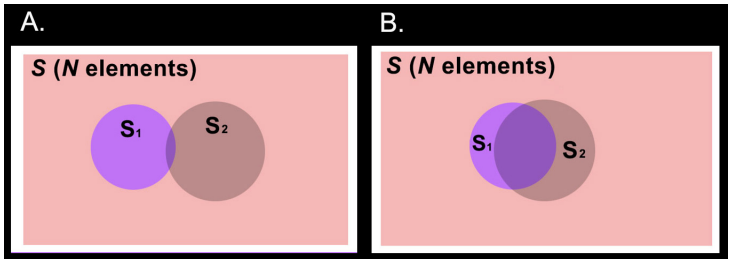
University of Pennsylvania

BIOL4536 Fall 2023

Professor: Gregory R. Grant

Final Exam (Practice #1)

Question 1. If we model pathway enrichment analysis with a hypergeometric distribution, then Which of the following has a smaller p-value?

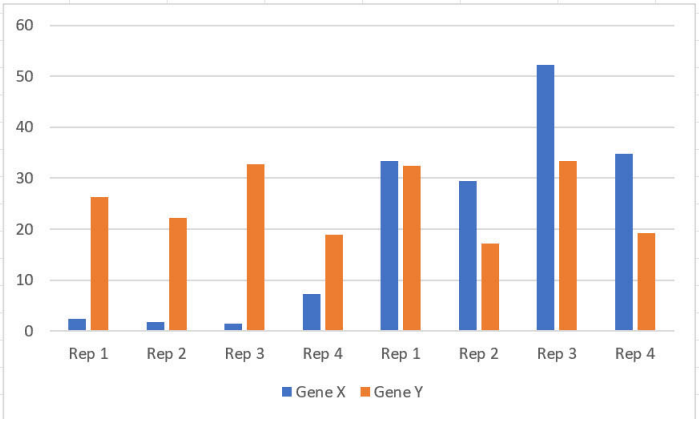


Question 2. What causes us to need to correct pathway enrichment p-values for multiple testing?

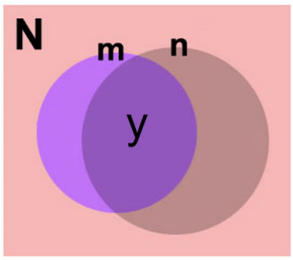
Question 3. True or False. When doing a differential expression pathway enrichment analysis, raising the q-value cutoff used to produce the DE list will always lead to an equal or lower enrichment q-values.

Question 4. This is a figure from the slides on GSEA (Gene Set Enrichment Analysis). Where do those numbers come from in the column all the way on the right with a question mark? In other words how are they defined? We're not looking for a mathematical formula, just their defining property.

	Condition 1				Condition 2				?
	Rep 1	Rep 2	Rep 3	Rep 4	Rep 1	Rep 2	Rep 3	Rep 4	
Gene X	2.4	1.8	1.4	7.2	33.4	29.42	52.17	34.83	0.936986436
Gene Y	26.3	22.24	32.75	18.91	32.4	17.2	33.4	19.23	0.039800984
Class	0	0	0	0	1	1	1	1	

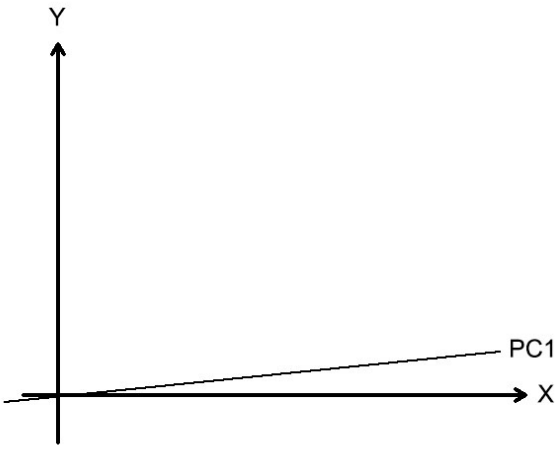


Question 5. This is the diagram we saw in class illustrating the hypergeometric random variable. Which of the things m, n, N and y are random quantities? Circle all that apply.



Question 6. Consider the PC1 subspace in the figure. Circle the one correct answer.

(A) The X loading is higher than the Y loading.
(B) The X loading is equal to the Y loading.
(C) The X loading is lower than the Y loading.
(D) None of the above can be said definitively.



Question 7. True or False. A latent variable can combine the information from multiple genes (dimensions) into one variable (dimension).

Question 8. In Principle Components Analysis, to obtain PC1, we project onto the one-dimensional subspace. In this subspace the data has

(A) Strictly greater variance as it had in the original space.
(B) Strictly less variance as it had in the original space.
(C) Greater or equal variance as it had in the original space.
(D) Less or equal variance as it had in the original space.

Question 9. True or False. Principle Components Analysis is a linear method and UMAP is non-linear.

Question 10. The null hypothesis for a Mann-Whitney test is (circle one):

(A) Equal means
(B) Equal means and variances
(C) Equal medians
(D) Equal distributions
(E) Equal standard deviations

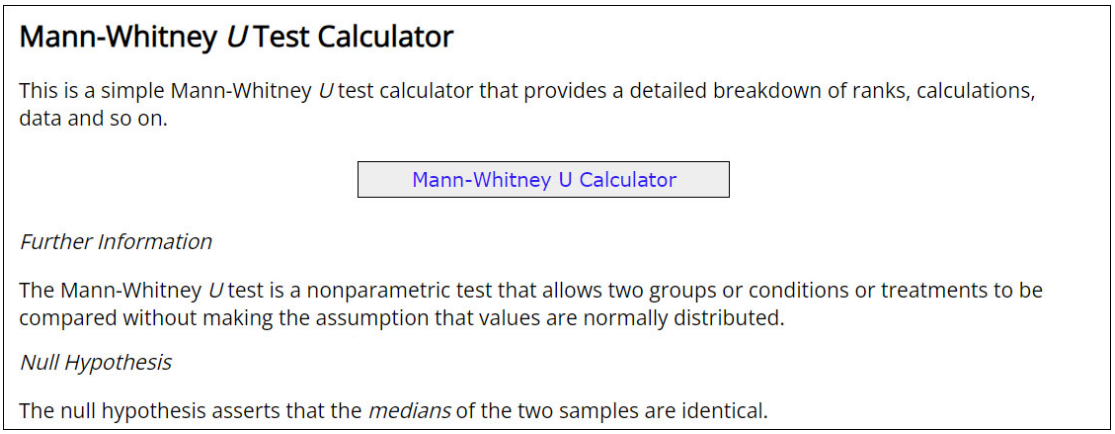
Question 11. In your own words, what's the difference between technical and biological variation in a data set.

Question 12. Consider the two data sets shown below. True or False. The Mann-Whitney p-value is the same for both data sets.

Data Set 1	
condition1	condition2
5	8
4	7
3	6

Data Set 2	
condition1	condition2
5	800
4	7
3	600

Question 13. This is a screen shot detail from a webpage with a Mann-Whitney calculator. The authors of this page made a grievous mistake, what is it?



Not that it matters, but the website is: `https://www.socscistatistics.com/tests/mannwhitney`

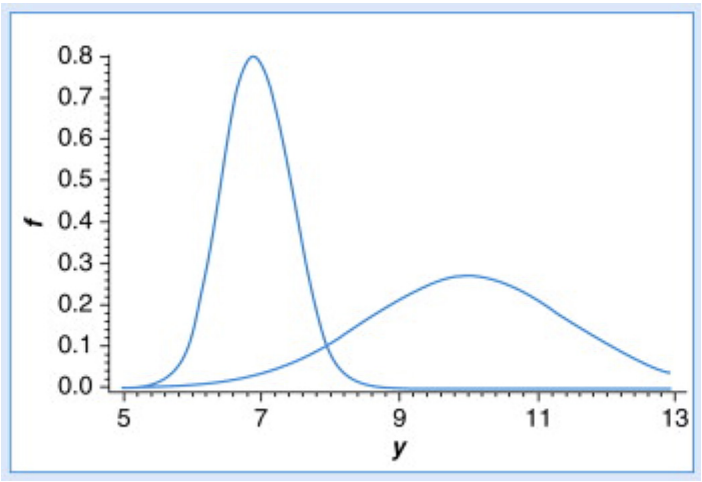
Question 14. True or False. Non-parametric tests tend to be blind to outliers.

Question 15. It would be legitimate to compare these two distributions using (circle all that apply)

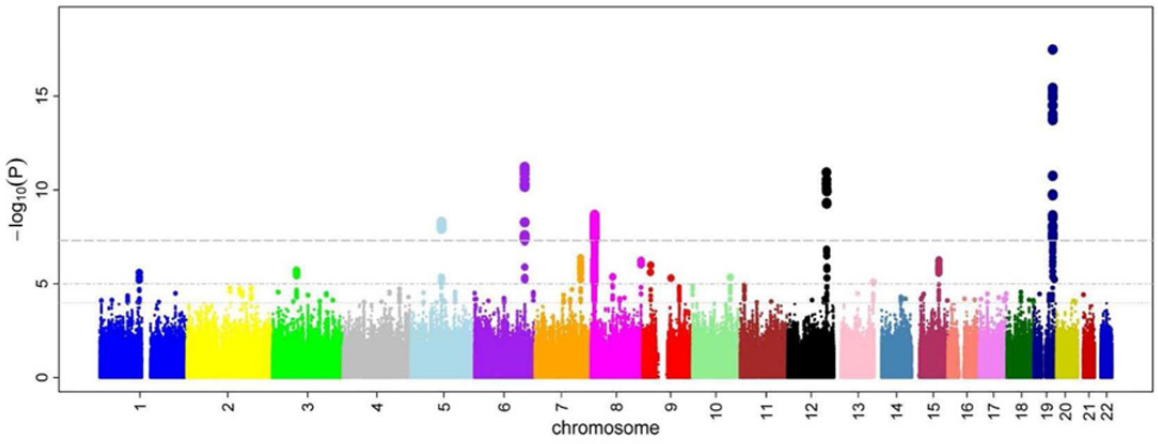
(A) Mann-Whitney

(B) A Permutation test

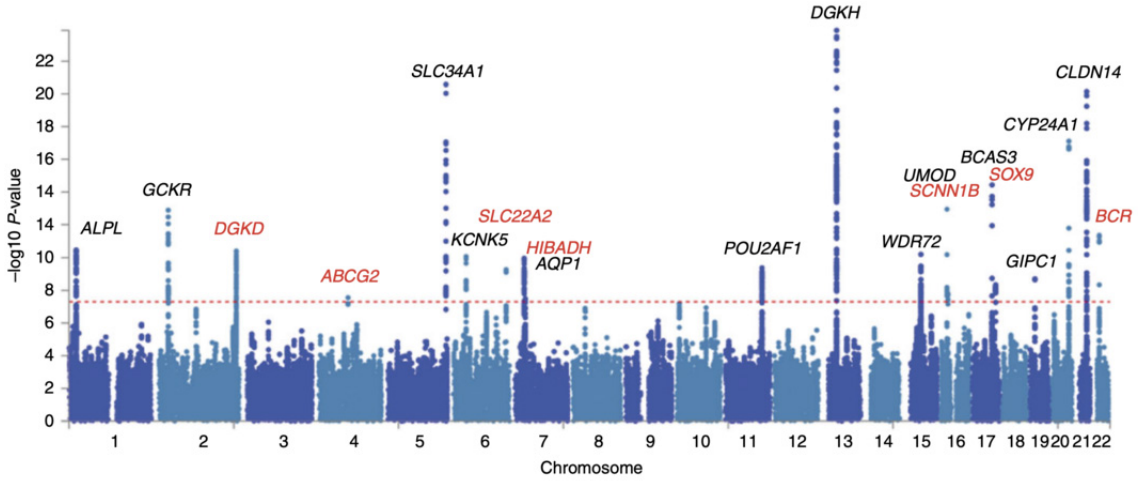
(C) A parametric T-test



Question 16. True or False. It is called a Manhattan plot because multiple points are in the exact same genomic location.



Question 17. True or False. At each locus in a Manhattan plot, the SNP with the smallest p-value (highest on the vertical axis) is the causative SNP.



Question 18. What is the multiple testing issue in GWAS?

(A) Multiple genes
(B) Multiple subjects
(C) Multiple phenotypes
(D) Multiple chromosomes
(E) Multiple SNPs

Question 19. Is it necessary to run a test for significant association on the following data?

	Controls	Cases
A/A	100	1000
A/B	200	2000
B/B	900	9000

Question 20. True or False. A polygenic risk score combines information from multiple SNPs into one score that assesses risk of one genetically associated condition.

Question 21. Connect the thing on the left with the correct thing on the right.

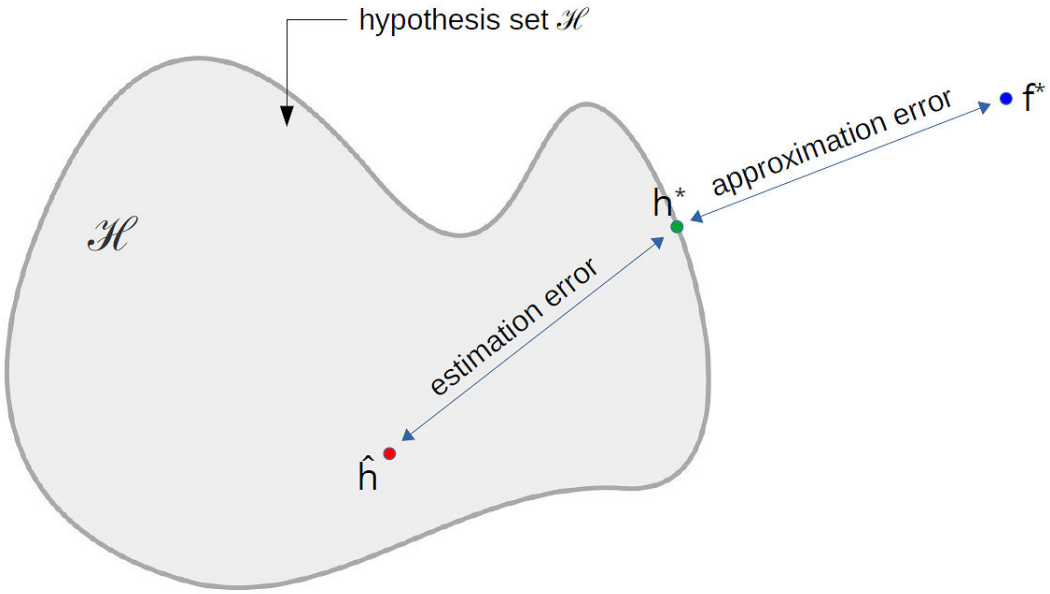
Unsupervised Learning	Determine how many disease subtypes there are.
Supervised Learning	Predict disease subtype using test results as features

Question 22. In the figure, which functions are generally unknowable? Circle all that apply.

(A) $\hat{h}$

(B) h^*

(C) f^*



Question 23. True or False. For a continuous dependent variable, we use the absolute value loss function $|\hat{y} - y|$ instead of the quadratic loss function $(\hat{y} - y)^2$ because the former is differentiable.

Question 24. True or False. An element of the hypothesis set is a function of the dependent variable.

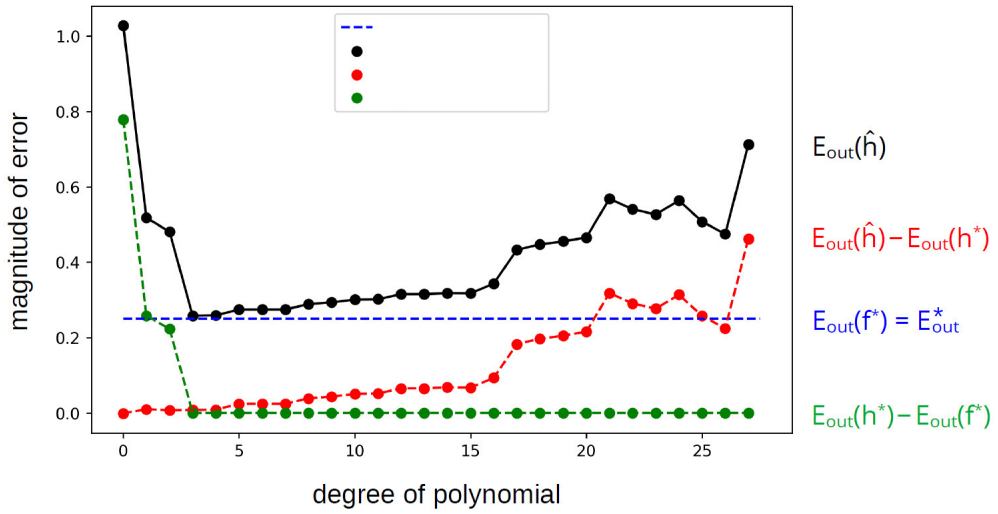
Question 25. In the figure, which color line represents the Bayes Risk?

(A) black

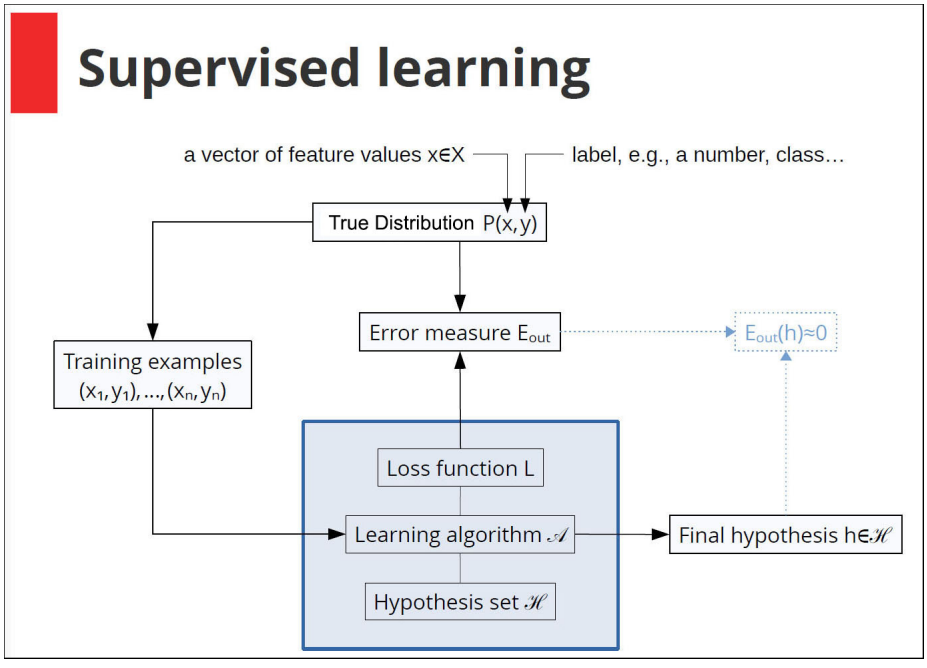
(B) blue

(C) orange

(D) green



Question 26. Refer to the diagram below. True or False, the Final hypothesis $h \in \mathcal{H}$ depends on the choice of Loss function L.



Question 27. Draw a line between each R data structure and the matching description

Data structure	Description
data frame	One-dimensional, where each element can have a different type
list	One-dimensional, where each element must have the same data type
matrix	Two-dimensional table, where all elements must have the same data type
vector	Two-dimensional table, where each column can have a different data type

Question 28. Here are the first six lines from a text file:

```
species,island,bill_length_mm,year
Adelie,Torgersen,39.1,2007
Adelie,Torgersen,39.5,2007
Adelie,Torgersen,40.3,2007
Adelie,Torgersen,NA,2007
Adelie,Torgersen,36.7,2007
```

Which R function would you use to read this file into R?

(A) read_xlsx()
(B) read_csv()
(C) read_tsv()
(D) read_RDS()

Question 29. Consider the following function definition:

```r
get_de_genes <- function(de_results,
                         de_method,
                         q_value_cutoff = 0.05,
                         log2_fc_cutoff = 1) {
    # Body of the function
}
```

If we call the function with the following code, what value is assigned to the 'q_value_cutoff' argument?

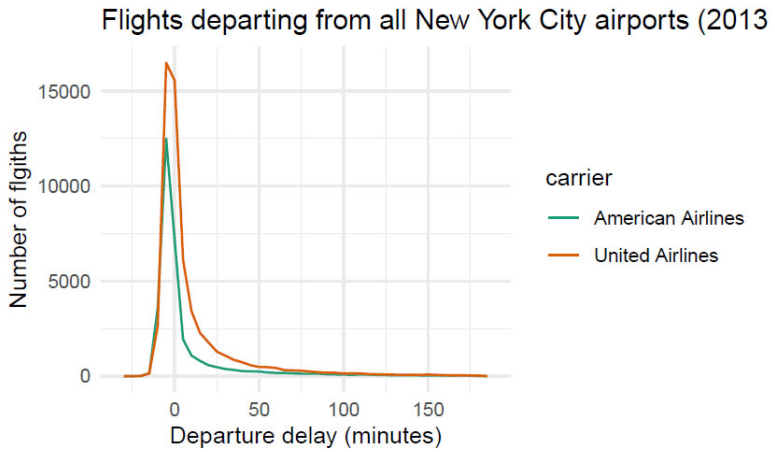
```r
input_data |>
    get_de_genes(log2_fc_cutoff = 2,
                 "limma")
```

(A) contents of 'intput_data'
(B) 2
(C) 0.05
(D) "limma"
(E) 1

Question 30. True or False. SummarizedExperiement objects are designed to store the results of an experimental assay, along with metadata describing the experiment?

Question 31. Consider this table and graph

```
# A tibble: 6 x 6
  flight carrier           sched_dep_time dep_delay sched_arr_time arr_delay
   <int> <chr>                      <int>     <dbl>          <int>     <dbl>
1   1545 United Airlines              515         2            819        11
2   1714 United Airlines              529         4            830        20
3   1141 American Airlines            540         2            850        33
4   1696 United Airlines              558        -4            728        12
5    301 American Airlines            600        -2            745         8
6    194 United Airlines              600        -2            917         7
```



To draw this graph, which aesthetic(s) would you need to map? (Circle all that apply)

(A) *x*
(B) *y*
(C) shape
(D) color

Question 32. True or False. These two R expressions are equivalent.

```r
penguins |> mutate(flipper_length_cm = flipper_length_mm / 10)

mutate(flipper_length_cm = flipper_length_mm / 10, penguins)
```

Question 33. Consider these two tibbles:

Tibble A:

```
# A tibble: 24 x 3
  gene_name sample_id   read_counts
  <chr>     <chr>             <int>
1 Lcn2      Saline_9574          63
2 Lcn2      Saline_9575          41
3 Lcn2      IL1B_9577         39976
4 Lcn2      IL1B_9578         44056
5 Ido2      Saline_9574        1734
6 Ido2      Saline_9575        1129
# i 18 more rows
```

Tibble B:

```
# A tibble: 6 x 5
  gene_name Saline_9574 Saline_9575 IL1B_9577 IL1B_9578
  <chr>           <int>       <int>     <int>     <int>
1 Lcn2               63          41     39976     44056
2 Ido2             1734        1129       280       230
3 Fam83a              6           5        94       210
# i 3 more rows
```

Which R function would you use to reshape Tibble A into Tibble B?

(A) `left_join()`
(B) `pivot_longer()`
(C) `pivot_wider()`
(D) `bind_rows()`