# CS 260: Foundations of Data Science

Prof. Thao Nguyen

Fall 2024

HAVERFORD
COLLEGE

Materials by Sara Mathieson

# Admin

- **Lab 5** grades & feedback posted on Moodle

- I will be away for a conference Nov 5-10
  - **No lab** on Nov 5
  - Prof. Mathieson will teach **Nov 6 lecture**
  - Will check my email but responses can be delayed

# CS@Haverford Fall Fling

- Tuesday Nov 12, 11:30am-1:30pm ET

# Outline for today

- Dimensionality reduction
- PCA for data visualization

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- Dimensionality reduction

- PCA for data visualization

# Principal Component Analysis (PCA)

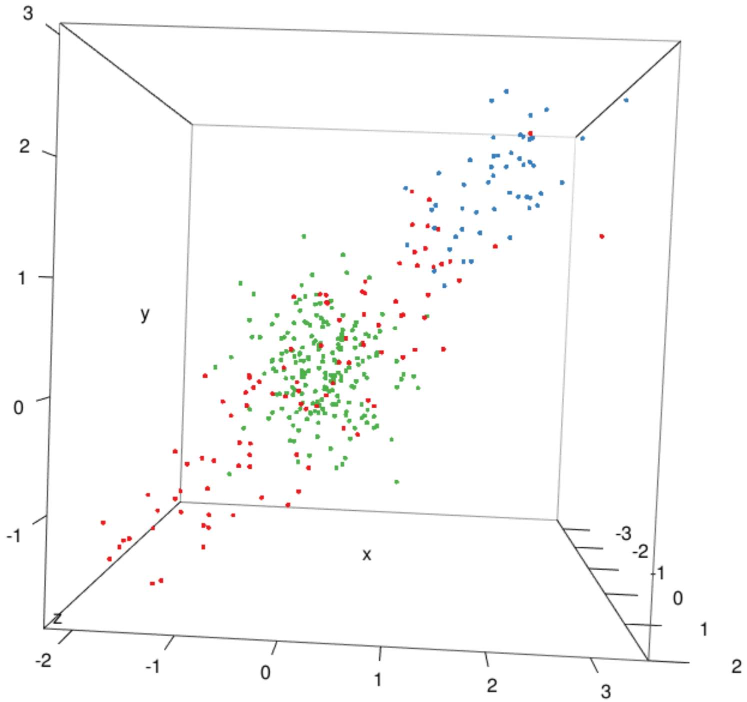
- Transforms *p*-dimensional data so that the new first dimension explains as much of the variation as possible, the new second explains as much of the remaining variation as possible, and so on

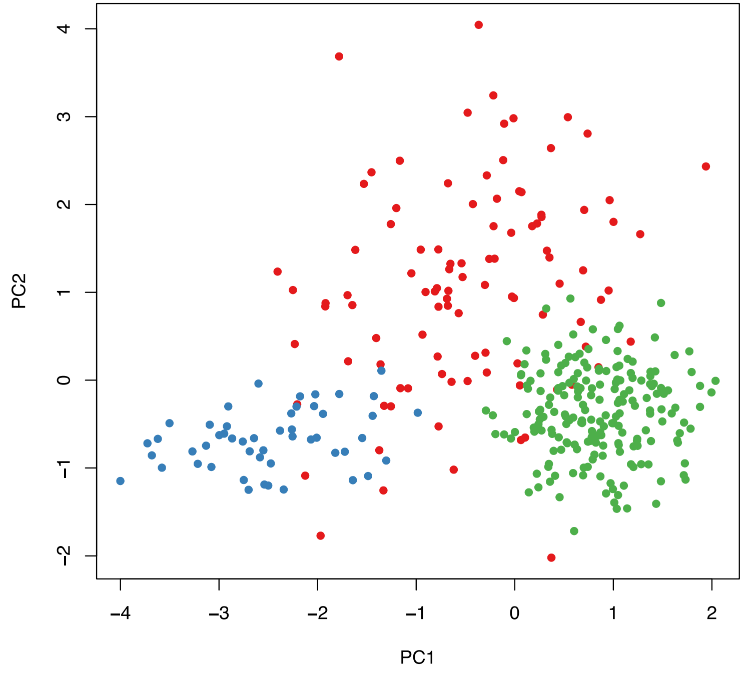
- PCA is a linear transformation

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

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

# PCA Example

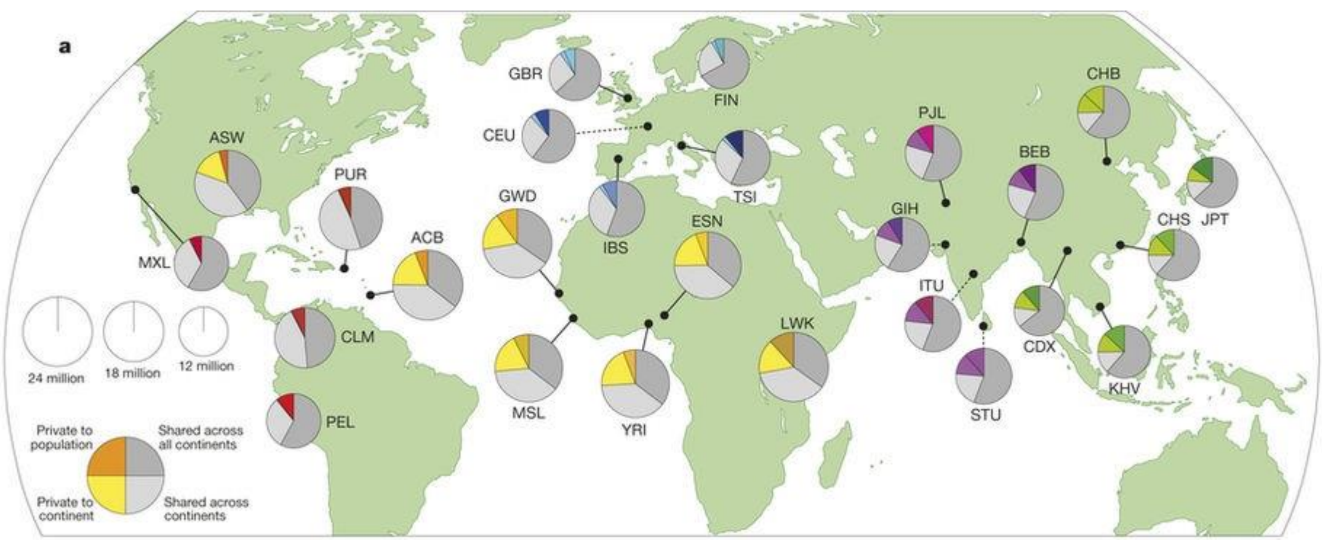



*Slide: Iain Mathieson*

# 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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#CHROM  POS     ID      REF     ALT     QUAL    FILTER  INFO    FORMAT  HG00096 HG00097 HG00099 HG00100 HG00101 HG00102 HG00103 HG00105 HG00106 HG00107 HG00108 HG00109 HG00110 HG00111
20      60343   .       G       A       100     PASS    .       GT      0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0
20      60419   .       A       G       100     PASS    .       GT      0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0
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20      60522   rs150241001     T       TC      100     PASS    .       GT      0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0
20      60568   .       A       C       100     PASS    .       GT      0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0
20      60571   rs116145529     C       A       100     PASS    .       GT      0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0
20      60579   .       G       A       100     PASS    .       GT      0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0
20      60649   .       A       G       100     PASS    .       GT      0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0
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20      60795   rs184056664     G       C       100     PASS    .       GT      0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0
20      60808   .       G       A       100     PASS    .       GT      0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0
20      60810   .       G       GA      100     PASS    .       GT      0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0
20      60826   .       A       G       100     PASS    .       GT      0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0
20      60828   rs187713677     T       G       100     PASS    .       GT      0|0     0|1     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0
20      60864   .       G       A       100     PASS    .       GT      0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0
20      60895   .       A       G       100     PASS    .       GT      0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0
20      60916   .       G       T       100     PASS    .       GT      0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0
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20      61070   .       C       T       100     PASS    .       GT      0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0
20      61098   rs6078030       C       T       100     PASS    .       GT      0|0     0|0     0|0     0|0     0|1     0|0     1|0     0|0     0|0     0|0     0|0     0|1     0|0
20      61118   .       A       G       100     PASS    .       GT      0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0
20      61138   rs140305189     C       CT      100     PASS    .       GT      0|0     0|1     0|0     0|0     0|1     0|0     0|0     0|1     0|0     0|0     0|0     0|0     0|0
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20      61329   rs182162684     C       T       100     PASS    .       GT      0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0
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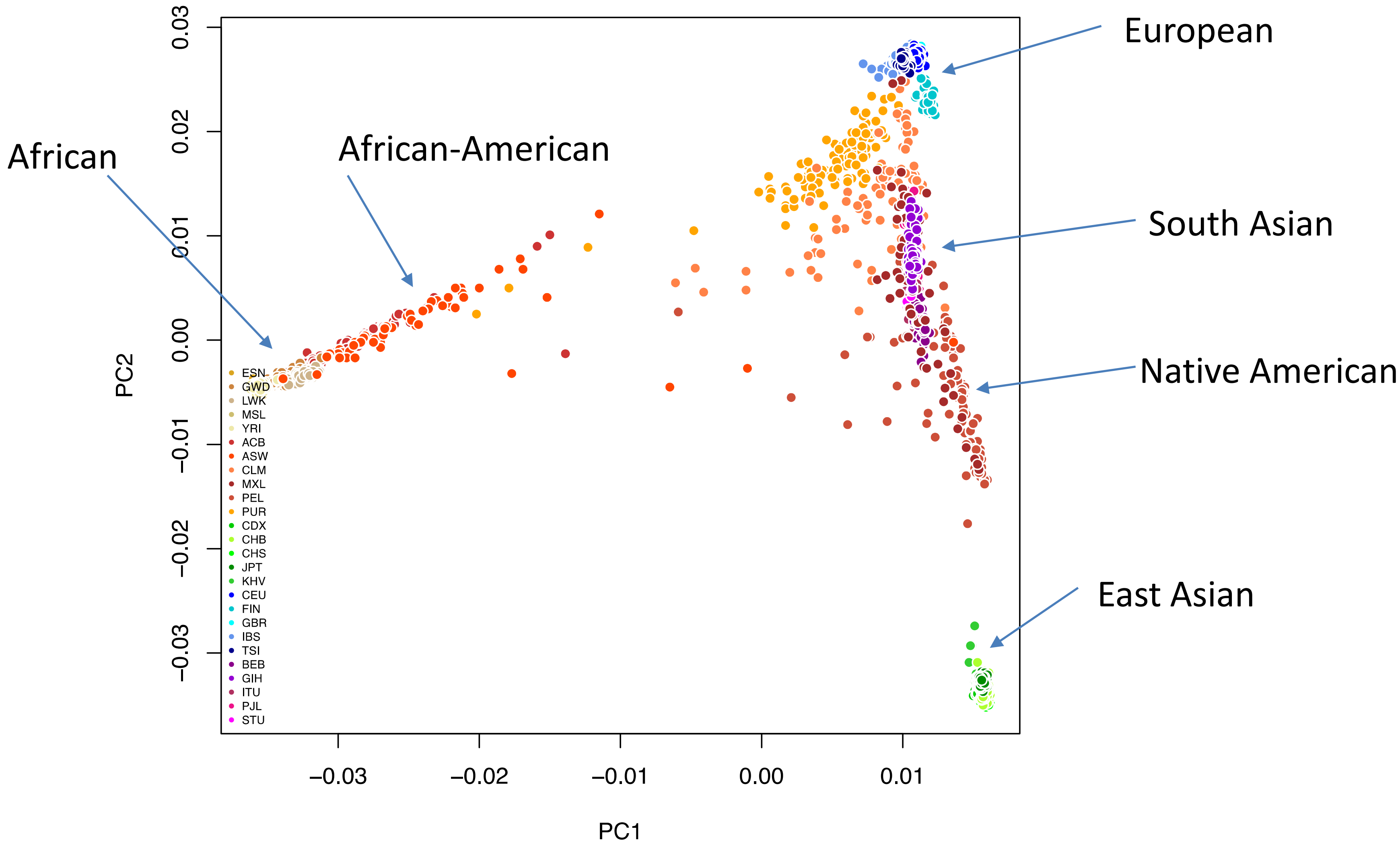
20      61409   rs139103017     A       G       100     PASS    .       GT      0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0
20      61437   .       A       C       100     PASS    .       GT      0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0
20      61450   .       T       C       100     PASS    .       GT      0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0
20      61517   rs187280035     C       T       100     PASS    .       GT      0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0
20      61538   .       A       G       100     PASS    .       GT      0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0
20      61638   .       C       T       100     PASS    .       GT      0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0
20      61651   rs76553454      C       A       100     PASS    .       GT      0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0
20      61711   rs369824431     G       T       100     PASS    .       GT      0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0
20      61724   rs142532139     A       C       100     PASS    .       GT      0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0
20      61795   rs4814683       G       T       100     PASS    .       GT      1|0     0|0     0|0     0|0     0|1     0|0     1|0     0|1     1|0     0|0     0|0     0|1     0|0
20      61955   .       C       T       100     PASS    .       GT      0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0
20      61972   .       T       C       100     PASS    .       GT      0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0
20      62100   rs6047235       T       C       100     PASS    .       GT      0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0
20      62174   .       AGATCAGTCCTTT   A       100     PASS    .       GT      0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0
20      62255   rs192879424     T       C       100     PASS    .       GT      0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0
20      62283   .       T       C       100     PASS    .       GT      0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0
20      62348   rs141113228     A       G       100     PASS    .       GT      0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0
20      62387   .       T       A       100     PASS    .       GT      0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0
20      62420   rs185326153     A       G       100     PASS    .       GT      0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0
20      62461   .       C       T       100     PASS    .       GT      0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0
20      62471   rs188652106     G       A       100     PASS    .       GT      0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0
20      62478   rs192812899     A       G       100     PASS    .       GT      0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0
20      62545   rs150267191     C       G       100     PASS    .       GT      0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0
20      62553   rs114190700     T       C       100     PASS    .       GT      0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0     0|0
```

# Global population structure

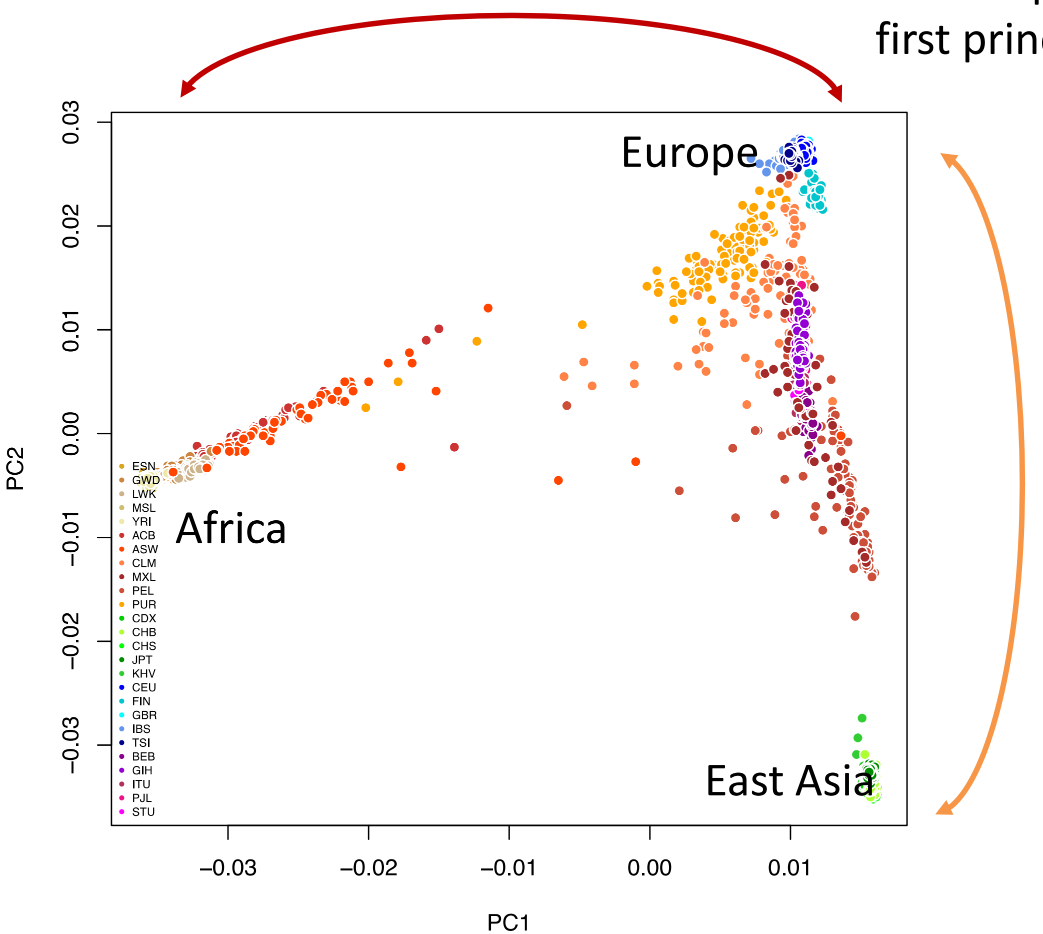


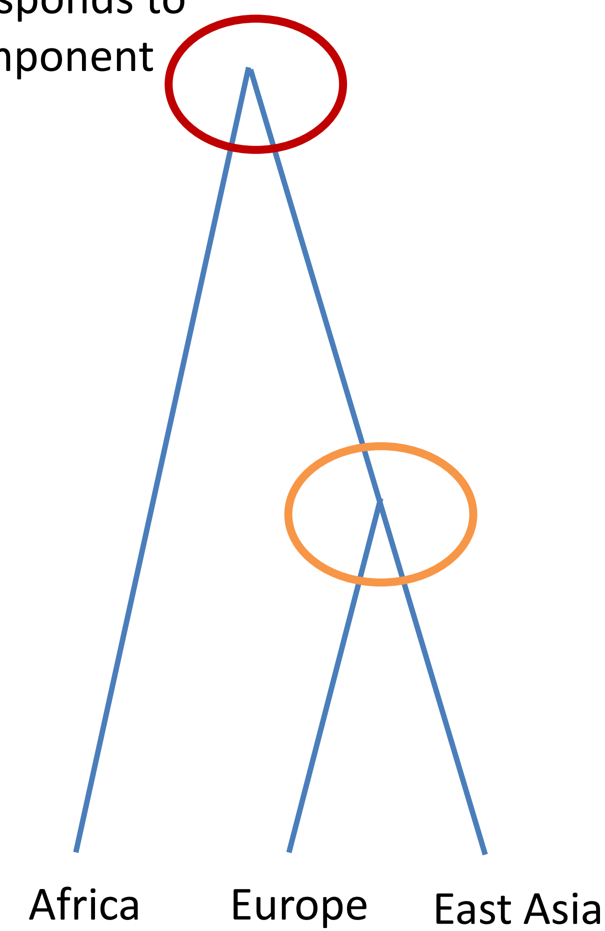

*Slide: Iain Mathieson*

# What causes these patterns?

## 1. Populations splits separate populations

Oldest split corresponds to first principal component



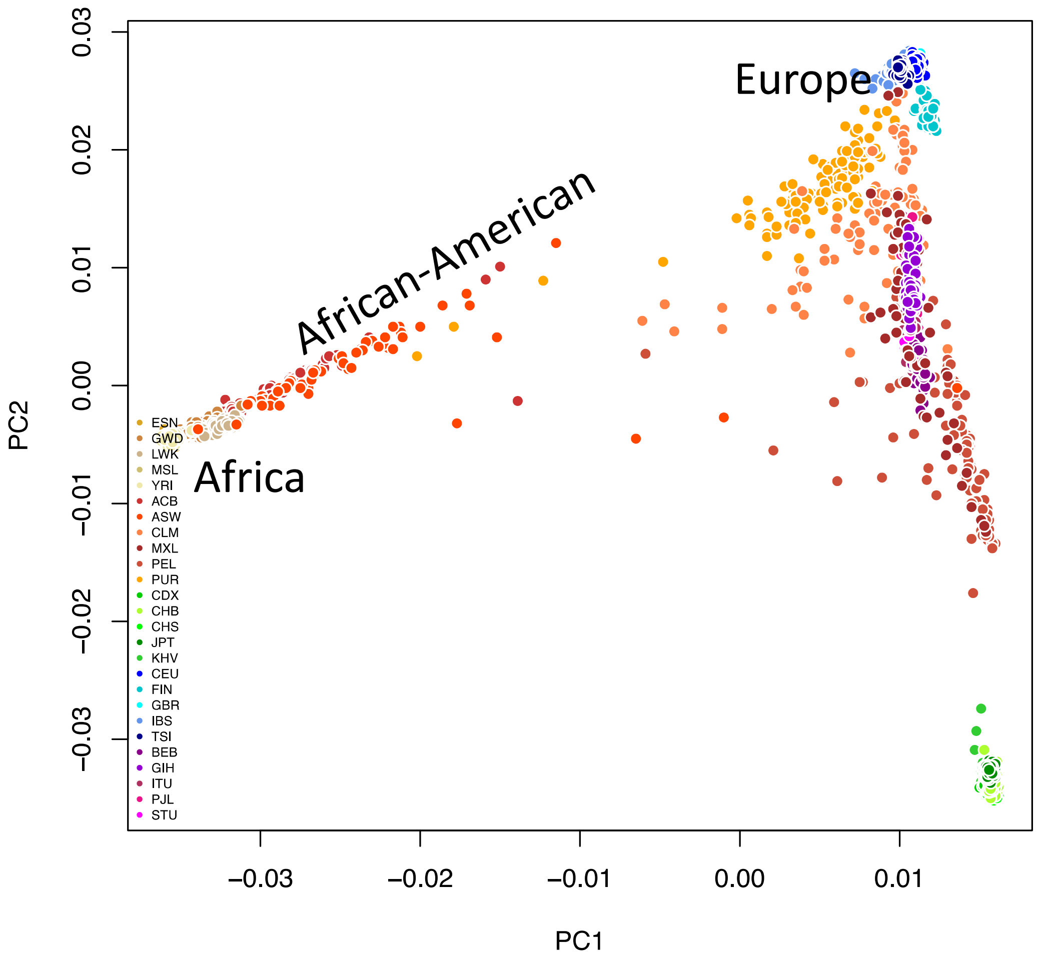



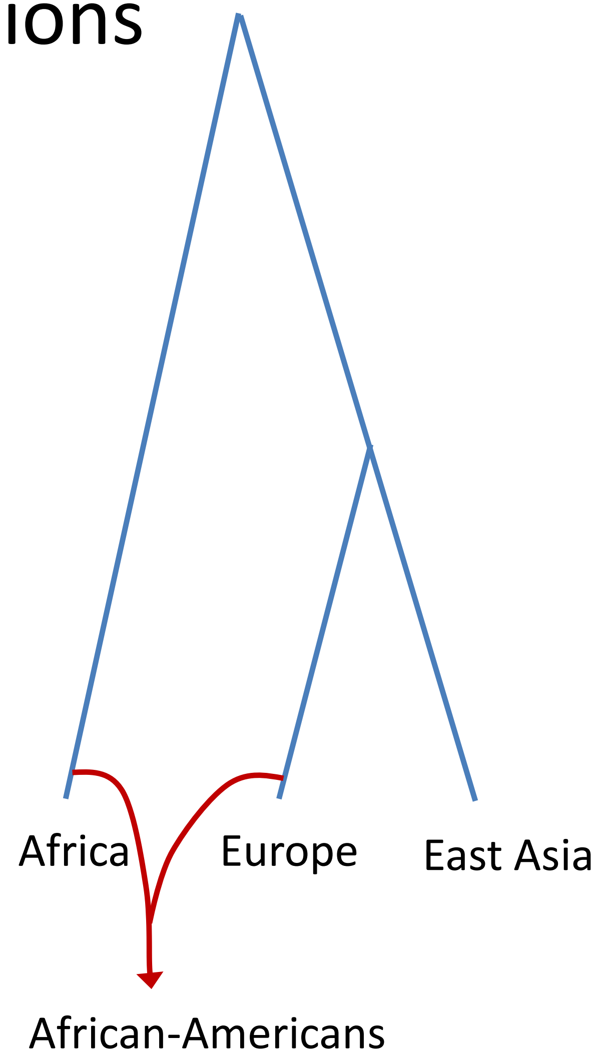

*Slide: Iain Mathieson*

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

# What causes these patterns?

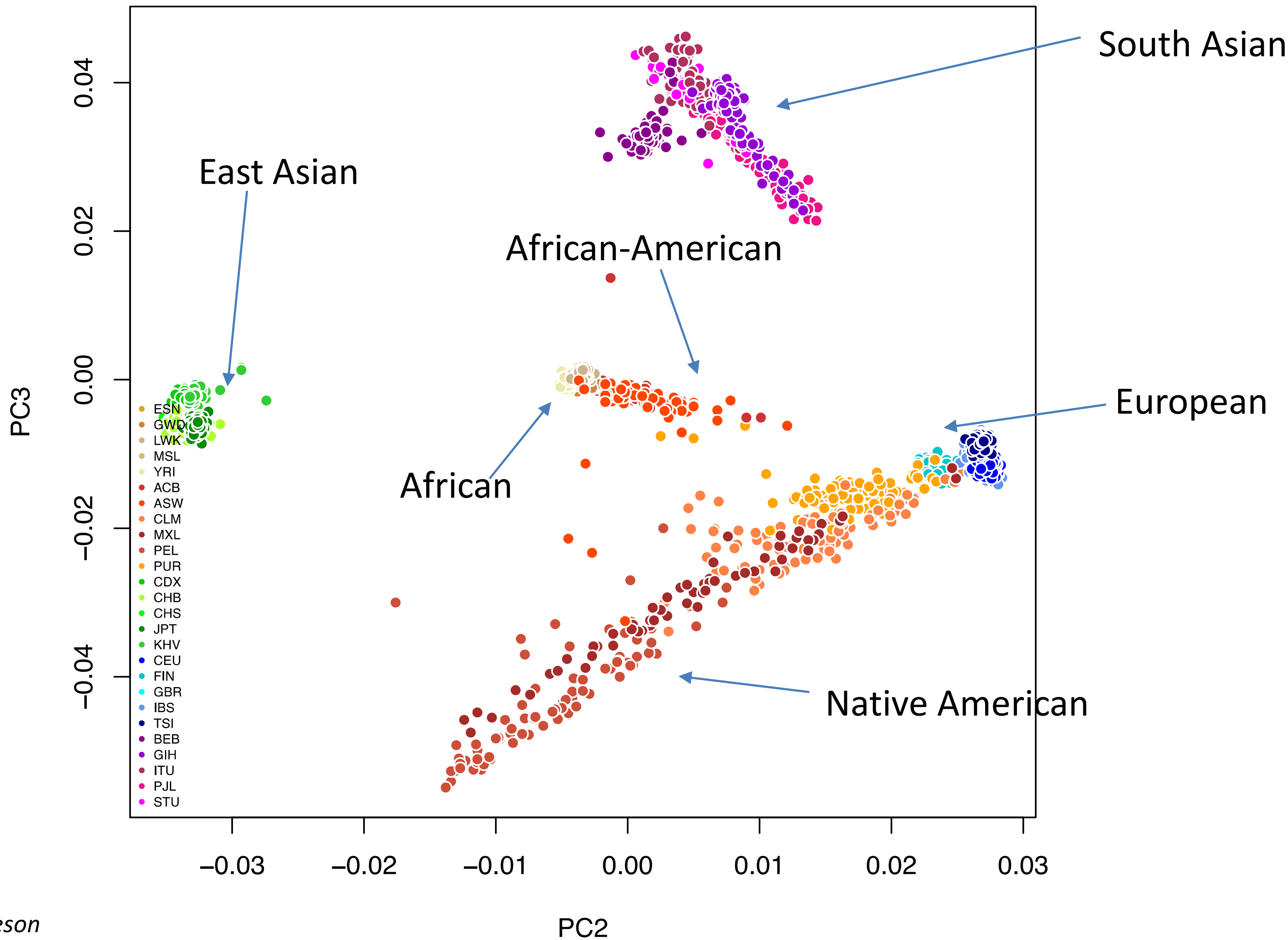
## 2. Admixture merges populations




*Slide: Iain Mathieson*

# Global population structure

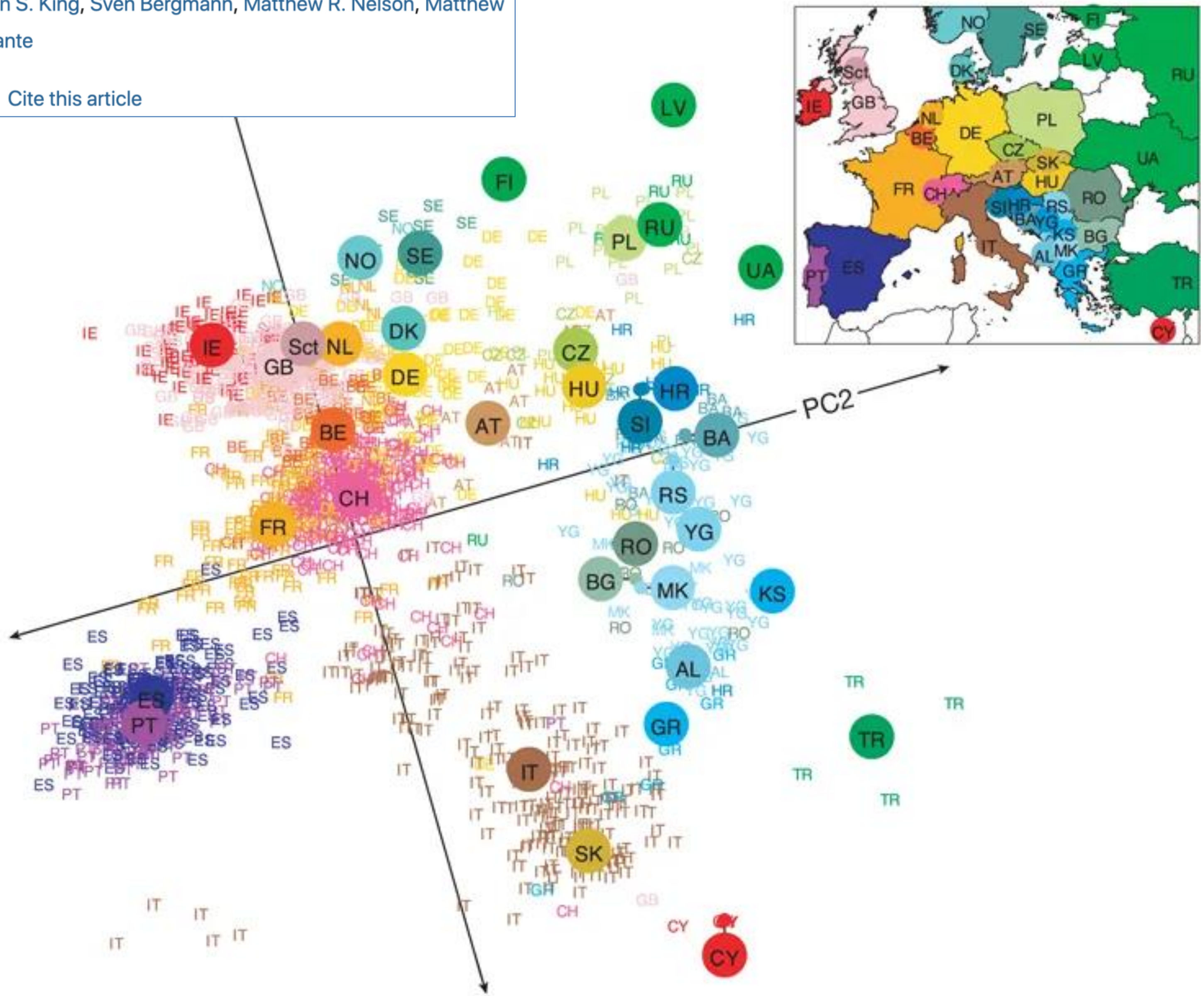



*Slide: Iain Mathieson*

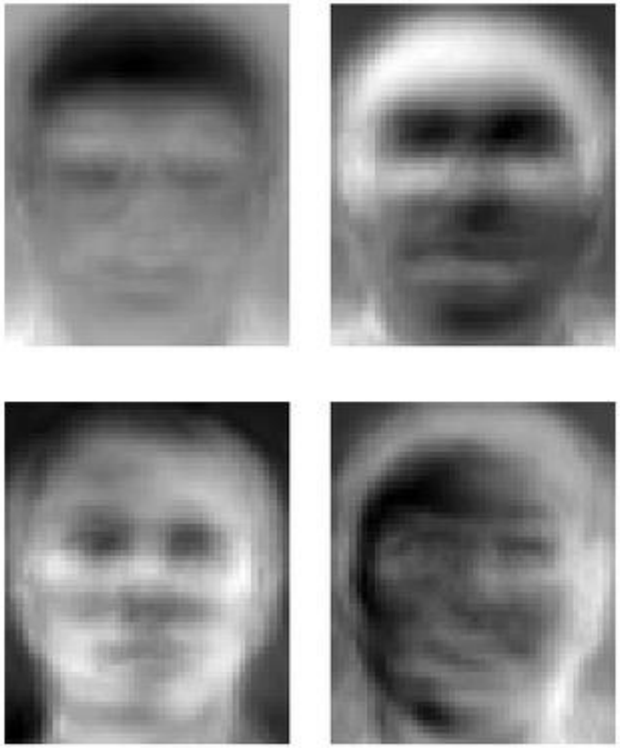
# Genes mirror geography within Europe

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



# PCA application: Eigenfaces



*Wikipedia*

- Low-dimensional representation of face images
- Used for face recognition/classification

# Outline for today

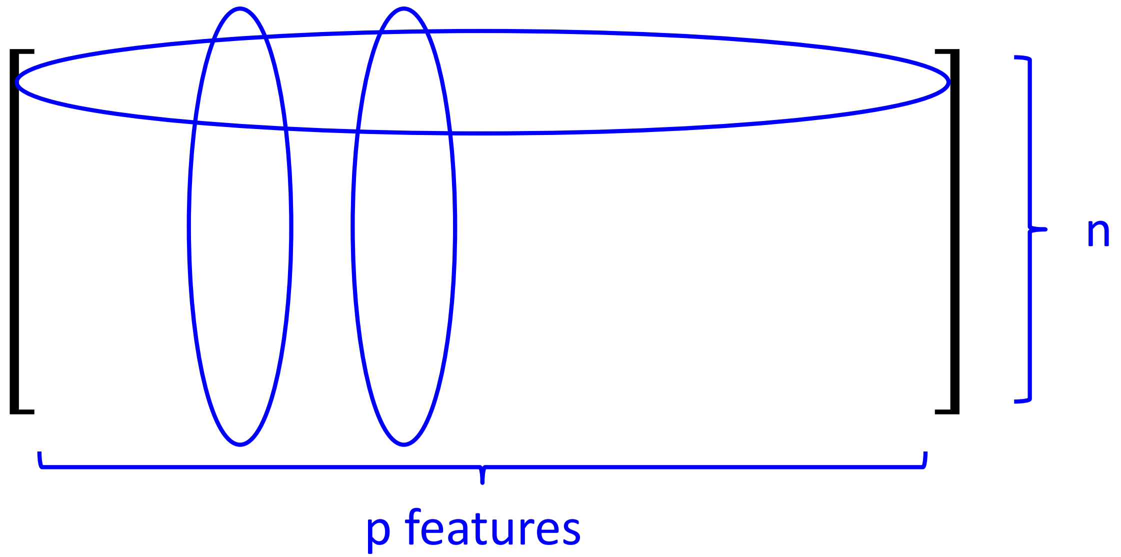
- Dimensionality reduction

- PCA for data visualization

# PCA Algorithm

<u>Step 1:</u>

$X_{orig} =$




p >> n

**Goal:** Create nx2 matrix for visualization

# PCA Algorithm

Step 2: Subtract off column-wise mean

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

$\overline{x_1} = 2.5$ $\overline{x_2} = 2$

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

# PCA Algorithm

<u>Step 3:</u> Compute covariance matrix A

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

2 features f, g

$A$ ↓ square & symmetric

Runtime $O(np^2)$

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

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

# PCA Algorithm

Step 4: Compute eigenvalues and eigenvectors of A

eigenvalue

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

eigenvector

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

Solve for $\lambda$ and plug into first equation to solve for $\vec{v}$

# PCA Algorithm

<u>Step 5:</u> Sort eigenvectors by eigenvalues (high->low)

$$W = \begin{bmatrix} \vdots & \vdots & & \vdots \\ \overrightarrow{v_1} & \overrightarrow{v_2} & \cdots & \overrightarrow{v_r} \\ \vdots & \vdots & & \vdots \end{bmatrix}$$

Column eigenvalues: $\lambda_1$, $\lambda_2$

first eigenvector

$pxr$

usually $r = 2$

And compute the transformed data:

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

# Handout 16

# Handout 16

**Step 1 & 2**

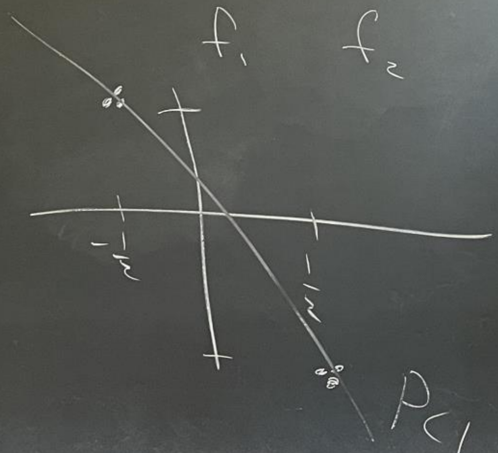
$$X = \begin{bmatrix} -1/2 & 1/2 \\ -1/2 & 1/2 \\ -1/2 & 1/2 \\ 1/2 & -1/2 \\ 1/2 & -1/2 \\ 1/2 & -1/2 \end{bmatrix}$$

$f_1 \quad f_2$

orig:

$\bar{f}_1 = \frac{1}{2}$

$\bar{f}_2 = \frac{1}{2}$




**Step 3**

$$A = \begin{bmatrix} var(f_1) & cov(f_1, f_2) \\ cov(f_2, f_1) & var(f_2) \end{bmatrix}$$

$\bar{f}_1 = 0$

$\bar{f}_2 = 0$

$$cov(f_1, f_2) = \frac{1}{6-1}\left(-\frac{1}{2}\cdot\frac{1}{2}\right)\cdot 6$$

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

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

Step 4

$$\det(A - \lambda I) = 0$$

$$\left(\begin{bmatrix} 3/10 & -3/10 \\ -3/10 & 3/10 \end{bmatrix} - \lambda \begin{bmatrix} 1 & 0 \\ 0 & 1 \end{bmatrix}\right) = 0$$

6

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

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

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

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

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

$$\lambda\left(\lambda - \frac{3}{5}\right) = 0 \Rightarrow \lambda_1 = \frac{3}{5}, \quad \lambda_2 = 0$$

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

$\lambda_1 = 3/5 \qquad \lambda_2 = 0$

$$T_2 = XW_2 = \begin{bmatrix} -1/2 & 1/2 \\ -1/2 & 1/2 \\ -1/2 & 1/2 \\ 1/2 & -1/2 \\ 1/2 & -1/2 \\ 1/2 & -1/2 \end{bmatrix} \begin{bmatrix} 1 & 1 \\ -1 & 1 \end{bmatrix} = \begin{bmatrix} -1 & 0 \\ -1 & 0 \\ -1 & 0 \\ 1 & 0 \\ 1 & 0 \\ 1 & 0 \end{bmatrix}$$

$\vec{v}_1 \qquad \vec{v}_2$