Classical Statistics Biological Big Data Supervised and Unsupervised Learning

Supervised Learning: Introduction

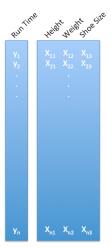
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A Simple Example

- ▶ Suppose we have n = 500 kids for whom we have p = 3 measurements: height, weight, and shoe size.
- ► We wish to predict these kids' 1600-meter run times using these measurements.

A Simple Example



Notation:

- ▶ *n* is the number of observations.
- ▶ p the number of variables/features/predictors.
- y is a n-vector containing response/outcome for each of n observations.
- ▶ X is a $n \times p$ data matrix.

Linear Regression on a Simple Example

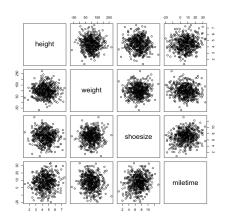
► You can perform linear regression to develop a model to predict run time using height, weight, and shoe size:

$$y = \beta_0 + \beta_1 X_1 + \beta_2 X_2 + \beta_3 X_3 + \epsilon$$

where y is run time, X_1, X_2, X_3 are height, weight, and shoe size, and ϵ is a noise term.

- ➤ You can look at the coefficients, p-values, and t-statistics for your linear regression model in order to interpret your results.
- ► You learned everything (or most of what) you need to analyze this data set in AP Statistics!

A Relationship Between the Variables?



Linear Model Output

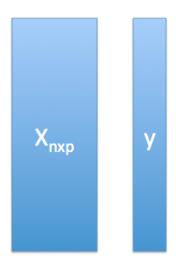
	Estimate	Std. Error	T-Stat	P-Value
Intercept	-2.265831	2.644654	-0.857	0.39199
height	1.074814	0.414789	2.591	0.00985 **
weight	-0.021155	0.008482	-2.494	0.01295 *
shoesize	0.955222	0.214449	4.454	1.04e-05 ***

 $RunTime \approx -2.27 + 1.07 \times Height - 0.021 \times Weight + 0.96 \times ShoeSize.$

Low-Dimensional Versus High-Dimensional

- ▶ The data set that we just saw is low-dimensional: $n \gg p$.
- Lots of the data sets coming out of modern biological techniques are high-dimensional: $n \approx p$ or $n \ll p$.
- This poses statistical challenges! AP Statistics no longer applies.

Low Dimensional



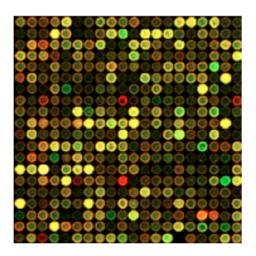
High Dimensional



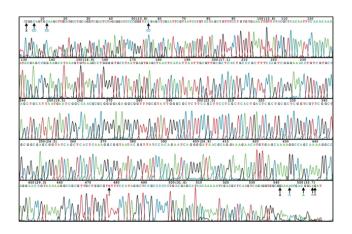
What Goes Wrong in High Dimensions?

- Suppose that we included many additional predictors in our model, such as
 - ► 50-yard dash time
 - ► Age
 - Zodiac symbol
 - ▶ Favorite color
 - ▶ Mother's birthday, in base 2
- ► Some of these predictors are useful, others aren't.
- ▶ If we include too many predictors, we will overfit the data.
- ► Overfitting: Model looks great on the data used to develop it, but will perform very poorly on future observations.
- ▶ When $p \approx n$ or p > n, overfitting is guaranteed unless we are very careful.

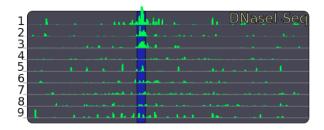
Gene Expression Data



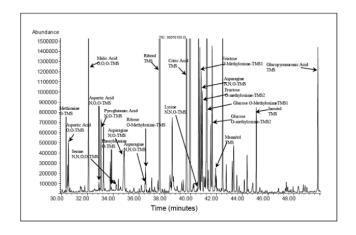
DNA Sequence Data



DNAse Hypersensitivity Data



Metabolomic Data



High-Dimensional Omics Analyses

For most omics analyses, we have many more variables than observations.... i.e. $p \gg n$.

- ▶ Predict risk of diabetes on the basis of DNA sequence data.... using n = 1000 patients and p = 3000000 variables.
- ► Cluster tissue samples on the basis of DNase hypersensitivity... using n = 200 cell types and p = 1000000000 variables.
- ▶ Identify genes whose expression is associated with survival time... using n = 250 cancer patients and p = 20000 variables.

Why Does Dimensionality Matter?

- Classical statistical techniques, such as linear regression, cannot be applied.
- ► Even very simple tasks, like identifying variables that are associated with a response, must be done with care.
- ► High risks of overfitting, false positives, and more.

This course: Statistical machine learning tools for big – mostly high-dimensional – data.

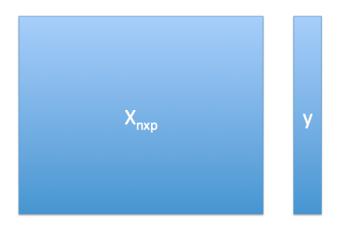
Statistical Machine Learning



Supervised and Unsupervised Learning

- ► Statistical machine learning can be divided into two main areas: supervised and unsupervised.
- ► Supervised Learning: Use a data set *X* to predict or detect association with a response *y*.
 - ► Regression
 - Classification
 - Hypothesis Testing
- ► Unsupervised Learning: Discover the signal in X, or detect associations within X.
 - ► Dimension Reduction
 - Clustering

Supervised Learning



Unsupervised Learning



This Course

- ► We will cover the big ideas in supervised learning for big data.
- ▶ The best way to use these methods: learn R.



"Course Textbook" . . . with applications in R



- ► Available for (free!) download from www.statlearning.com.
- ► An accessible introduction to statistical machine learning, with an R lab at the end of each chapter!!
- ▶ We will go through some of these R labs in class.
- ► To learn more, go through them on your own!

Let's Try Out Some R!

Chapter 2 R lab www.statlearning.com