CS 260: Foundations of Data Science

Prof. Sara Mathieson

Spring 2025

HAVERFORD
COLLEGE

Admin

- Reminder: **pair work is required** for the final project – find a partner ASAP or email me!

- **Lab 7 and project proposal** (both short)
  - Due Wednesday

Outline

- Finish data visualization intro
- Dimensionality reduction
- PCA for data visualization

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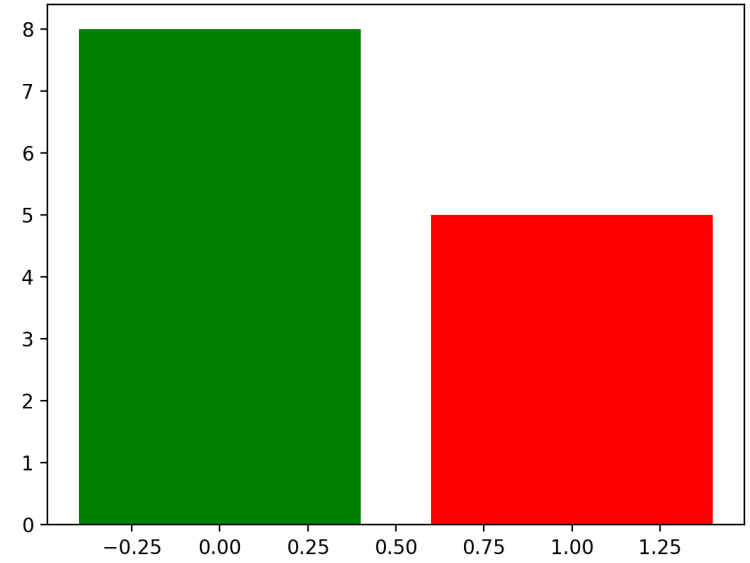
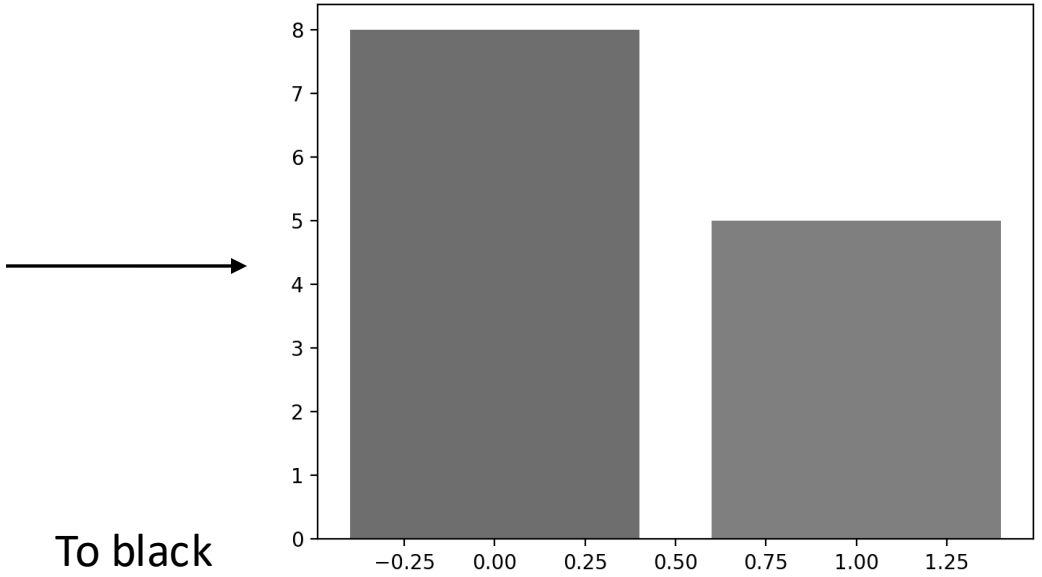
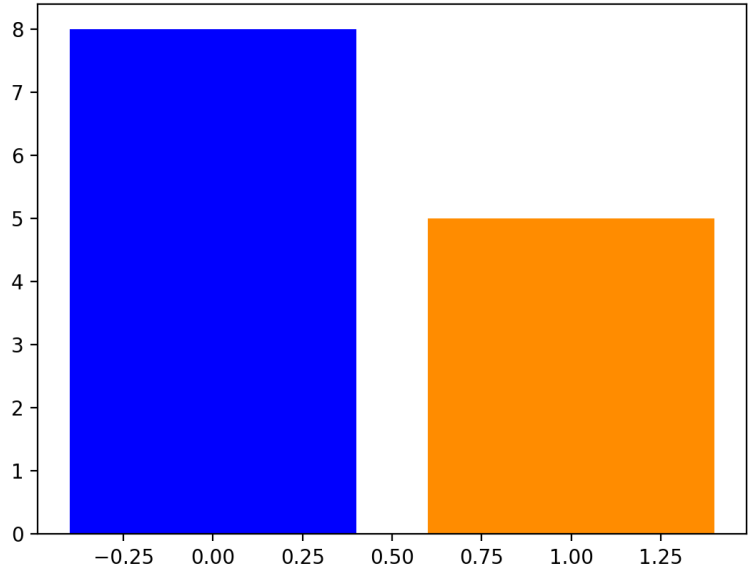
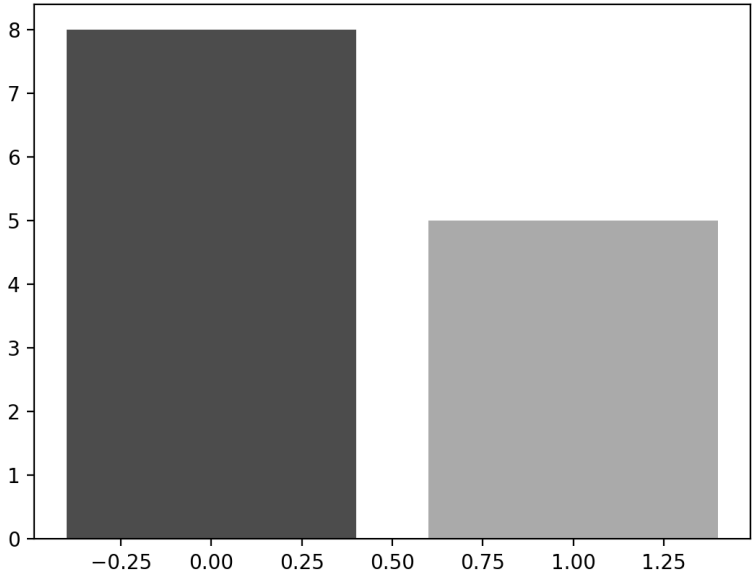
Red/green vs. blue/orange





To black and white





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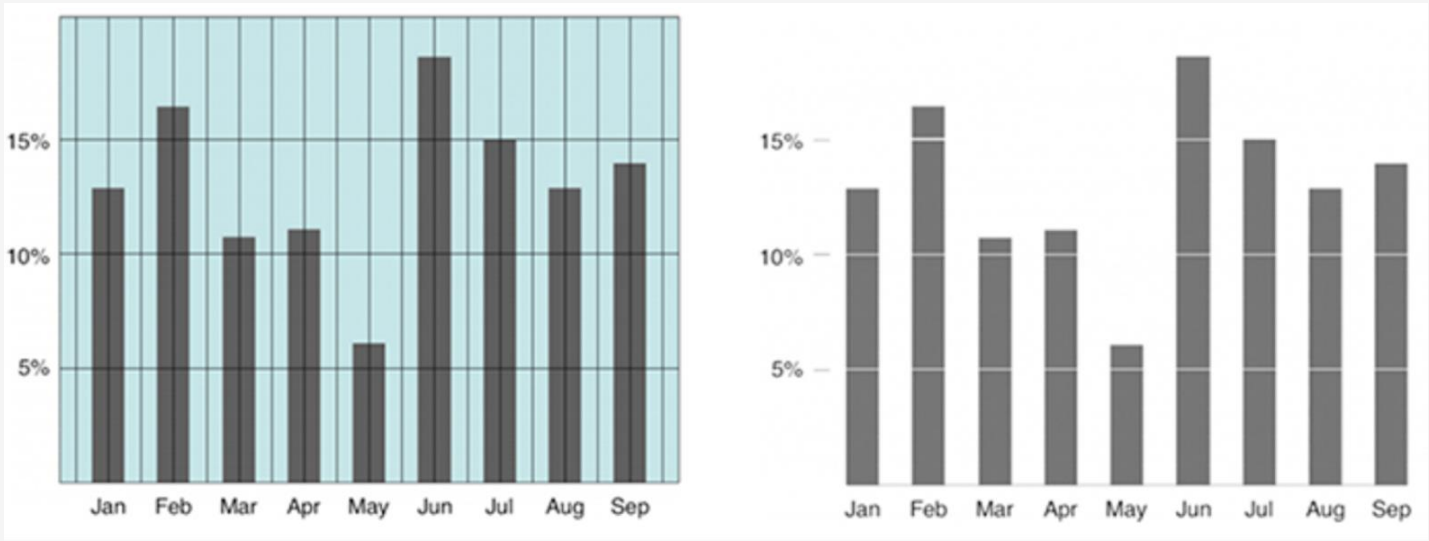

Slide from: Eric Miller

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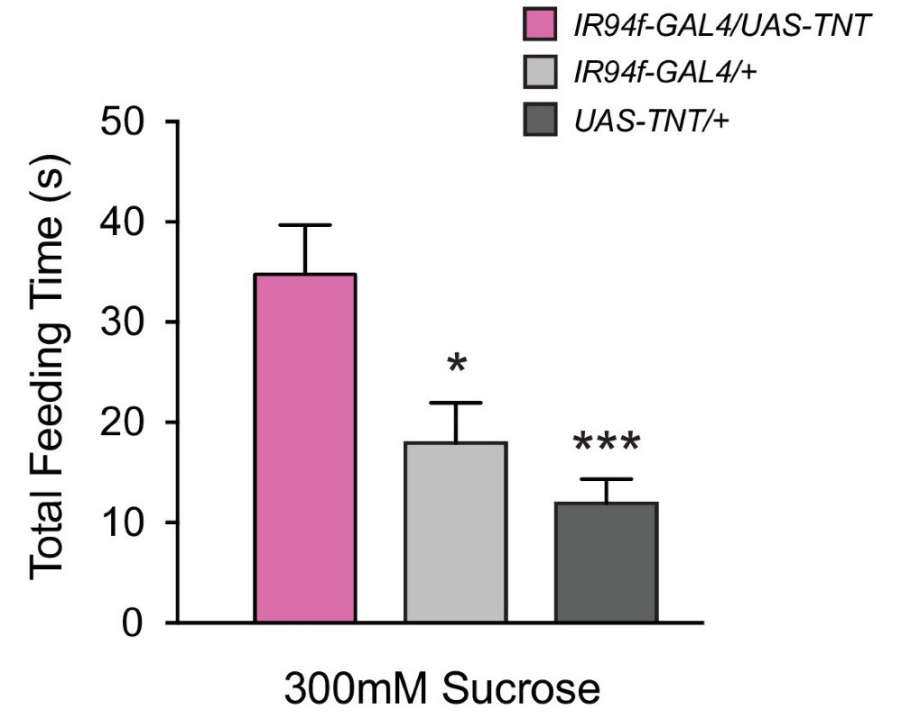

Slide from: Eric Miller

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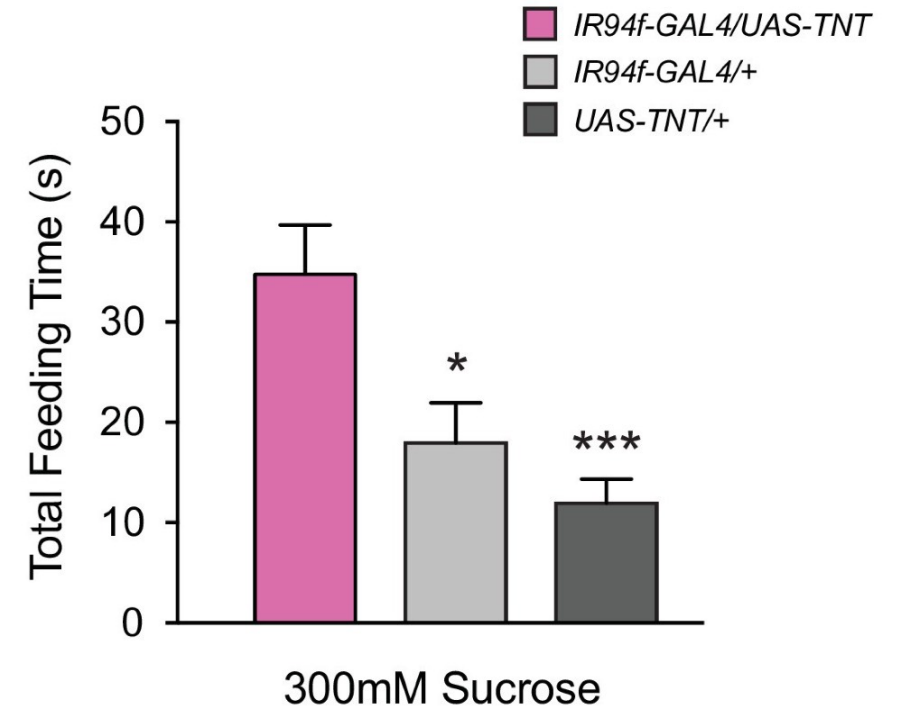

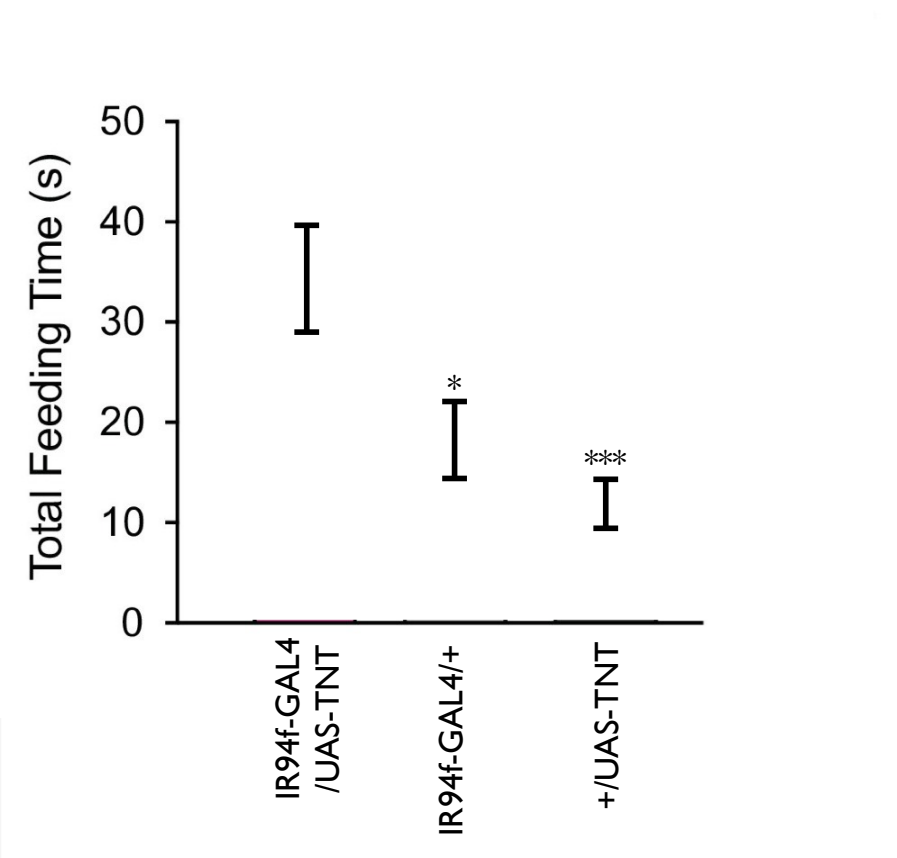

Slide from: Eric Miller

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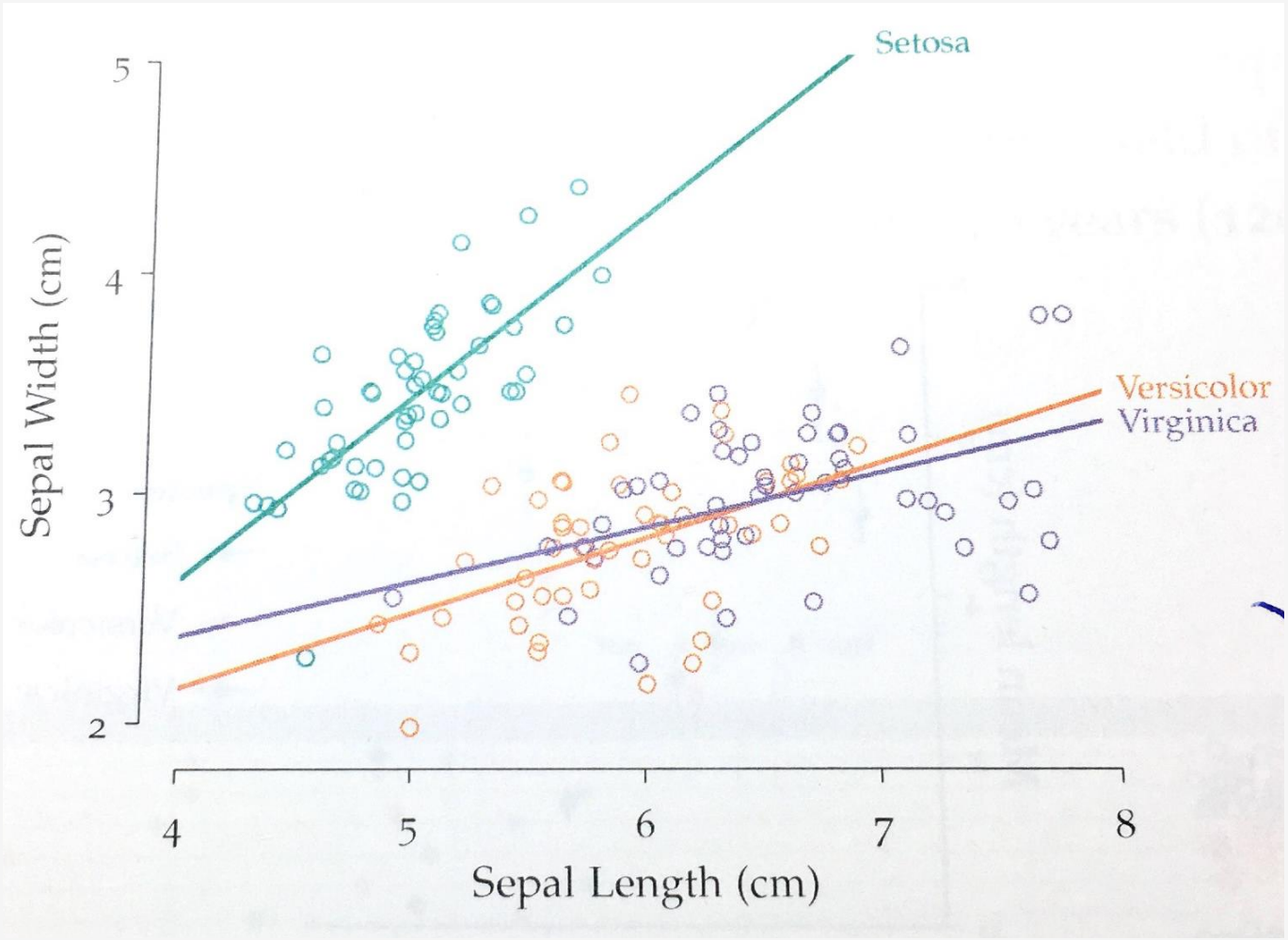


Where is the legend?

Slide from: Eric Miller

Data::Ink

- Remove excess ink
- Show distributions, instead of bars
- Can you remove the legend?
- Remove double encodings
- Is a log scale appropriate?
- What do the ‘error bars’ represent?

Slide from: Eric Miller

Outline

- Finish data visualization intro
- Dimensionality reduction
- PCA for data visualization

Principal Components Analysis (PCA)

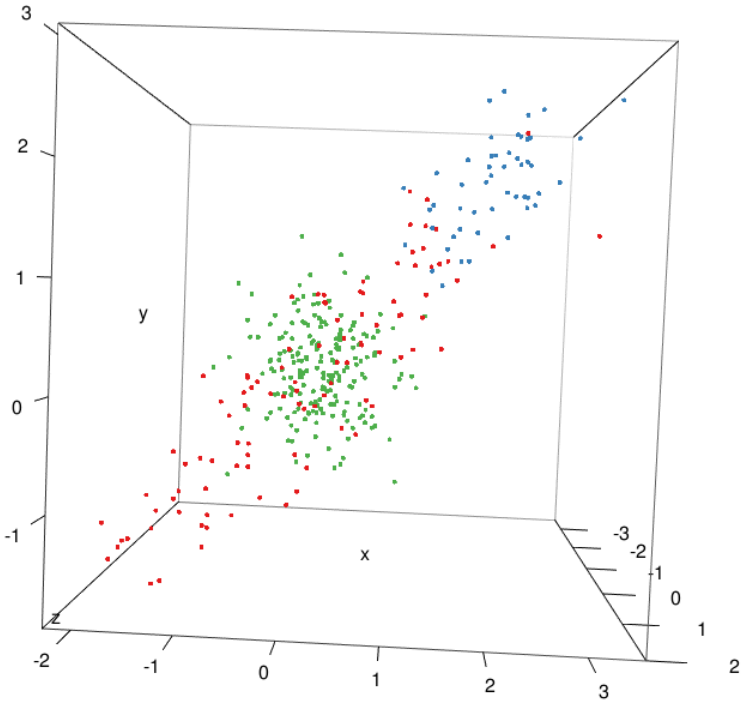
- Transforms *p*-dimensional data so that the new first dimension explains as much of the variation as possible, the new second explains as much of the remaining variation as possible, and so on

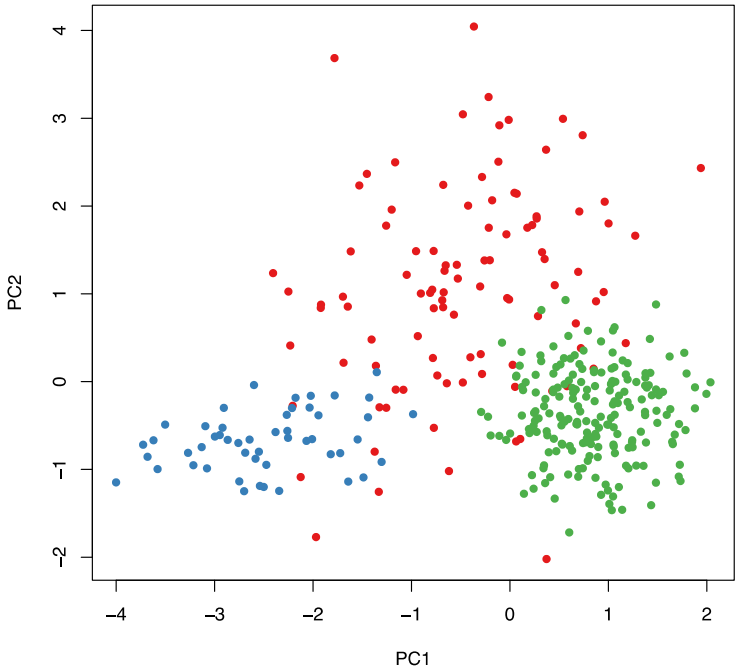
- Typically, we look at the first few dimensions of the transformed data and use as a means of dimensionality reduction and visualization

- PCA is a linear transformation

- PCA is often used for:
  - Data visualization
  - Infer qualitative relationships between groups

Principal component analysis

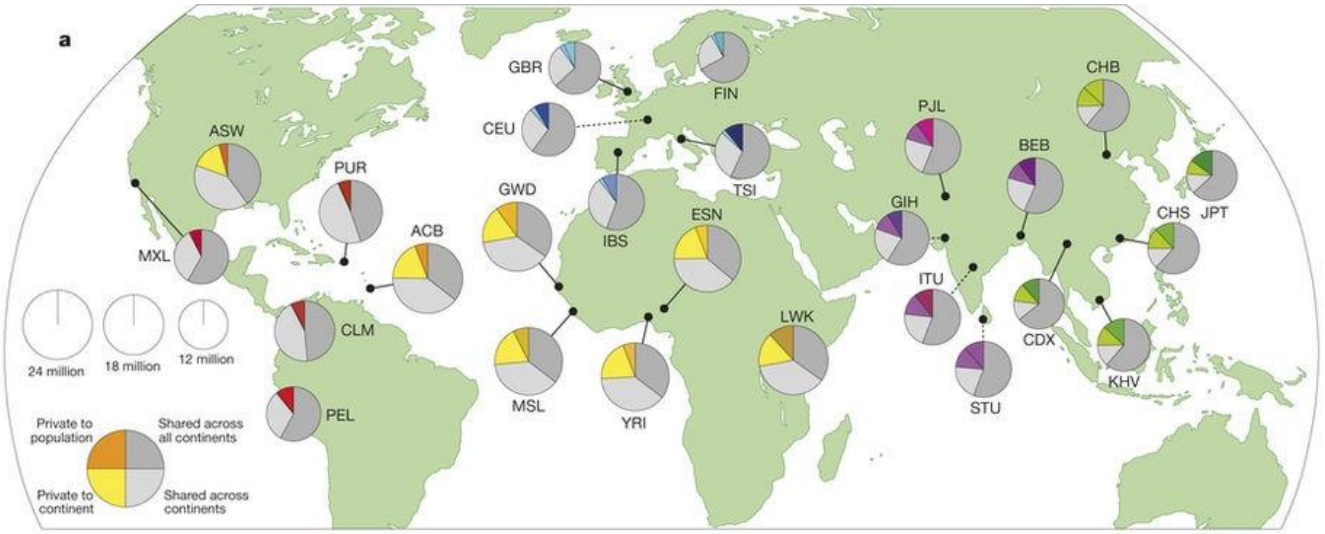




Slide: Iain Mathieson

The 1000 Genomes project

- Whole-genome sequence data from 2504 individuals from 26 populations
- A catalog of human genetic variation, useful as a reference or imputation panel
- Completely public. Download from ftp://ftp-trace.ncbi.nih.gov/1000genomes/



The 1000 Genomes Project Consortium, 2015; https://www.nature.com/articles/nature15393

Slide: Iain Mathieson

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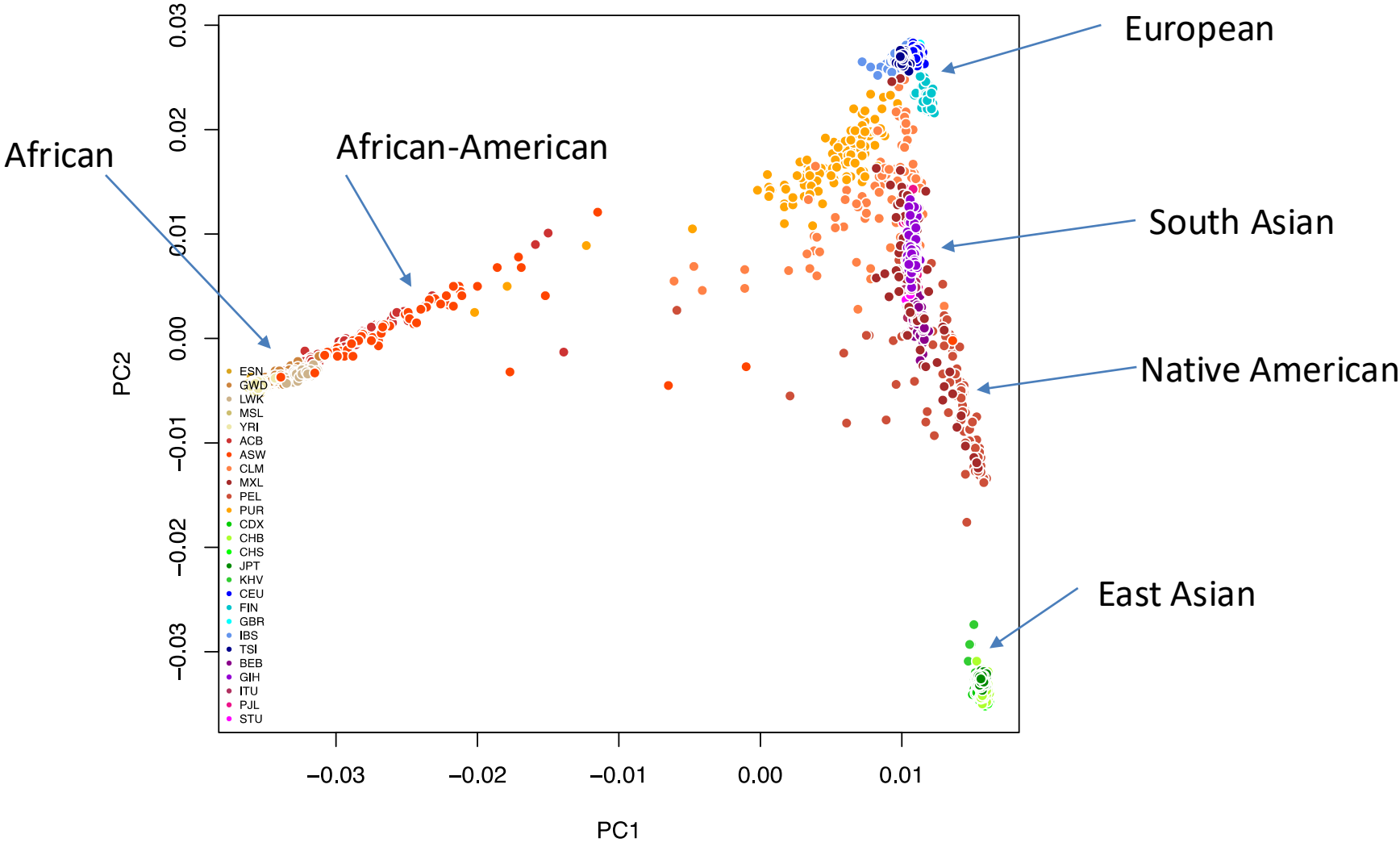
```

Global population structure

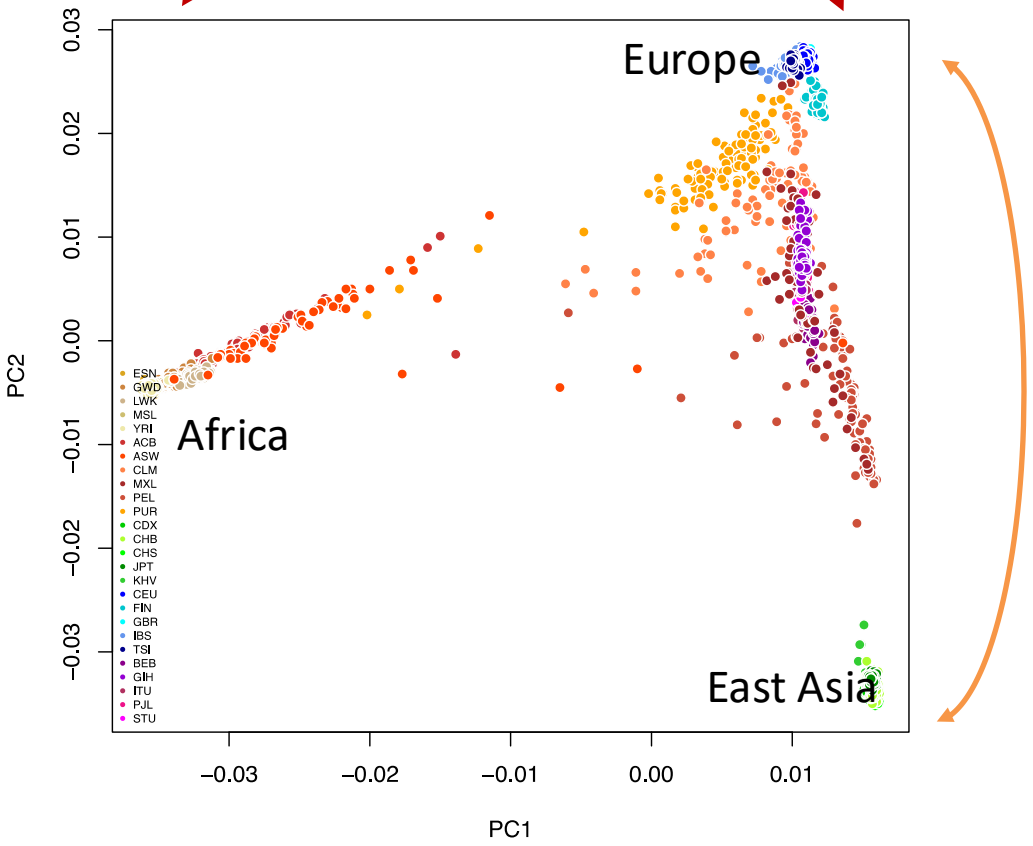


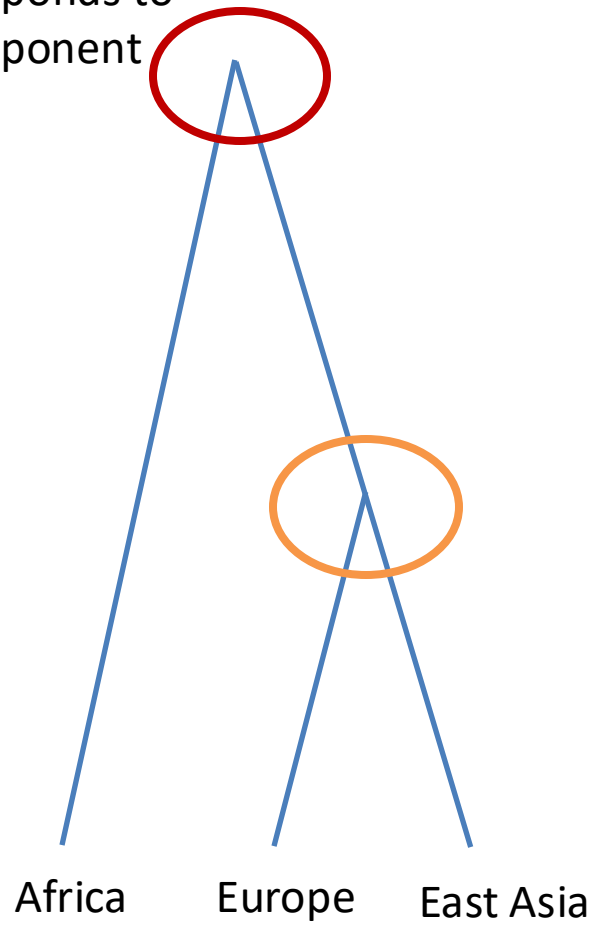
Slide: Iain Mathieson

What causes these patterns?

1. Populations splits separate populations

Oldest split corresponds to first principal component



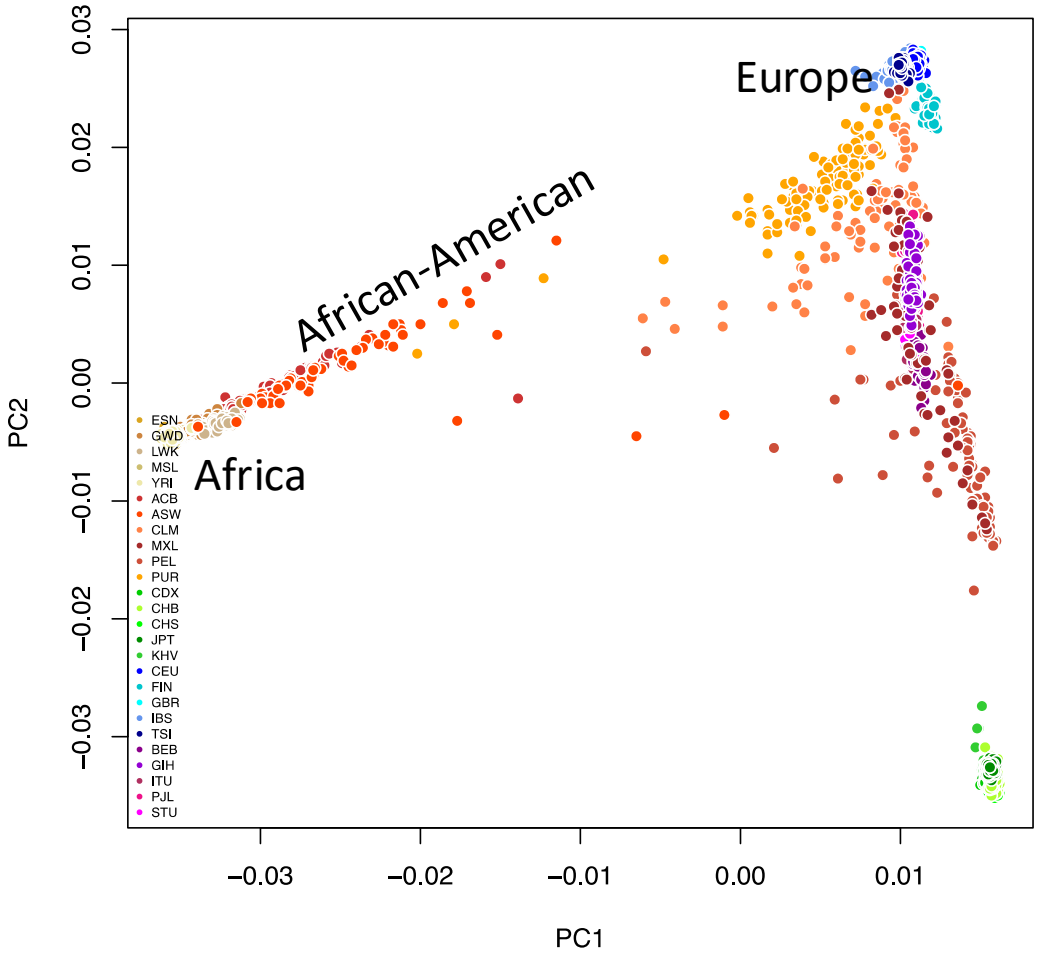


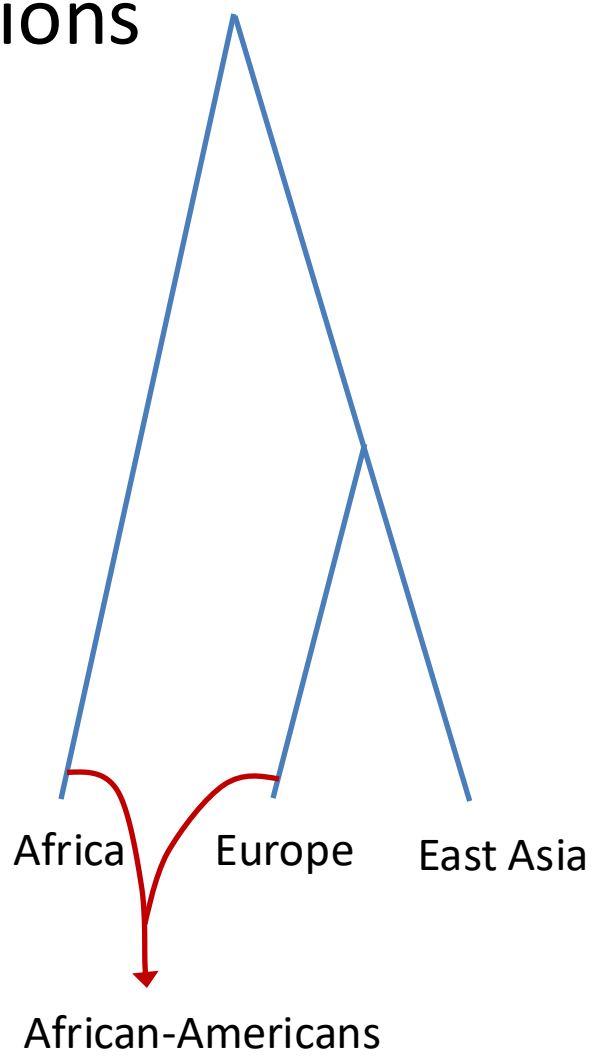
Slide: Iain Mathieson

McVean 2009: http://journals.plos.org/plosgenetics/article?id=10.1371/journal.pgen.1000686

What causes these patterns?

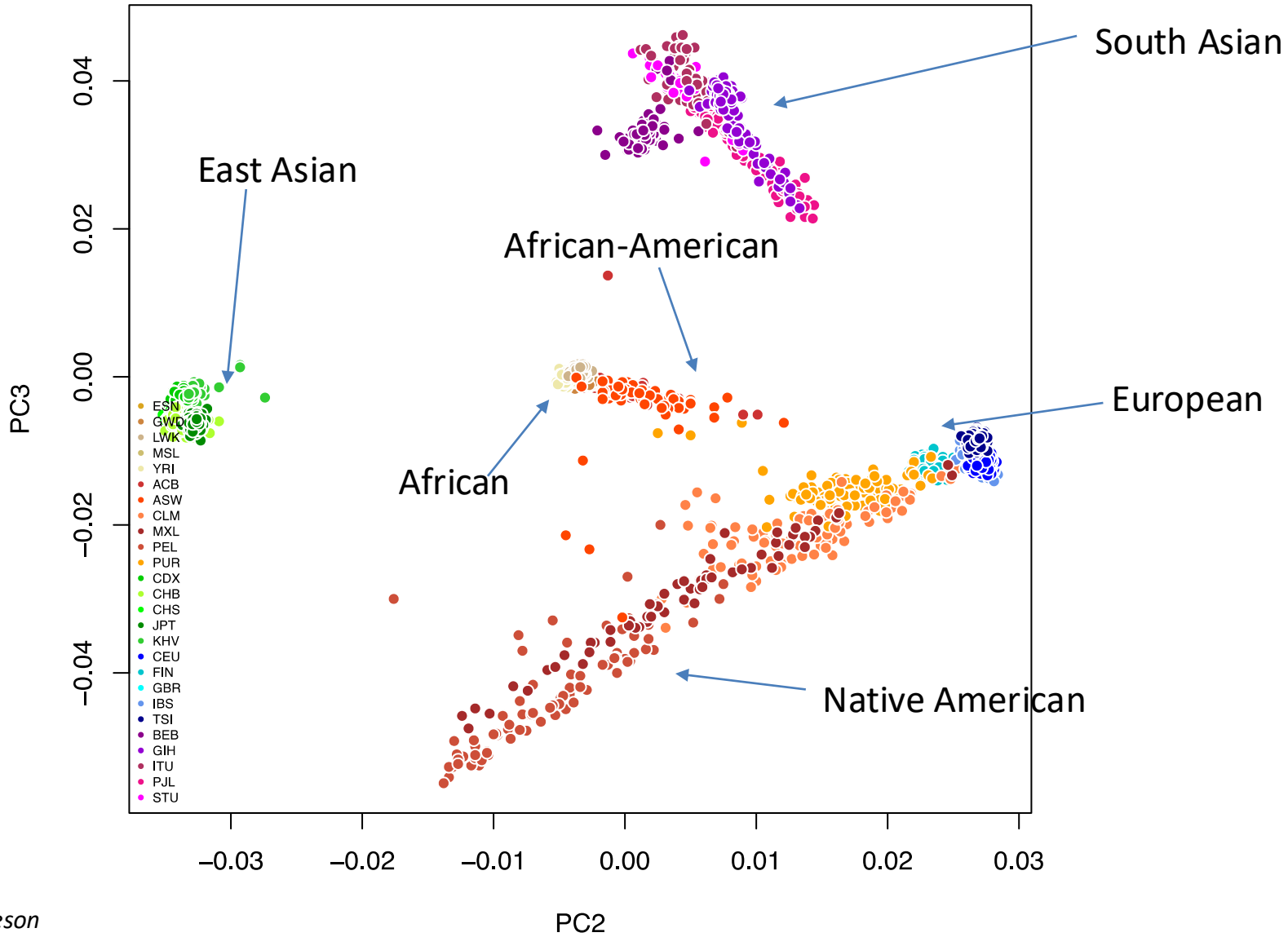
2. Admixture merges populations





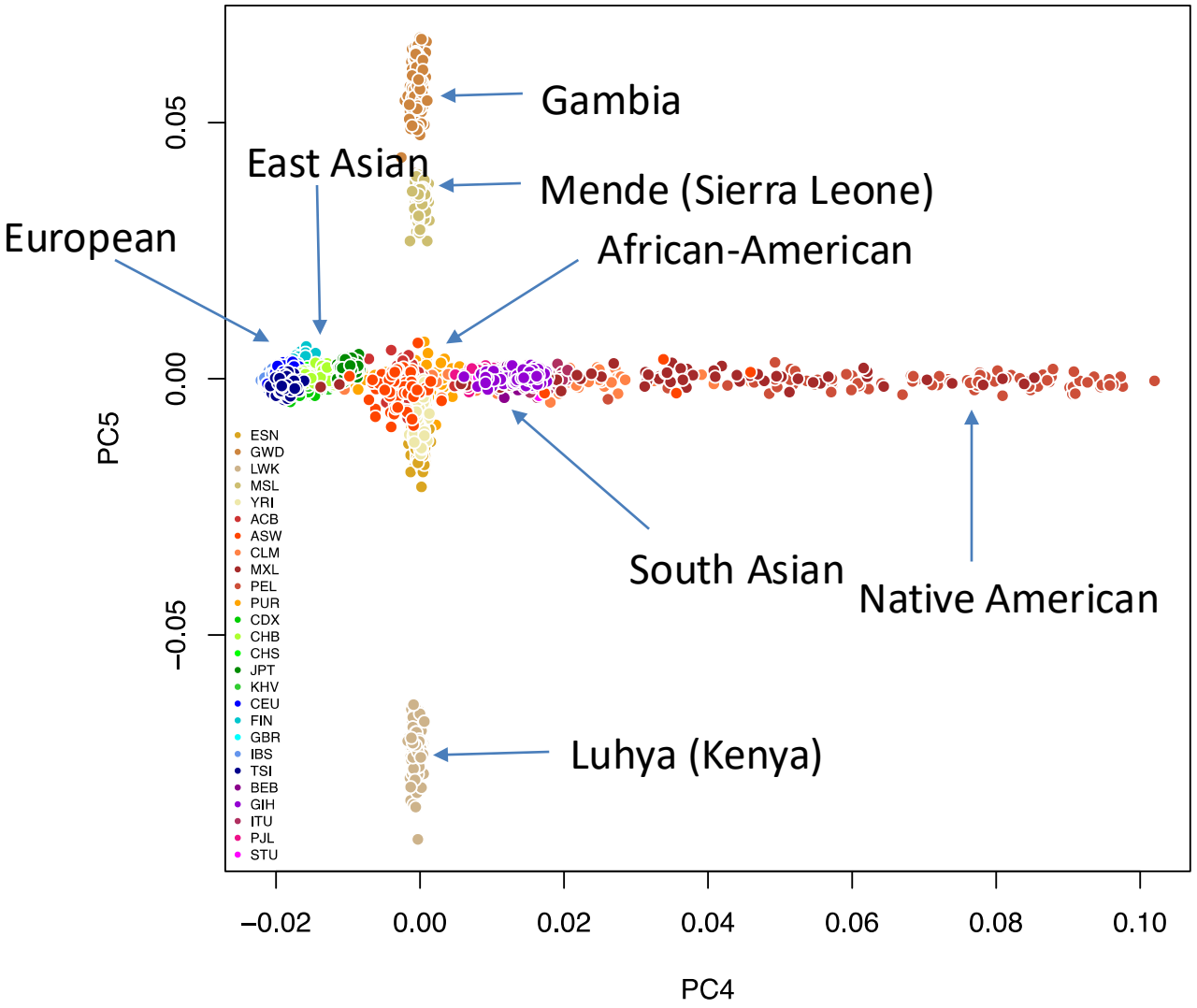
Slide: Iain Mathieson

Global population structure



Slide: Iain Mathieson

Global population structure

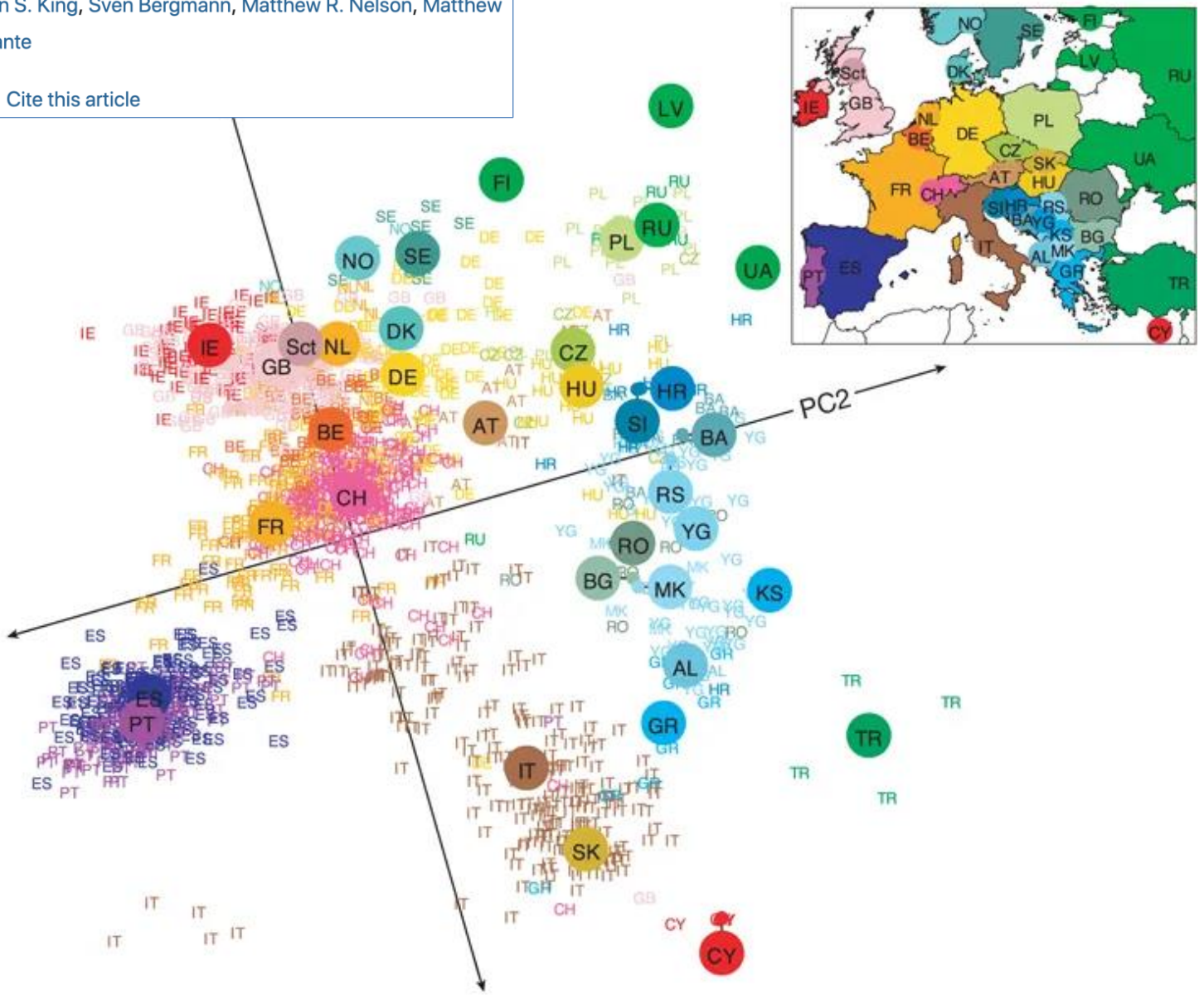


Slide: Iain Mathieson

Genes mirror geography within Europe

John Novembre ✉, Toby Johnson, Katarzyna Bryc, Zoltán Kutalik, Adam R. Boyko, Adam Auton, Amit Indap, Karen S. King, Sven Bergmann, Matthew R. Nelson, Matthew Stephens & Carlos D. Bustamante

Nature **456**, 98–101(2008) | Cite this article

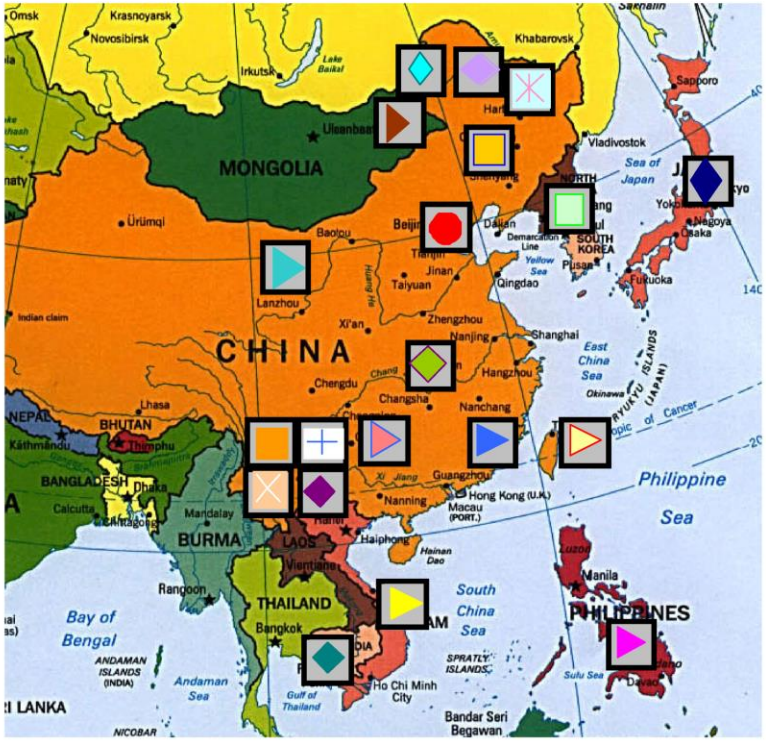


Analysis of East Asia Genetic Substructure Using Genome-Wide SNP Arrays

Chao Tian, Roman Kosoy, Annette Lee, Michael Ransom, John W. Belmont, Peter K. Gregersen, Michael F. Seldin

Published: December 5, 2008 • https://doi.org/10.1371/journal.pone.0003862

C



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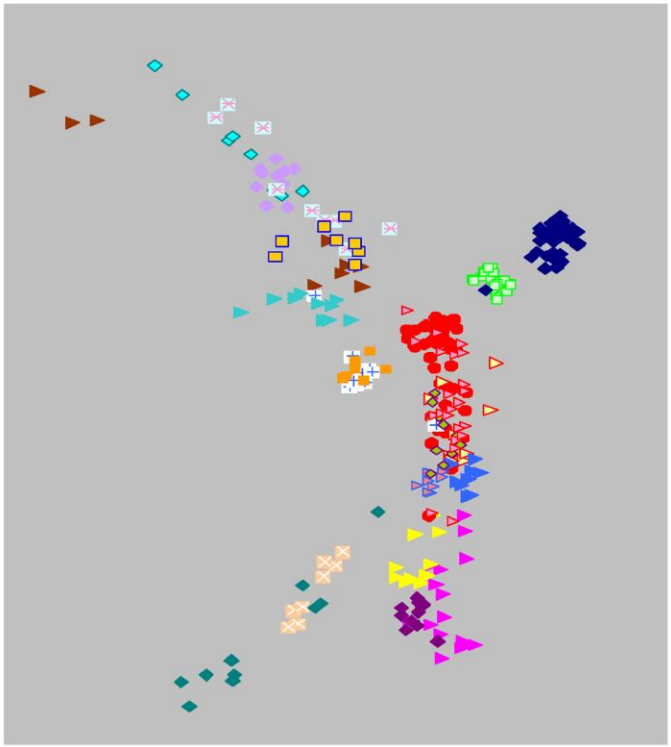

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XIBO
CHA
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YAK

Article | Published: 25 August 2010

A genomic analysis identifies a novel component in the genetic structure of sub-Saharan African populations

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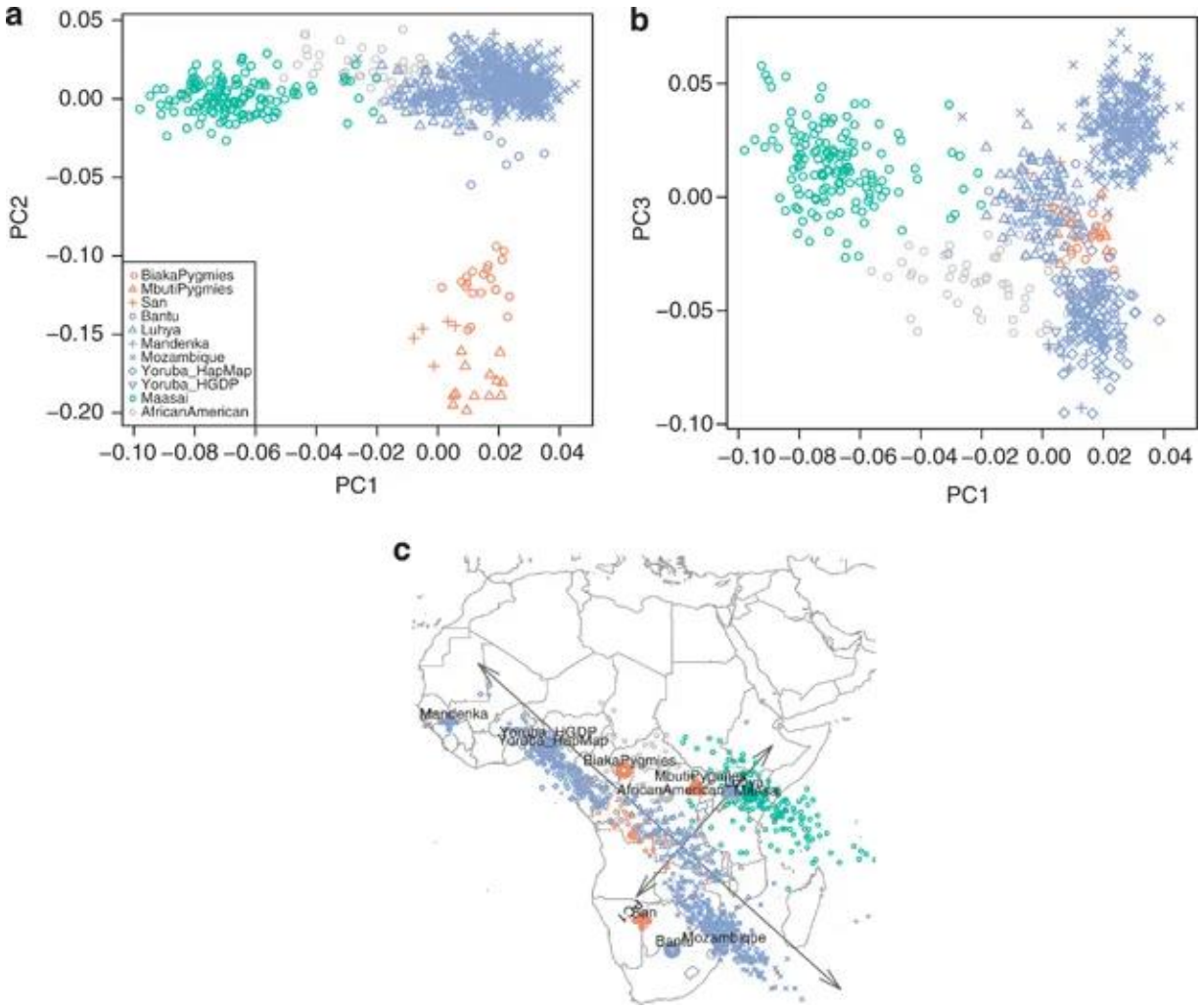


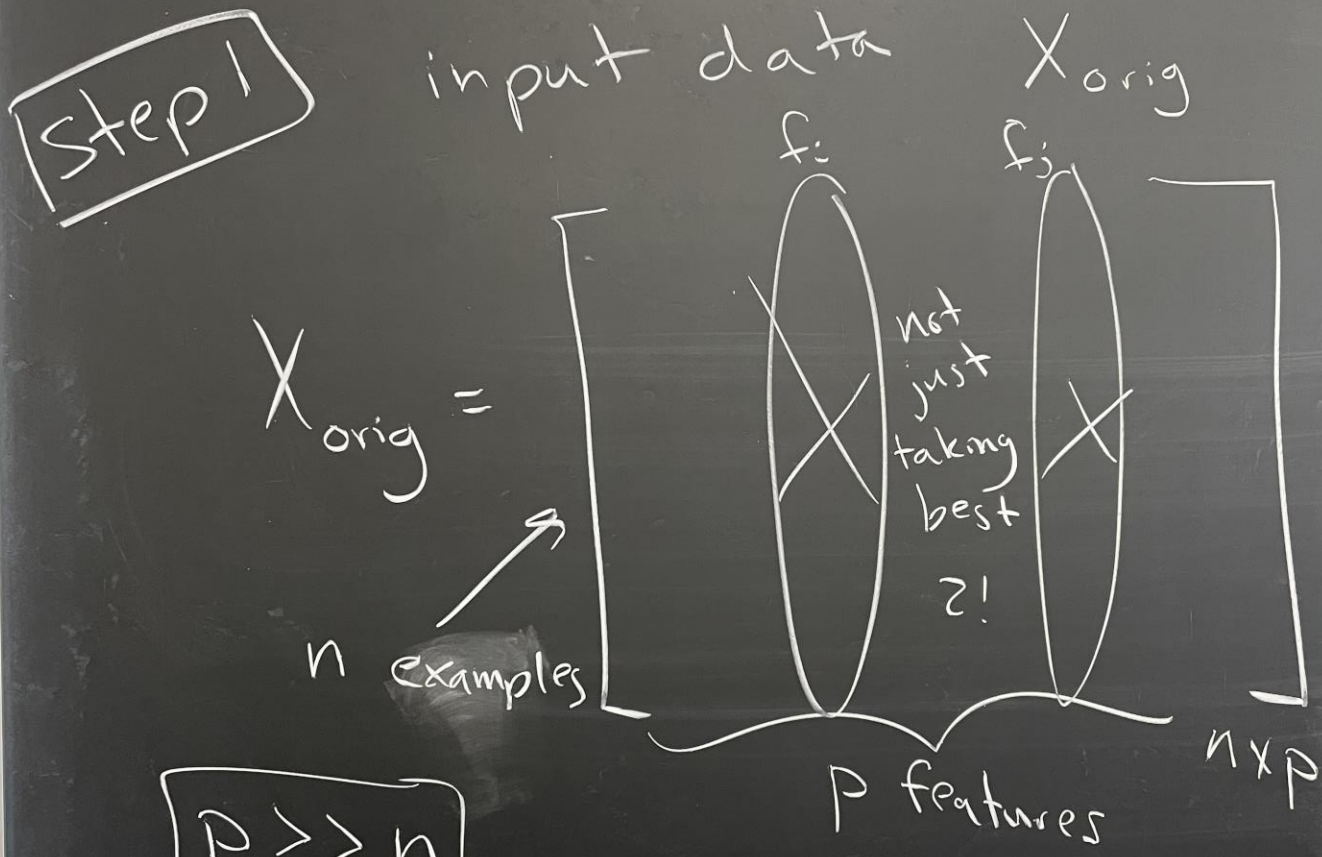
Outline

- Finish data visualization intro
- Dimensionality reduction
- PCA for data visualization

Principal Component Analysis

Step 1

input data X_{orig}



$p >> n$

sometimes!

(PCA)

goal: create $n \times r$ matrix for visualization

$r = 2$ (often)

no label! (i.e. $\vec{y}$)

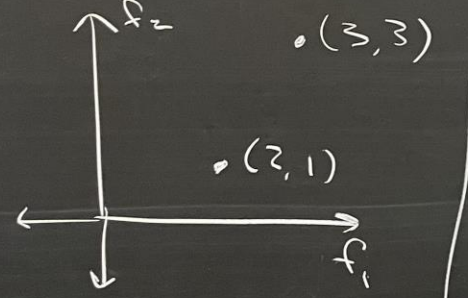
unsupervised

Step 2 Subtract of column-wise mean

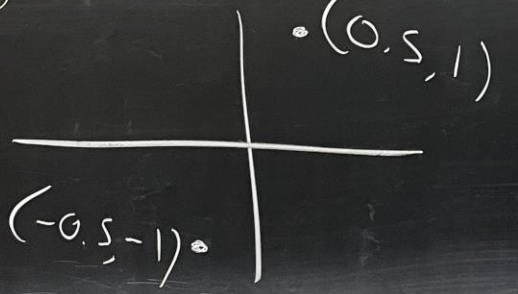
$$X_{orig} = \begin{matrix} f_1 & f_2 \\ \end{matrix} \begin{bmatrix} 2 & 1 \\ 3 & 3 \end{bmatrix}$$

$\bar{f}_1 = 2.5$

$\bar{f}_2 = 2$



$$X = \begin{bmatrix} -0.5 & -1 \\ 0.5 & 1 \end{bmatrix}$$



Step 3 compute covariance matrix A

$$cov(f,g) = \frac{1}{n-1} \sum_{i=1}^{n} (f_i - \bar{f})(g_i - \bar{g})$$

$$cov(f,f) = var(f) = \frac{1}{n-1} \sum_{i=1}^{n} (f_i - \bar{f})^2$$

$(n-1$ to make unbiased$)$

for 2 features

$A^T = A$ Symmetric

$$A = \begin{bmatrix} cov(f,f) & cov(f,g) \\ cov(g,f) & cov(g,g) \end{bmatrix}_{p \times p}$$

Step 4 Compute eigenvalues & eigenvectors of A

$$A\vec{v} = \lambda\vec{v} \Rightarrow \det(A - \lambda I) = 0$$

Solve for λ

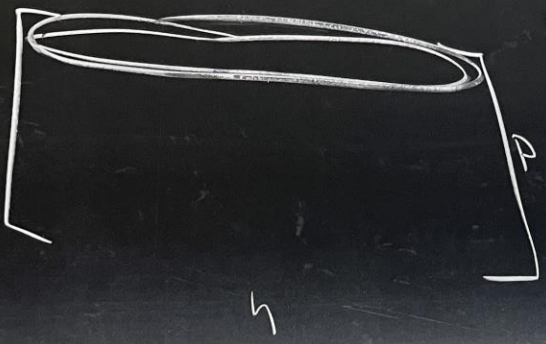
plug back in

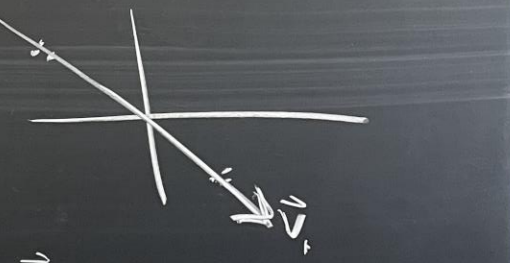
identity

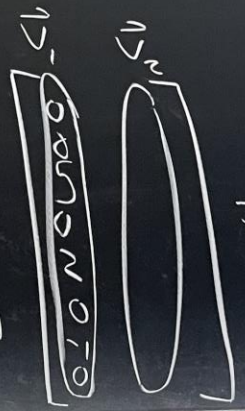
$$I = \begin{bmatrix} 1 & 0 \\ 0 & 1 \end{bmatrix}$$

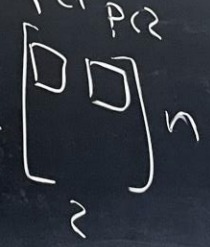
$p = 2$

$$\det\begin{bmatrix} a & b \\ c & d \end{bmatrix} = ad - bc$$









Step 5

$$W = \begin{bmatrix} | & | & & | \\ \vec{v}_1 & \vec{v}_2 & \cdots & \vec{v}_r \\ | & | & & | \end{bmatrix}$$

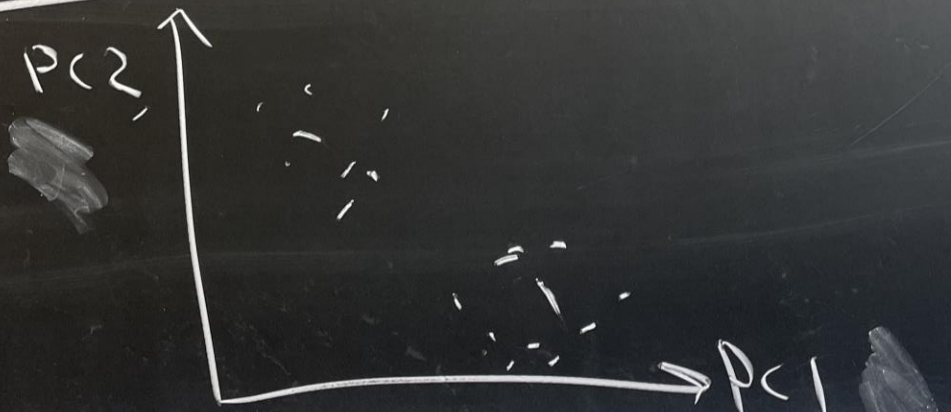
matrix of eigenvectors

$\lambda_1 \quad \lambda_2 \cdots \lambda_r$

$$T_{nxr} = X_{nxp} W_{pxr}$$

transformed data

Step 6 plot to visualize!



Handout 16

Step 1

$$X_{orig} = \begin{bmatrix} 0 & 1 \\ 0 & 1 \\ 0 & 1 \\ 1 & 0 \\ 1 & 0 \\ 1 & 0 \end{bmatrix} \quad \begin{matrix} \\ \\ \\ \\ \\ (f_1) \end{matrix} \quad \begin{matrix} \\ \\ \\ \\ \\ (f_2) \end{matrix}$$

$\bar{f}_{1,orig} = \frac{1}{2}$, $\bar{f}_{2,orig} = \frac{1}{2}$

Step 2

$$X = \begin{bmatrix} -1/2 & 1/2 \\ -1/2 & 1/2 \\ -1/2 & 1/2 \\ 1/2 & -1/2 \\ 1/2 & -1/2 \\ 1/2 & -1/2 \end{bmatrix}$$

$\bar{f}_1 = 0$, $\bar{f}_2 = 0$

Step 3

$$\text{var}(f_1) = \frac{1}{6-1}\left(\left(-\tfrac{1}{2}\right)\left(-\tfrac{1}{2}\right)\cdot 6\right) = \frac{1}{5}\cdot\frac{1}{4}\cdot 6$$

$$= \frac{3}{10}$$

$$\text{cov}(f_1, f_2) = \frac{1}{5}\left(-\tfrac{1}{2}\right)\left(\tfrac{1}{2}\right)\cdot 6 = \frac{-3}{10}$$

$$A = \begin{bmatrix} 3/10 & -3/10 \\ -3/10 & 3/10 \end{bmatrix}$$

Step 4

$$\det\left(\begin{bmatrix} 3/10 & -3/10 \\ -3/10 & 3/10 \end{bmatrix} - \underbrace{\begin{bmatrix} \lambda & 0 \\ 0 & \lambda \end{bmatrix}}_{\lambda I}\right) = 0$$

$$\det\begin{bmatrix} 3/10 - \lambda & -3/10 \\ -3/10 & 3/10 - \lambda \end{bmatrix} = 0$$

$$\left(\frac{3}{10} - \lambda\right)^2 - \left(-\frac{3}{10}\right)^2 = 0$$

$$\cancel{\left(\frac{3}{10}\right)^2} - 2\cdot\frac{3}{10}\lambda + \lambda^2 - \cancel{\left(\frac{3}{10}\right)^2} = 0$$

$$\lambda^2 - \frac{3}{5}\lambda = 0$$

$$\lambda\left(\lambda - \frac{3}{5}\right) = 0$$

Sort by high → low magnitude

$$\lambda_1 = \frac{3}{5},\ \lambda_2 = 0$$

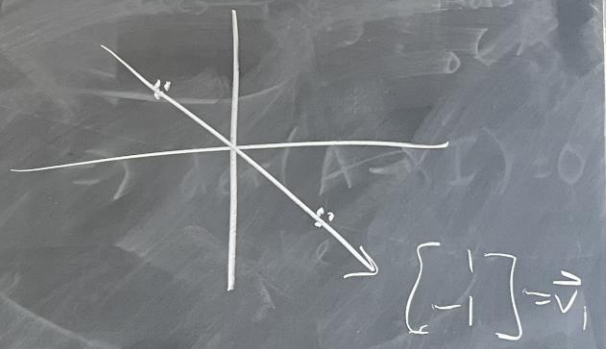
$$A\vec{v} = \lambda\vec{v}$$

$W = \begin{bmatrix} \vec{v}_1 & \vec{v}_2 \\ 1 & 1 \\ -1 & 1 \end{bmatrix}$ matrix

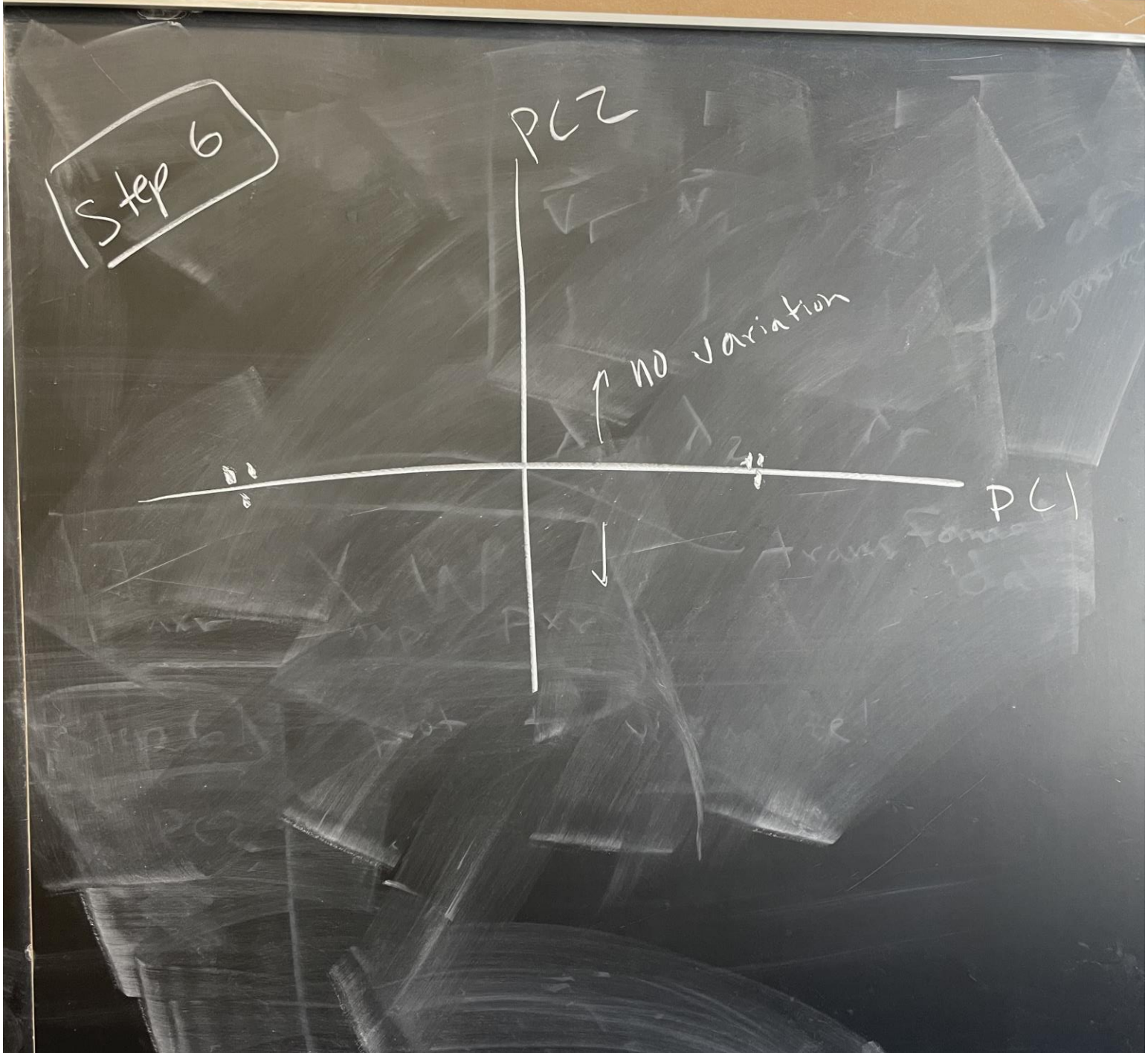
$\lambda_1 = \frac{3}{5}$ $\lambda_2 = 0$

$T_2 = X W_2 = \begin{bmatrix} -\frac{1}{2} & \frac{1}{2} \\ \vdots & \vdots \\ \frac{1}{2} & -\frac{1}{2} \\ \vdots & \vdots \end{bmatrix}$ dot product $\begin{bmatrix} 1 & 1 \\ -1 & 1 \end{bmatrix} = \begin{bmatrix} -1 & 0 \\ -1 & 0 \\ -1 & 0 \\ 1 & 0 \\ 1 & 0 \\ 1 & 0 \end{bmatrix}$

$P(1)$ $P(2)$



$\begin{bmatrix} 1 \\ -1 \end{bmatrix} = \vec{v}_1$


Step 6
PC2
↑ no variation
PC1
↓