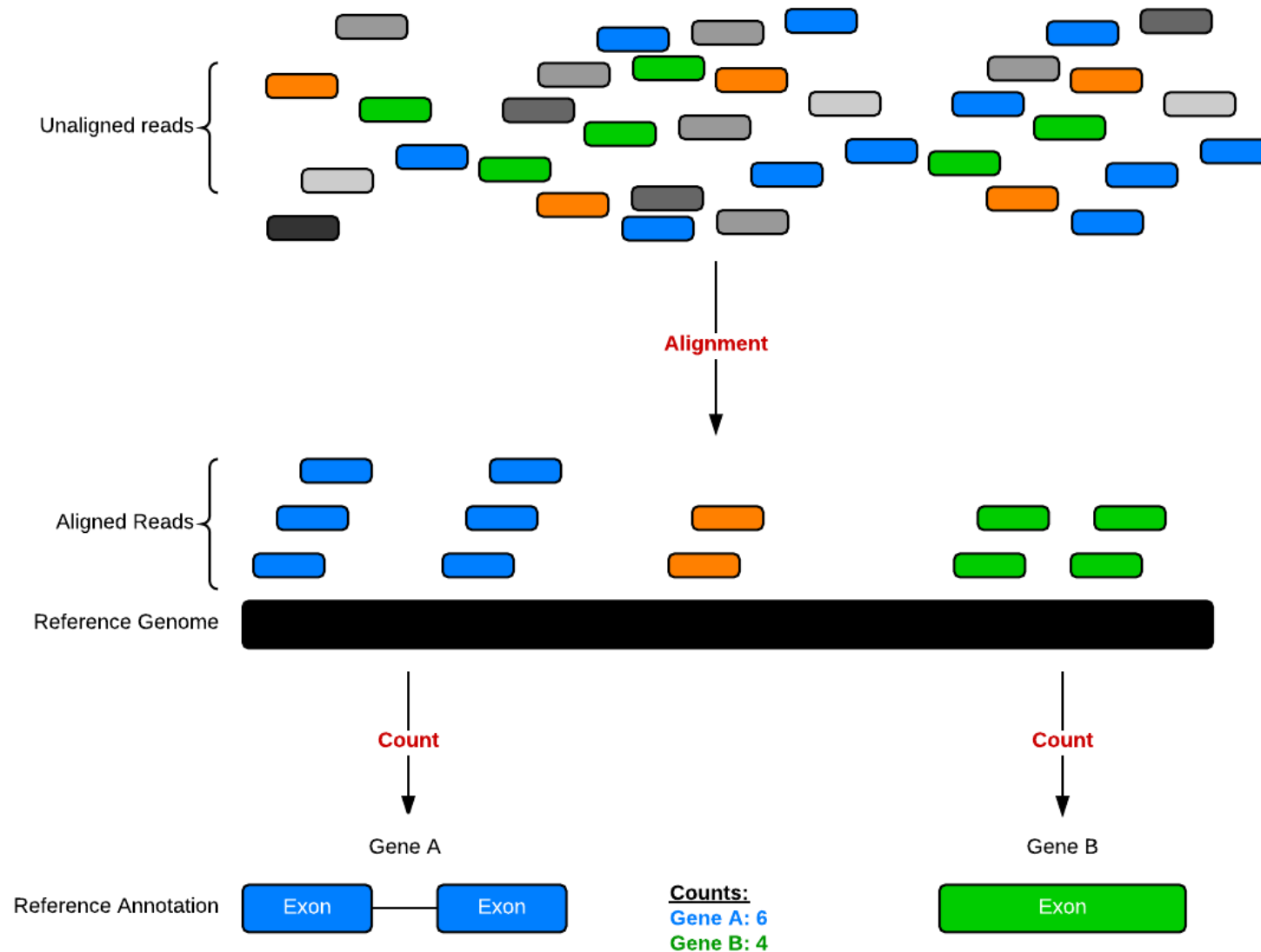
PAH
 Ref Phe → Tyr
PAH
 P1 Phe ~~→~~ Tyr



Genomic Data

- **Genomics** (SNP microarray, CNV microarray and long or short read DNA-seq/WGS)
- **Transcriptomics** (microarray, bulk RNA-seq, single-cell RNA-seq and spatial transcriptomics)
- **Proteomics** (protein microarrays and mass spectrometry)
- **Metabolomics** (mass spectrometry and Nuclear Magnetic Resonance Spectroscopy)
- **Epigenomics**
- **Methylomics** (methylation microarray, WGBS, EM-seq and long-read sequencing)
- **Metagenomics** (long or short read DNA-seq)

How to generate genomic data: RNA-seq



RNA-seq count table

countData

gene	ctrl_1	ctrl_2	exp_1	exp_1
geneA	10	11	56	45
geneB	0	0	128	54
geneC	42	41	59	41
geneD	103	122	1	23
geneE	10	23	14	56
geneF	0	1	2	0
...
...
...

colData

id	treatment	sex
ctrl_1	control	male
ctrl_2	control	female
exp_1	treatment	male
exp_2	treatment	female

Sample names:

ctrl_1, ctrl_2, exp_1, exp_2

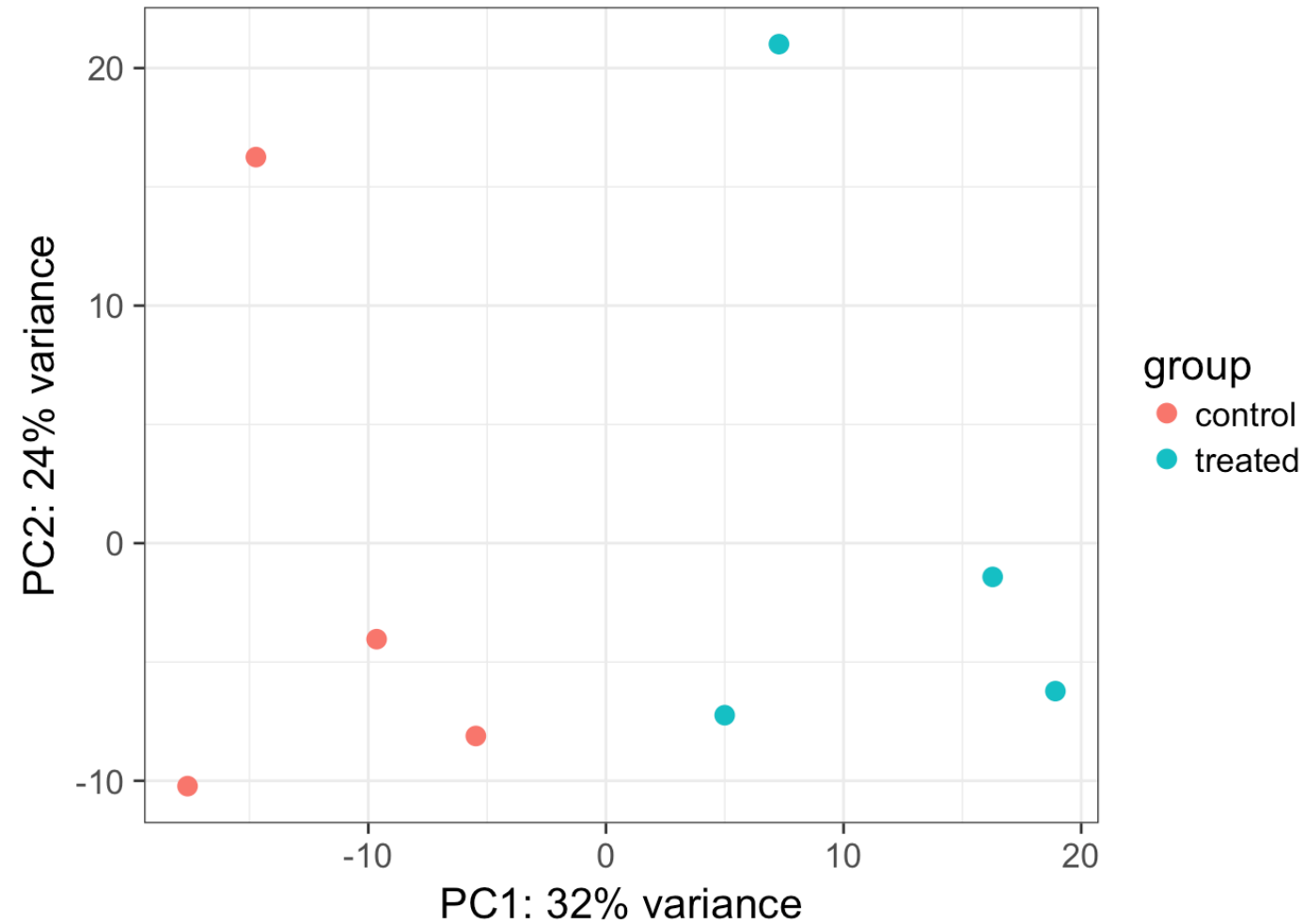
```
## # A tibble: 38,694 x 9
##       ensgene SRR1039508 SRR1039509 SRR1039512 SRR1039513 SRR1039516
##       <chr>         <dbl>         <dbl>         <dbl>         <dbl>
## 1 ENSG00000000003      723          486          904          445      1170
## 2 ENSG00000000005         0             0             0             0
## 3 ENSG000000000419    467          523          616          371      582
## 4 ENSG000000000457    347          258          364          237      318
## 5 ENSG000000000460     96           81           73           66      118
## 6 ENSG000000000938         0             0             1             0
## 7 ENSG000000000971   3413         3916         6000         4308     6424
## 8 ENSG000000001036   2328         1714         2640         1381     2165
## 9 ENSG000000001084     670          372          692          448      917
## 10 ENSG000000001167    426          295          531          178      740
## # ... with 38,684 more rows, and 3 more variables: SRR1039517 <dbl>,
## #   SRR1039520 <dbl>, SRR1039521 <dbl>
```

countData is the count matrix
(number of reads mapping to each gene for each sample)

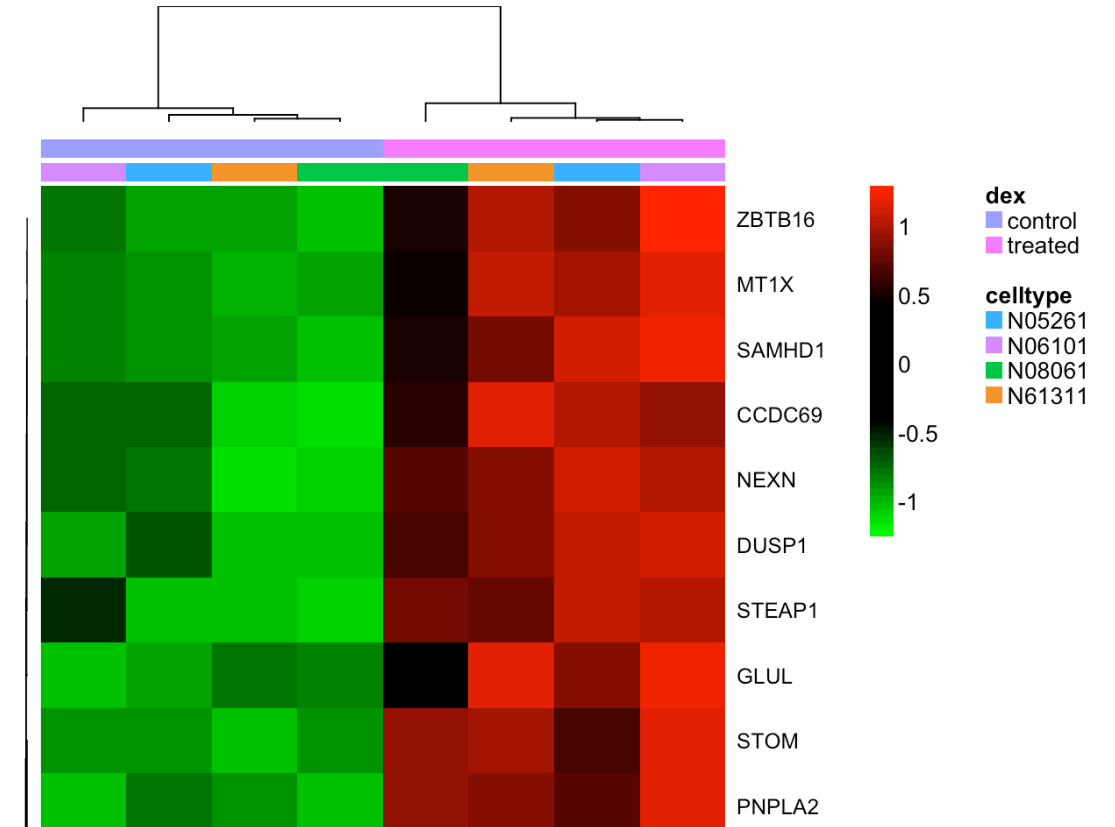
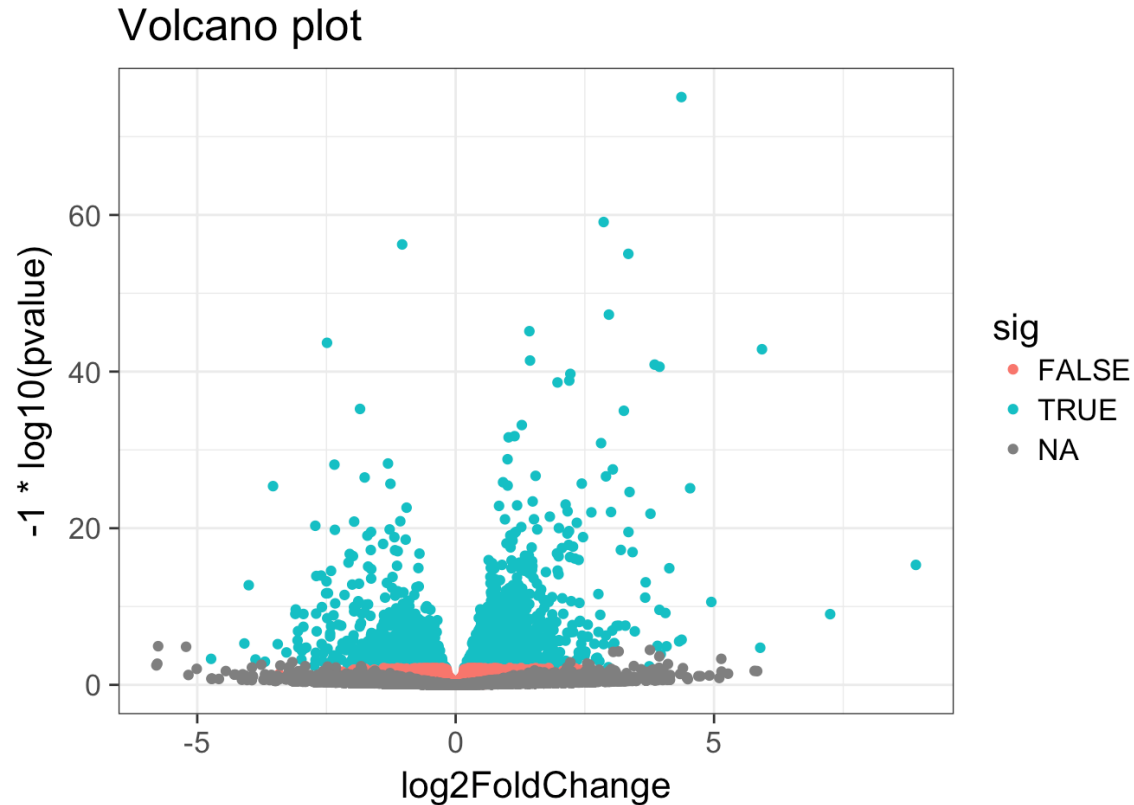
colData describes metadata about the *columns* of countData

First column of colData must match column names of countData (-1st)

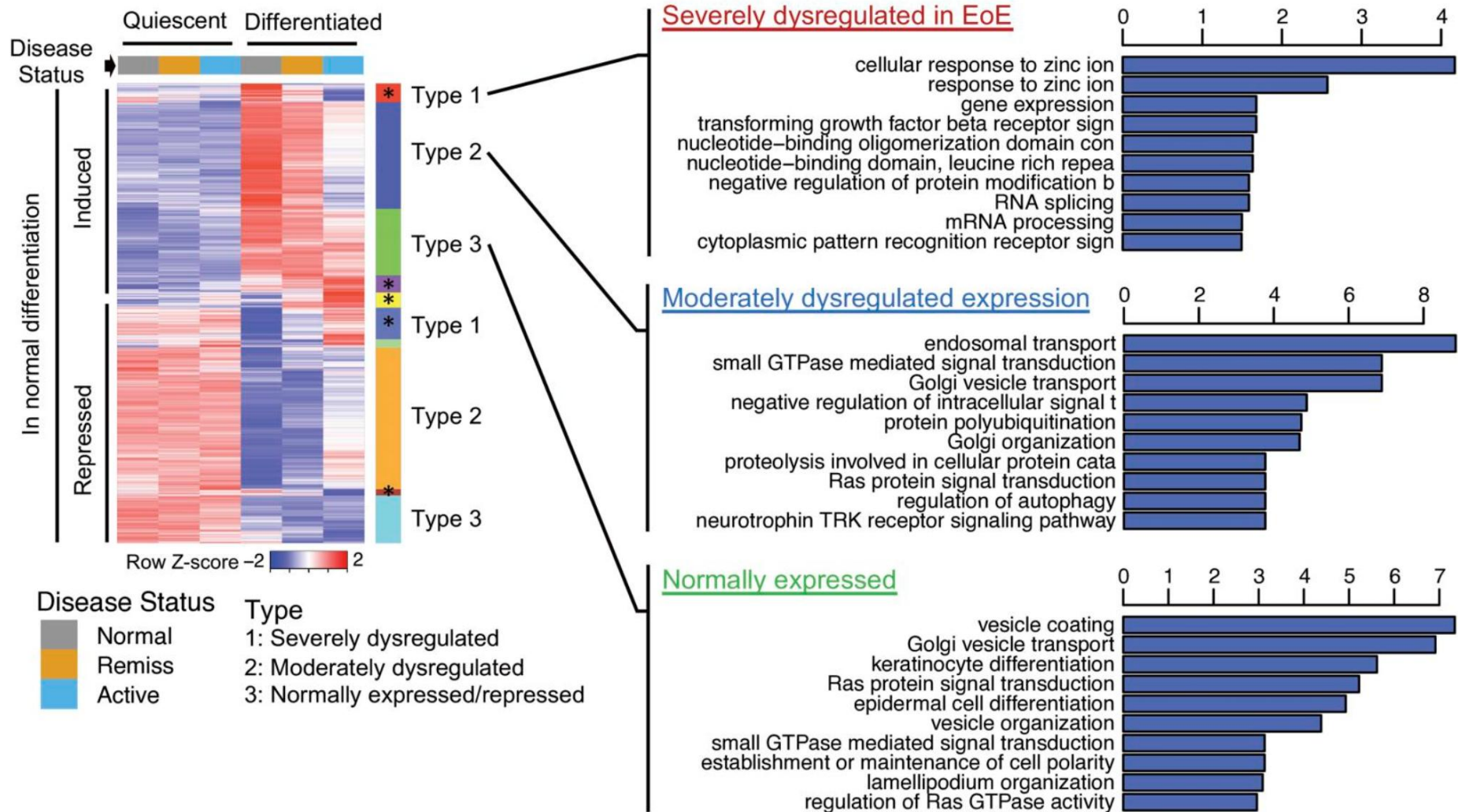
RNA-seq Downstream Analysis



RNA-seq Downstream Analysis



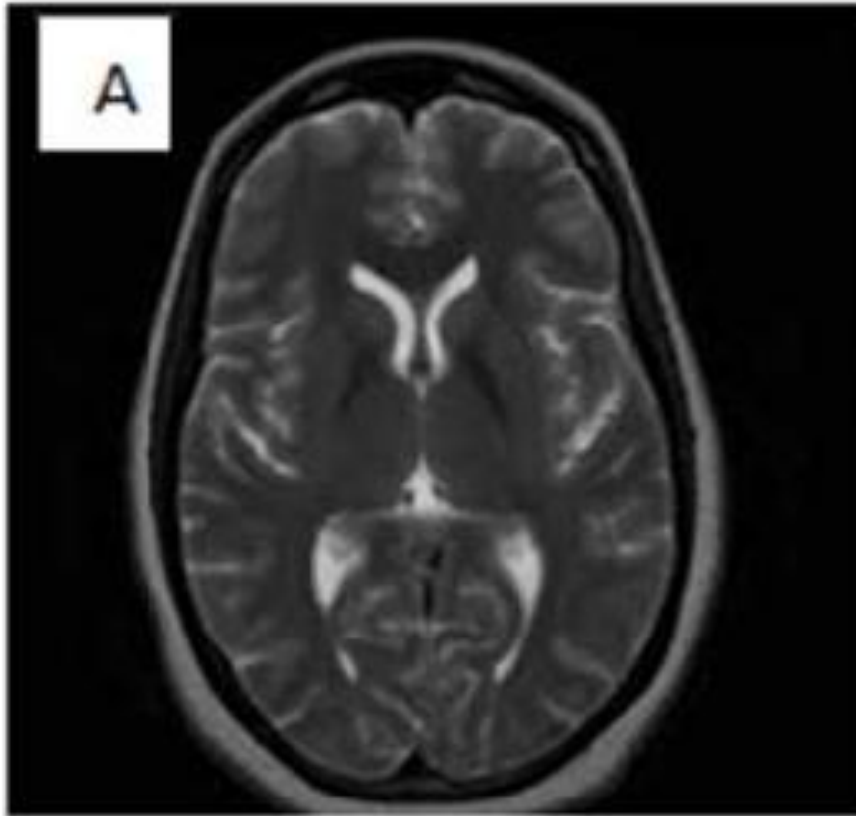
RNA-seq Downstream Analysis



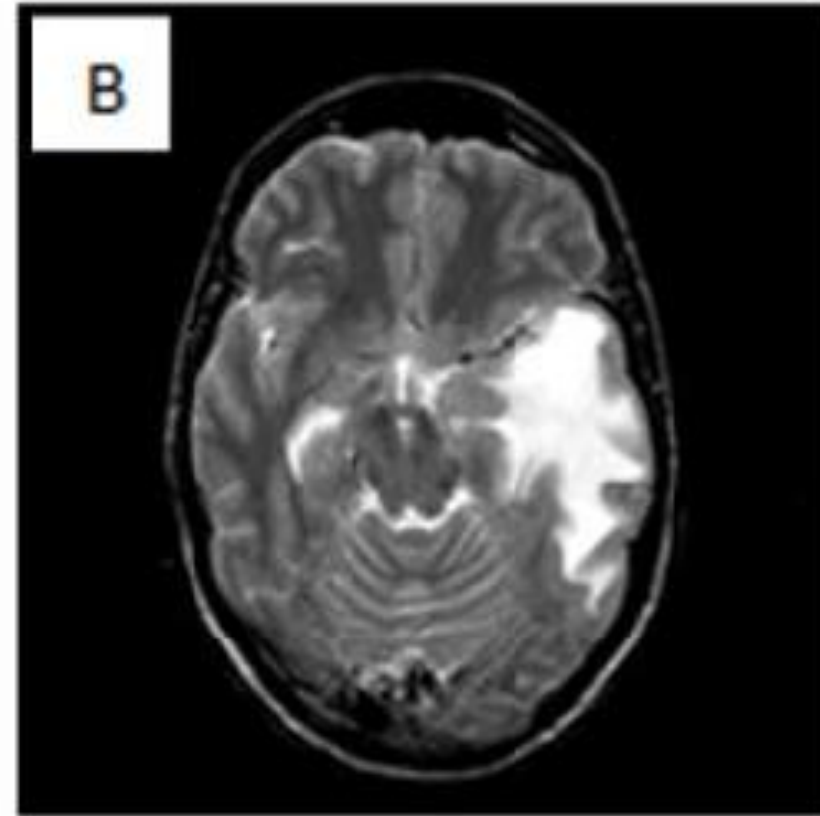
Medical Data

- Text (Patient Medical Record or Health Record)
- Image (Hematoxylin Eosin (HE), Immunohistochemistry (IHC) CT and MRI)
- Video (Medical Ultrasonography and Endoscopy)
- Signal (Electrocardiogram (ECG) and Electroencephalogram (EEG))

MRI



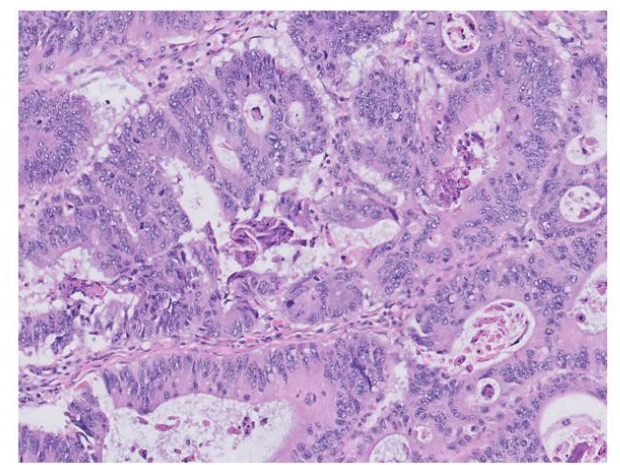
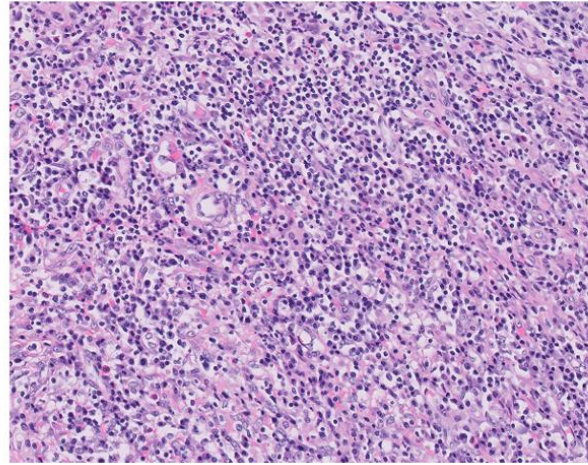
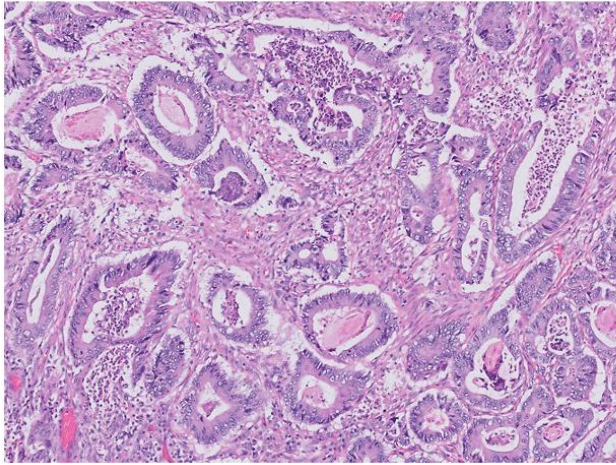
(A)-Normal brain



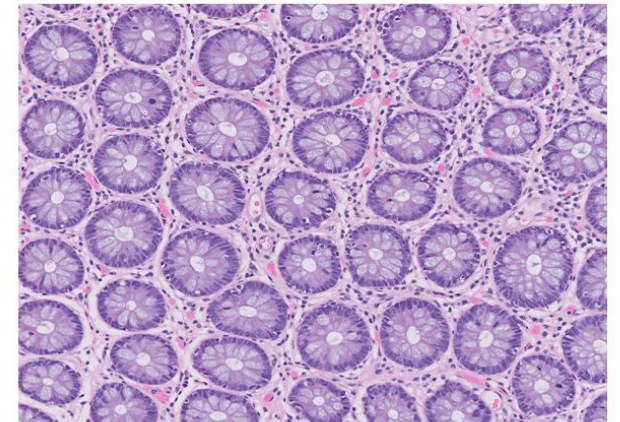
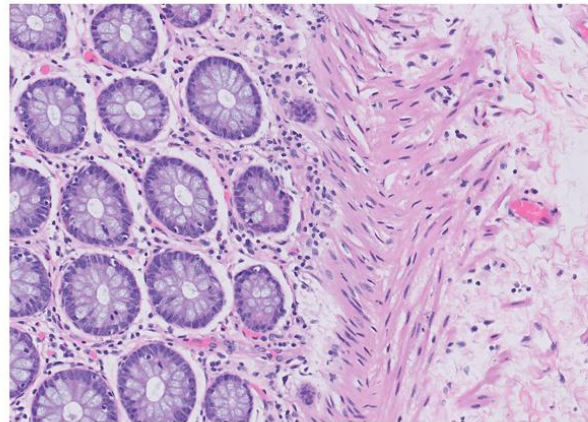
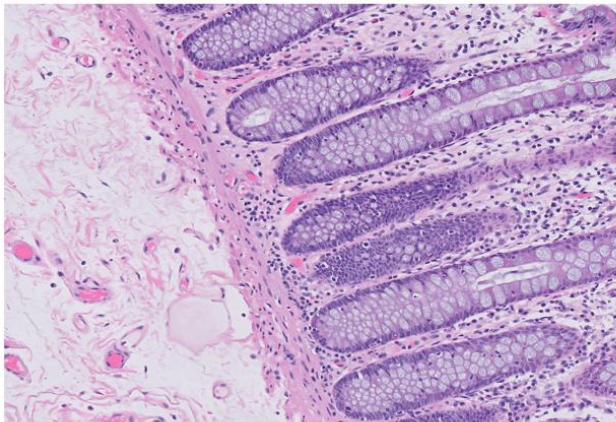
(B)-Tumor Brain

Hematoxylin Eosin

**Normal
(10X)**



**Tumor
(10X)**

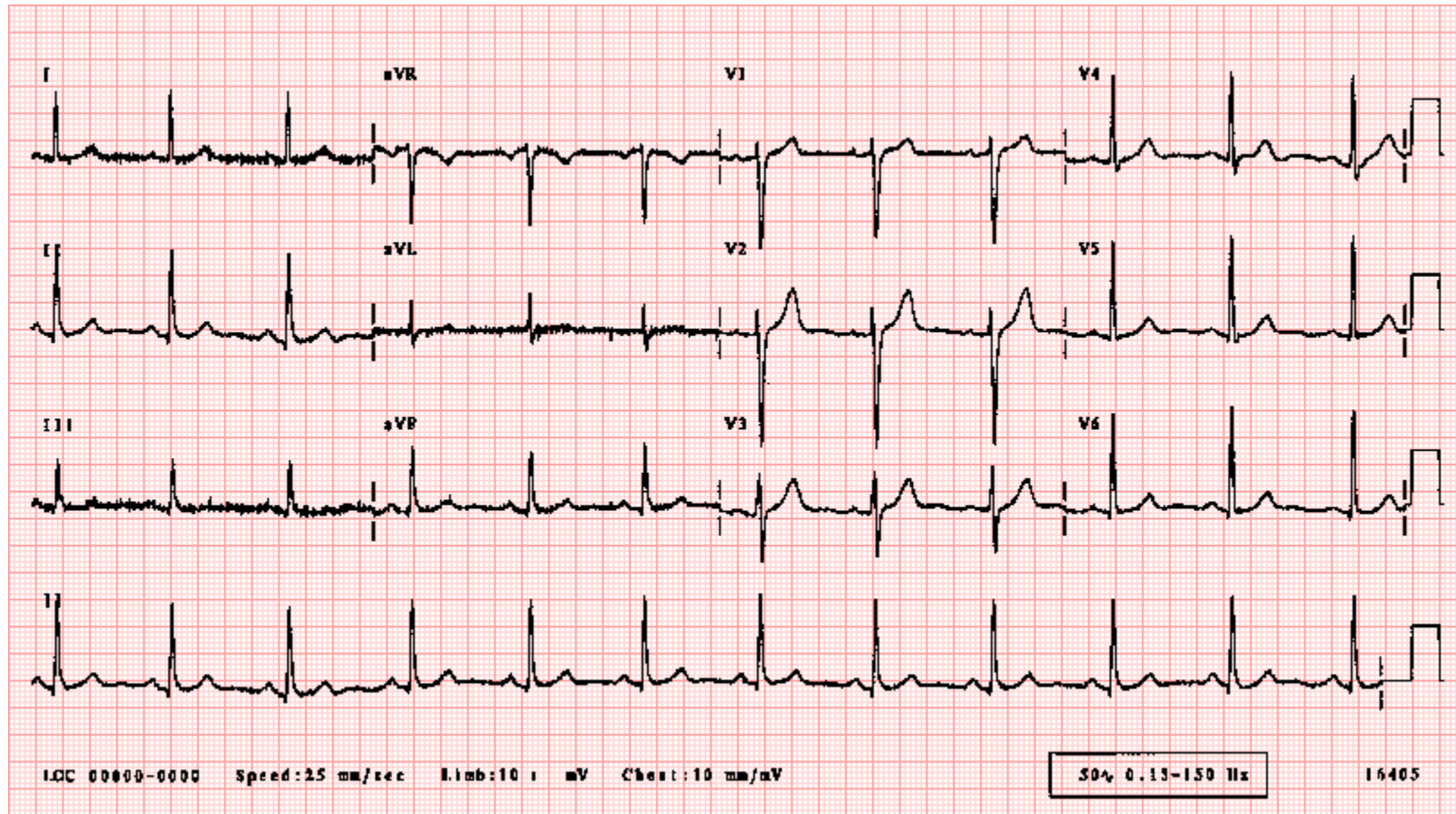


Medical Ultrasonography



https://commons.wikimedia.org/wiki/File:Ultrasound_Scan_ND_0105143736_1439320.gif

ECG



Thank you!