

Unsupervised learning

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Learning goals

- ▶ **Understand Unsupervised Learning:** Define unsupervised learning and its objectives.
- ▶ **Dimensionality Reduction (PCA):** Explain PCA for reducing dimensionality while preserving variance. Understand the meaning and interpretation of principal components and loading vectors.
- ▶ **K-means Clustering:** Understand k-means algorithm steps: initialisation, assignment, and update. Learn techniques to select the number of clusters.
- ▶ **Hierarchical Clustering:** Understand agglomerative clustering and how dendograms represent clusters. Compare similarity metrics (Euclidean distance, correlation) linkage methods (single, complete, average) and their impact.

Material

- ▶ Chapter 12 of “*An introduction to statistical learning with applications in python*, G. James, D. Witten, T. Hastie, R. Tibshirani, J. Taylor”

Overview

- ▶ Supervised vs unsupervised learning
- ▶ Dimensionality reduction
 - ▶ Principal component analysis (PCA)
- ▶ Clustering
 - ▶ K-means
 - ▶ Hierarchical clustering

Instructions

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Difference between supervised and unsupervised learning

With both supervised and unsupervised learning we have a set of features X_1, X_2, \dots, X_p for each observation:

- ▶ With **supervised learning** (or learning with a teacher) we also have an associated variable Y (a label).
- ▶ With **unsupervised learning** (or learning without a teacher) we don't have the label.

Unsupervised learning

The goal of unsupervised learning is to find similarities among observations based on the set of features.

There are two categories of methods:

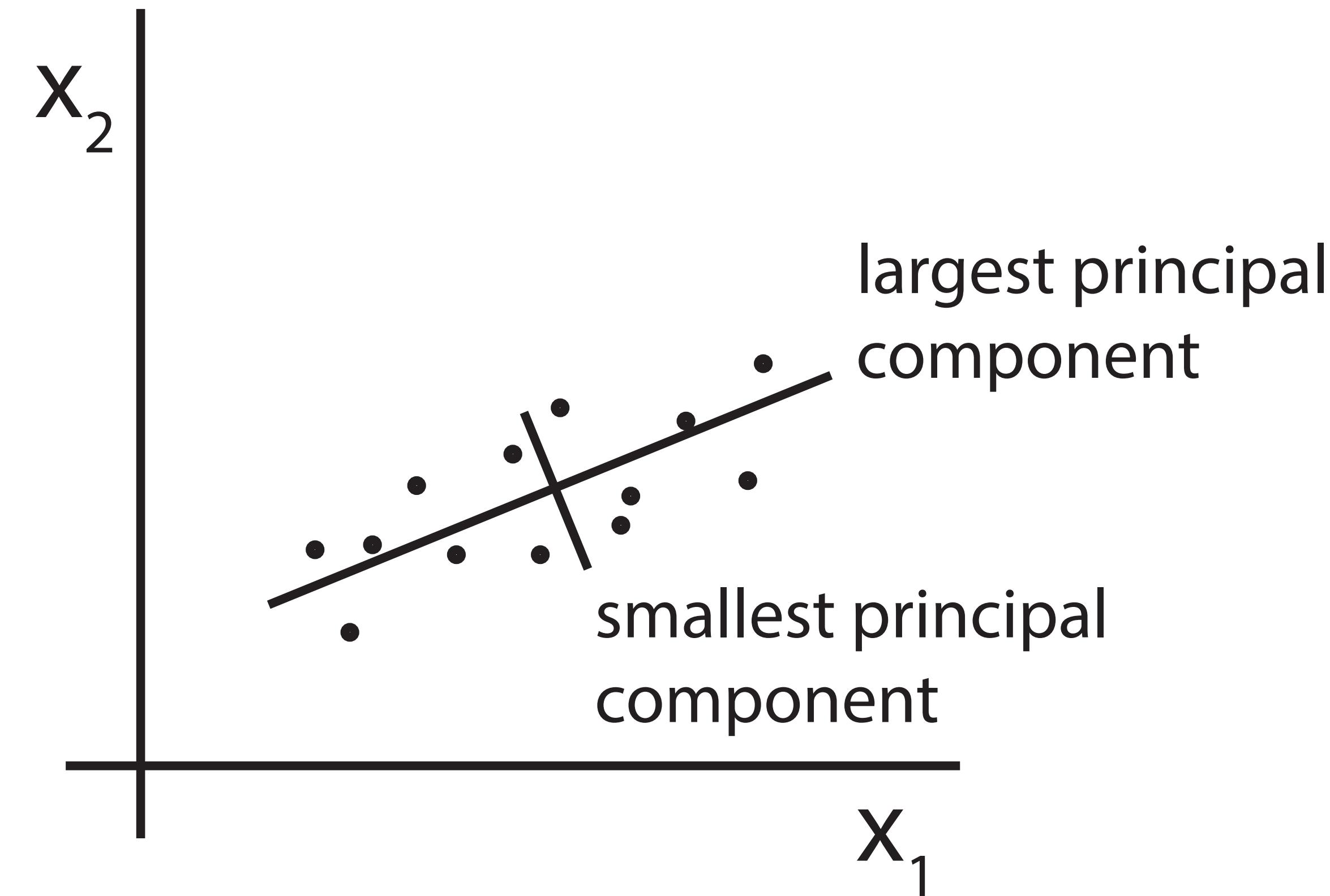
- ▶ **Dimensionality reduction:** methods to reduce the dimensions of the input features to facilitate visualisation and identification of groups.
- ▶ **Clustering:** methods to discover unknown groups (clusters) in data.

Use of unsupervised learning

- ▶ No need for labels (often difficult to retrieve).
- ▶ Used for exploratory data analysis or for preprocessing before applying supervised learning techniques.
- ▶ No quantitative metrics to measure success; evaluation based on heuristic arguments.

Principal component analysis (PCA)

Idea: from the p variables (often correlated), derive a smaller subset of variables that explain most of the variability of the original set.



Principal component analysis (PCA)

Use of PCA:

- ▶ For feature reduction: derive a smaller subset of variables (the principal components) that are a good representation of the original data. These can then be used for supervised learning problems.
- ▶ For data visualisation: be able to visualise data in a smaller dimension. This can be used for exploratory data analysis or to identify groups of observations with similar characteristics.

First principal component

Starting from a set of features X_1, X_2, \dots, X_p , the **first principal component** is the normalised linear combination of the features:

$$Z_1 = \phi_{11}X_1 + \phi_{21}X_2 + \dots + \phi_{p1}X_p$$

Where:

- ▶ $\sum_{j=1}^p \phi_{j1}^2 = 1$
- ▶ $\phi_{11}, \phi_{21}, \dots, \phi_{p1}$ are the **loadings** of the first principal component
- ▶ $\phi_1 = (\phi_{11}, \phi_{21}, \dots, \phi_{p1})^T$ is the **first principal component loading vector**

Computing the first principal component

Consider a dataset with n observations x_1, x_2, \dots, x_n , where each $x_i = (x_{i1}, x_{i2}, \dots, x_{ip})^T$, or in matrix form \mathbf{X} of size $n \times p$.

Since we are only interested in the variance, we assume that each feature has 0 mean, i.e. the column means of \mathbf{X} are 0.

We want to find the linear combination of the sample feature value:

$$z_{i1} = \phi_{11}x_{i1} + \phi_{21}x_{i2} + \dots + \phi_{p1}x_{ip}$$

With the largest sample variance, subject to $\sum_{j=1}^p \phi_{j1}^2 = 1$.

Computing the first principal component

This is equivalent to solve the optimisation problem:

$$\underset{\phi_{11}, \dots, \phi_{p1}}{\text{maximize}} \left\{ \frac{1}{n} \sum_{i=1}^n \left(\sum_{j=1}^p \phi_{j1} x_{ij} \right)^2 \right\} \quad \text{subject to } \sum_{j=1}^p \phi_{j1}^2 = 1.$$

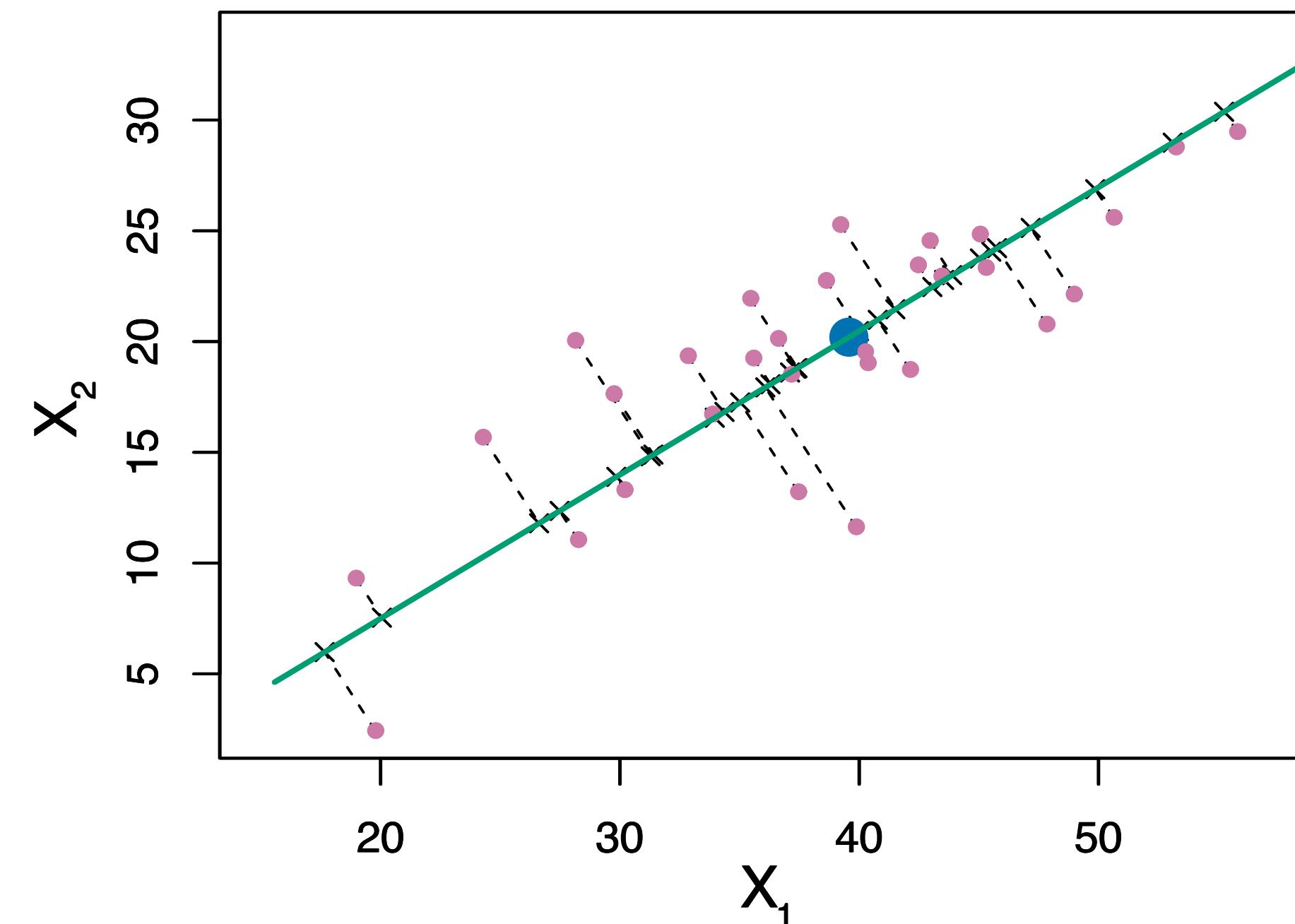
z_{i1}

Since each feature has zero mean (i.e. $\frac{1}{n} \sum_{i=1}^n x_{ij} = 0$), also the average of z_{11}, \dots, z_{n1} is zero. Hence what we are maximising is the sample variance of

$$z_1, \text{ which is } \frac{1}{n} \sum_{i=1}^n z_{i1}^2.$$

Interpreting the first principal component

- ▶ The **first principal component loading vector** $\phi_1 = (\phi_{11}, \phi_{21}, \dots, \phi_{p1})^T$ define the direction along which the data vary the most.
- ▶ The **first principal component scores** are the projections of the n points x_1, \dots, x_n onto this direction.



Computing the second principal component

Given the first principal component Z_1 , the second principal component Z_2 is the linear combination of the features that has maximal variance and is uncorrelated with Z_1 .

The principal component scores take the form:

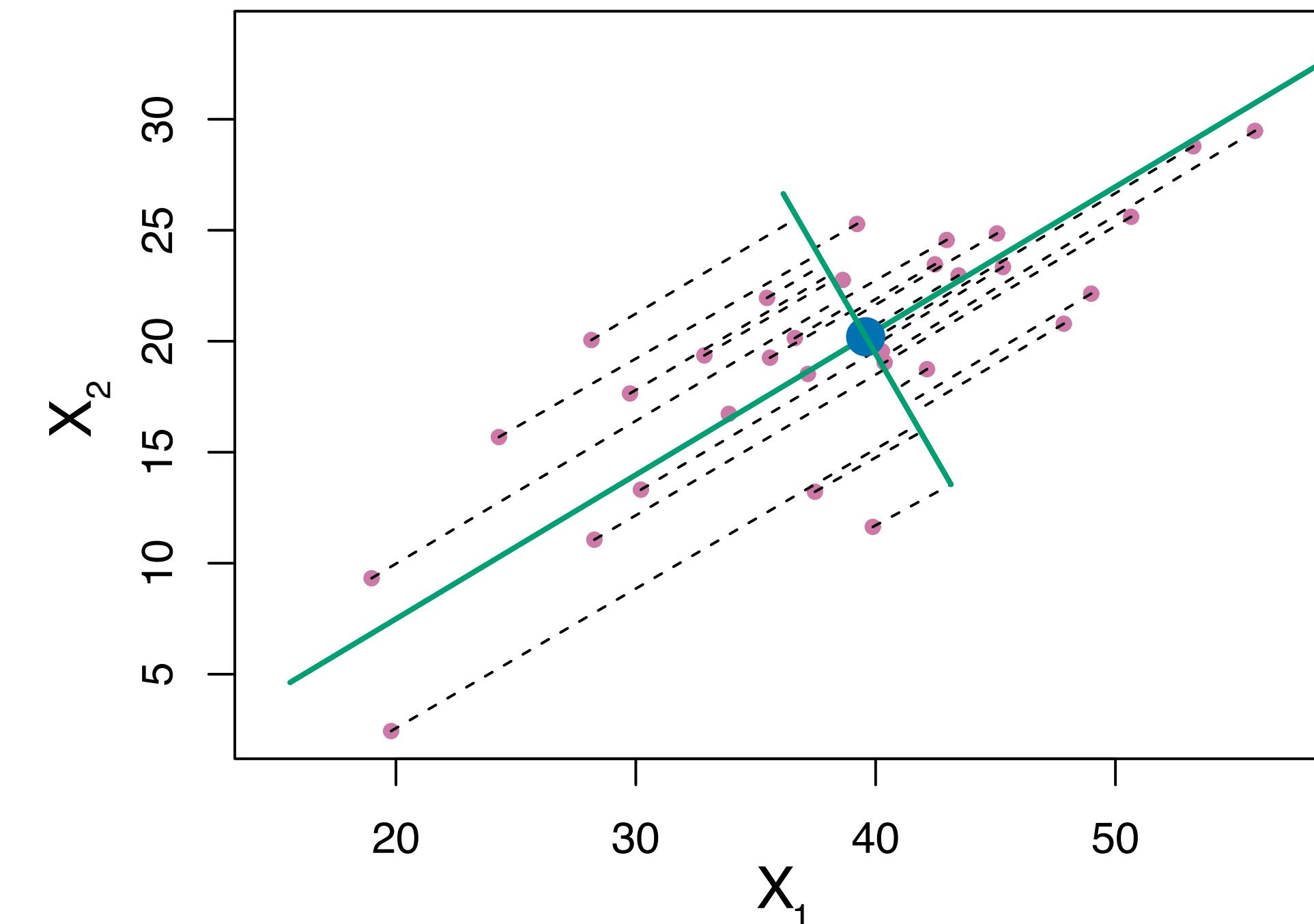
$$z_{i2} = \phi_{12}x_{i1} + \phi_{22}x_{i2} + \dots + \phi_{p2}x_{ip}$$

That maximise the variance, under the constraints that:

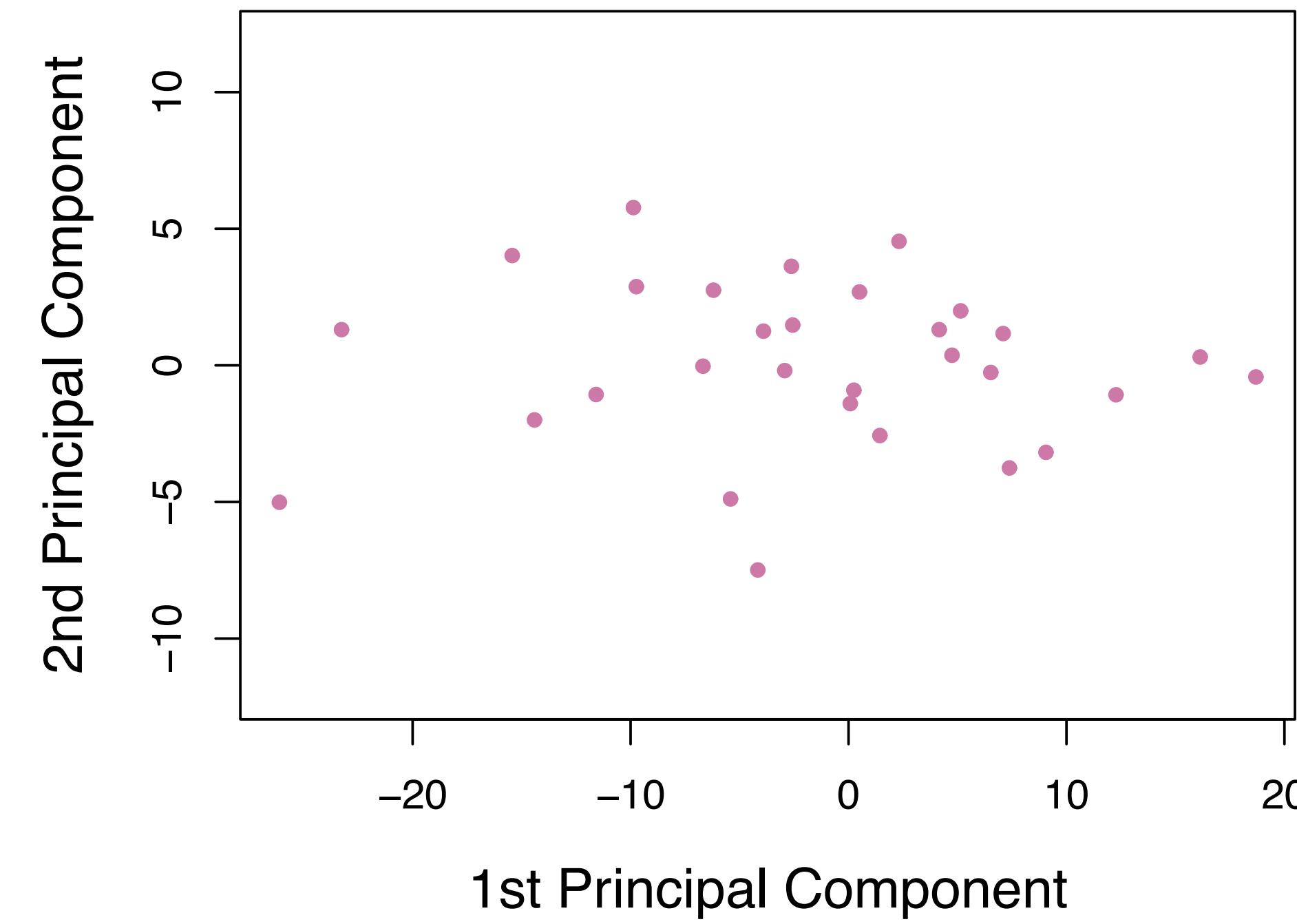
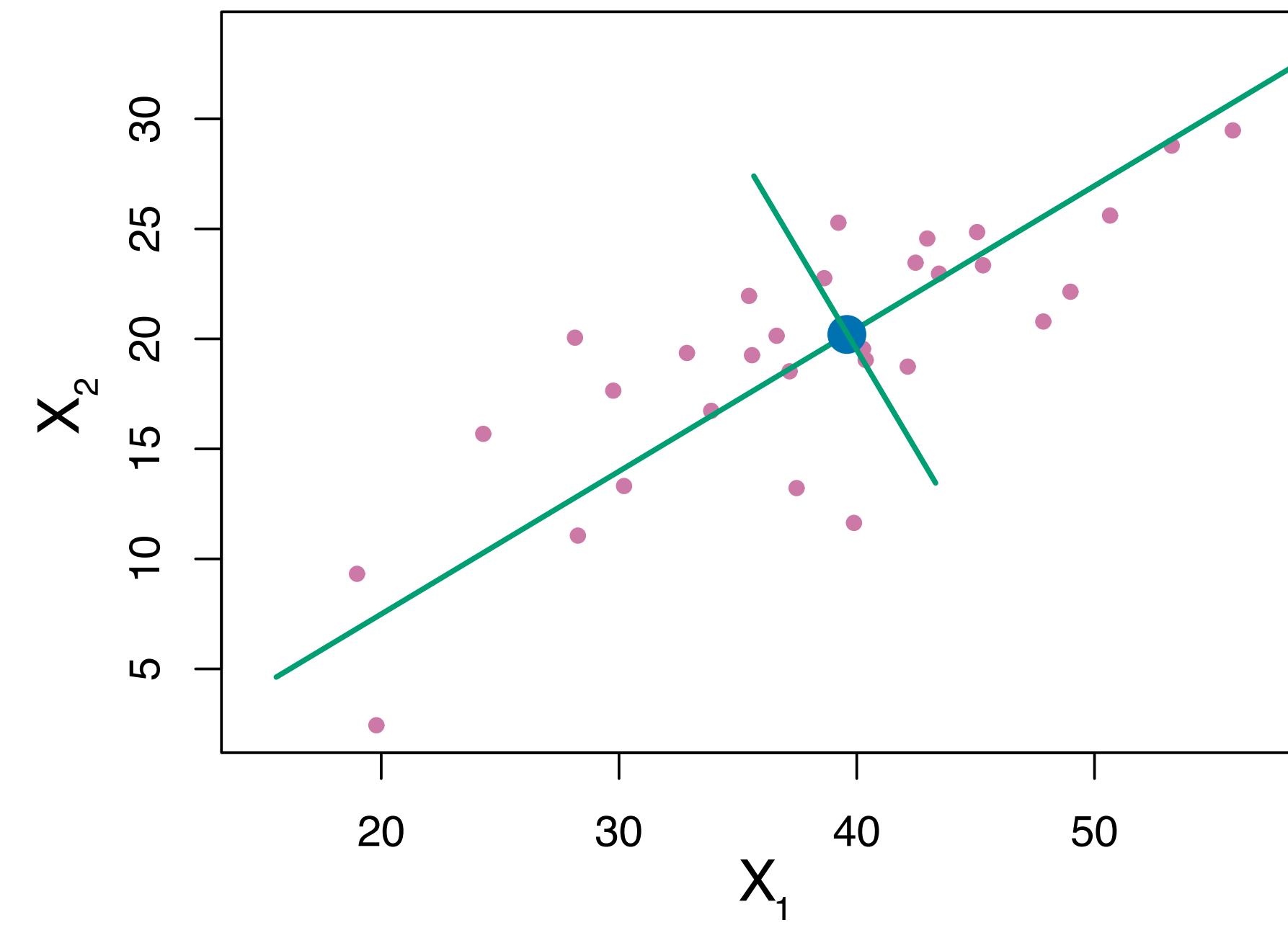
- ▶ $\sum_{j=1}^p \phi_{j2}^2 = 1$
- ▶ The direction of ϕ_2 orthogonal (perpendicular) to the direction of ϕ_1

Interpreting the second principal component

- ▶ The direction of the **second principal component loading vector** ϕ_2 is orthogonal (perpendicular) to the direction of ϕ_1 .
- ▶ The **second principal component scores** are the projections of the n points x_1, \dots, x_n onto the direction of ϕ_2 .



Visualising the first two principal components



When do you expect PCA to have poor performance in feature reduction

(Multiple choices possible)

- ▶ When the features are highly correlated
- ▶ When the features are uncorrelated
- ▶ When the features are dominated by noise

PCA computation using singular value decomposition

The optimisation problem can be solved via singular value decomposition of the the $n \times p$ data matrix \mathbf{X} :

$$\mathbf{X} = \mathbf{U}\mathbf{D}\mathbf{V}^T$$

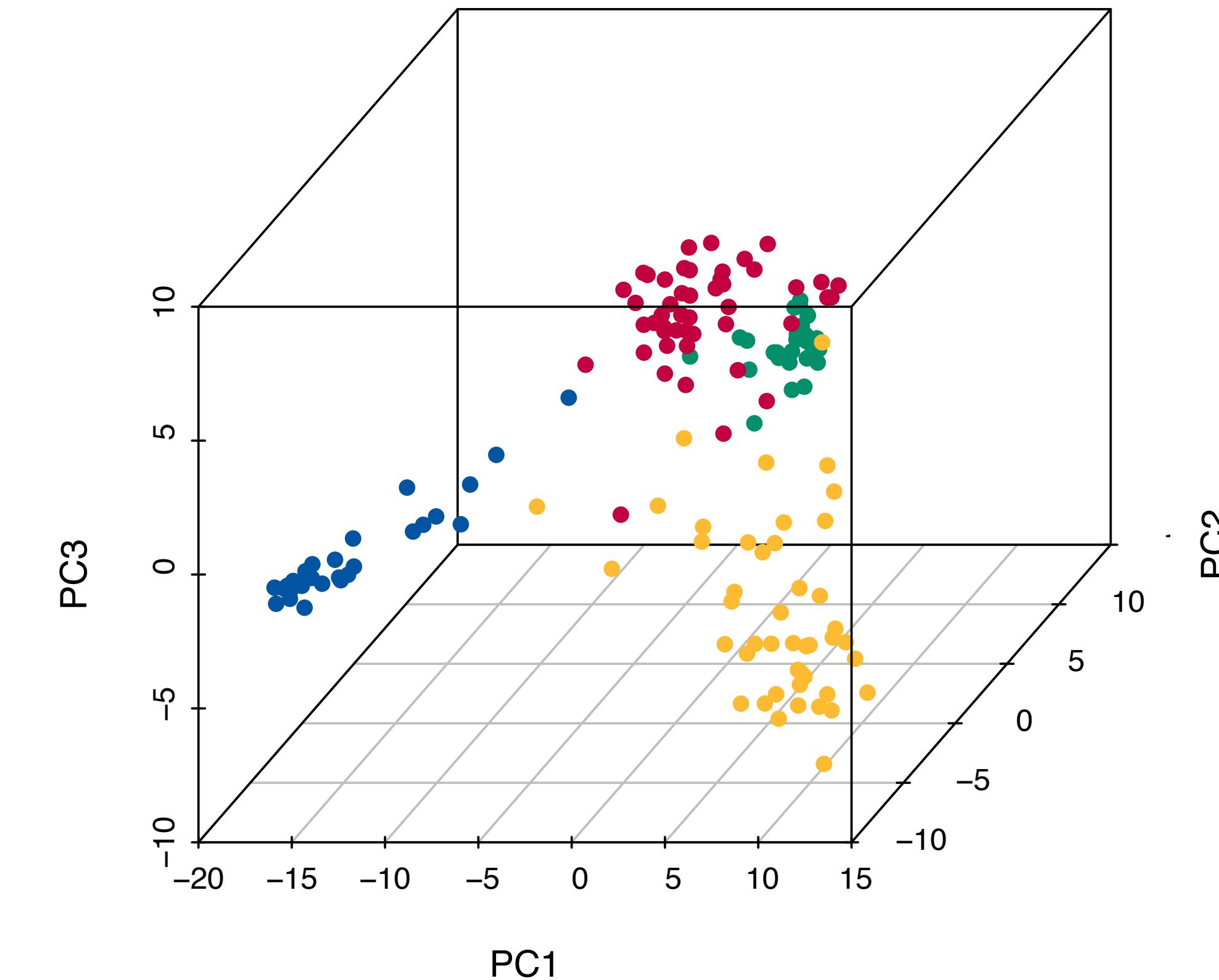
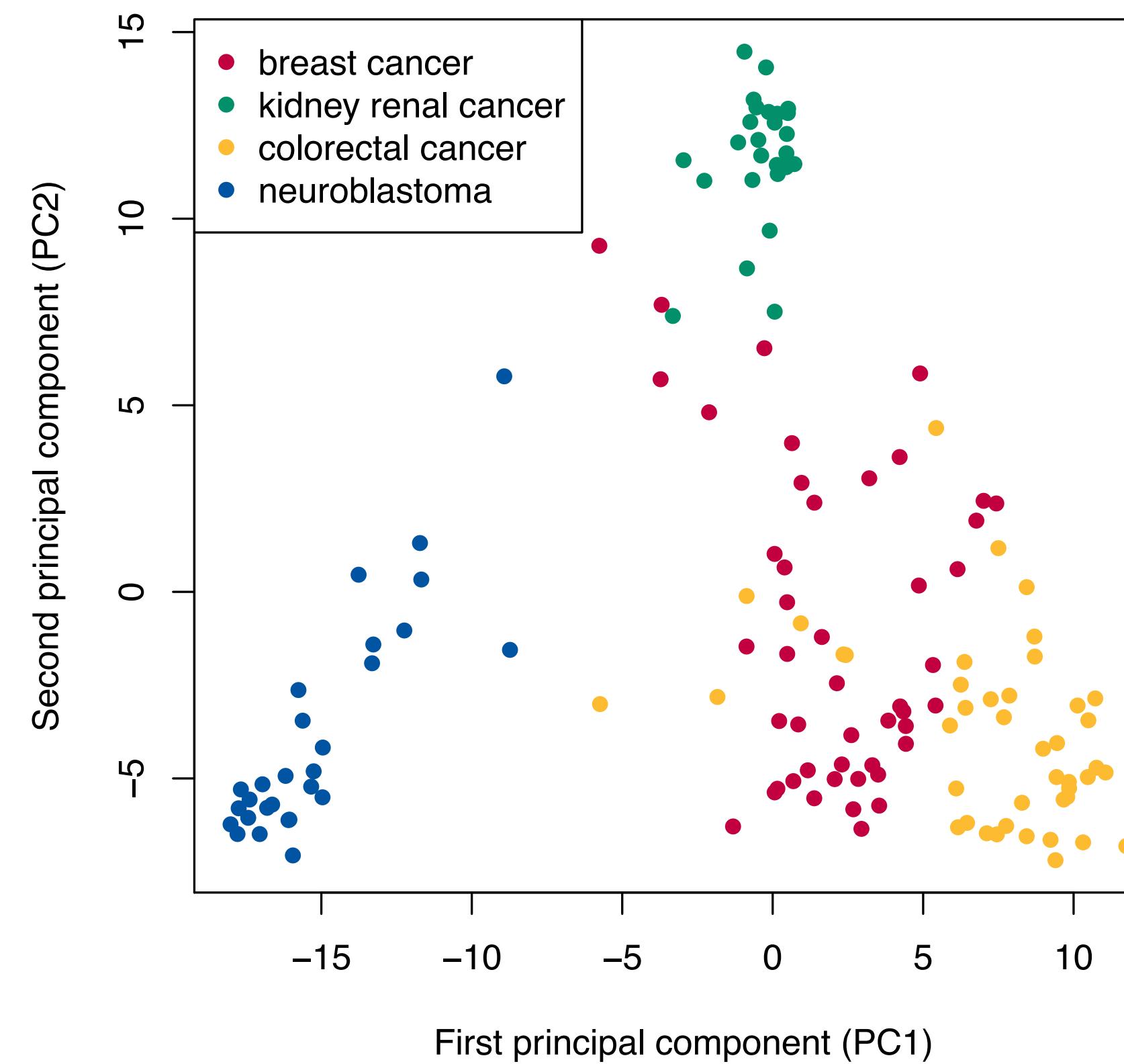
For $n > p$ this is a unique decomposition such that:

- ▶ \mathbf{U} is a $n \times p$ orthogonal matrix (i.e. $\mathbf{U}^T\mathbf{U} = \mathbf{I}_p$)
- ▶ \mathbf{D} is a $p \times p$ diagonal matrix with $d_j > 0$ and $d_j \geq d_{j+1}$ known as singular values
- ▶ \mathbf{V} is a $p \times p$ orthogonal matrix (i.e. $\mathbf{V}^T\mathbf{V} = \mathbf{I}_p$)

The columns of \mathbf{UD} are the principal components of \mathbf{X} and $\frac{d_1^2}{n}, \frac{d_2^2}{n}, \dots, \frac{d_p^2}{n}$ is the variance explained by each principal component.

Example: GDSC dataset - scores

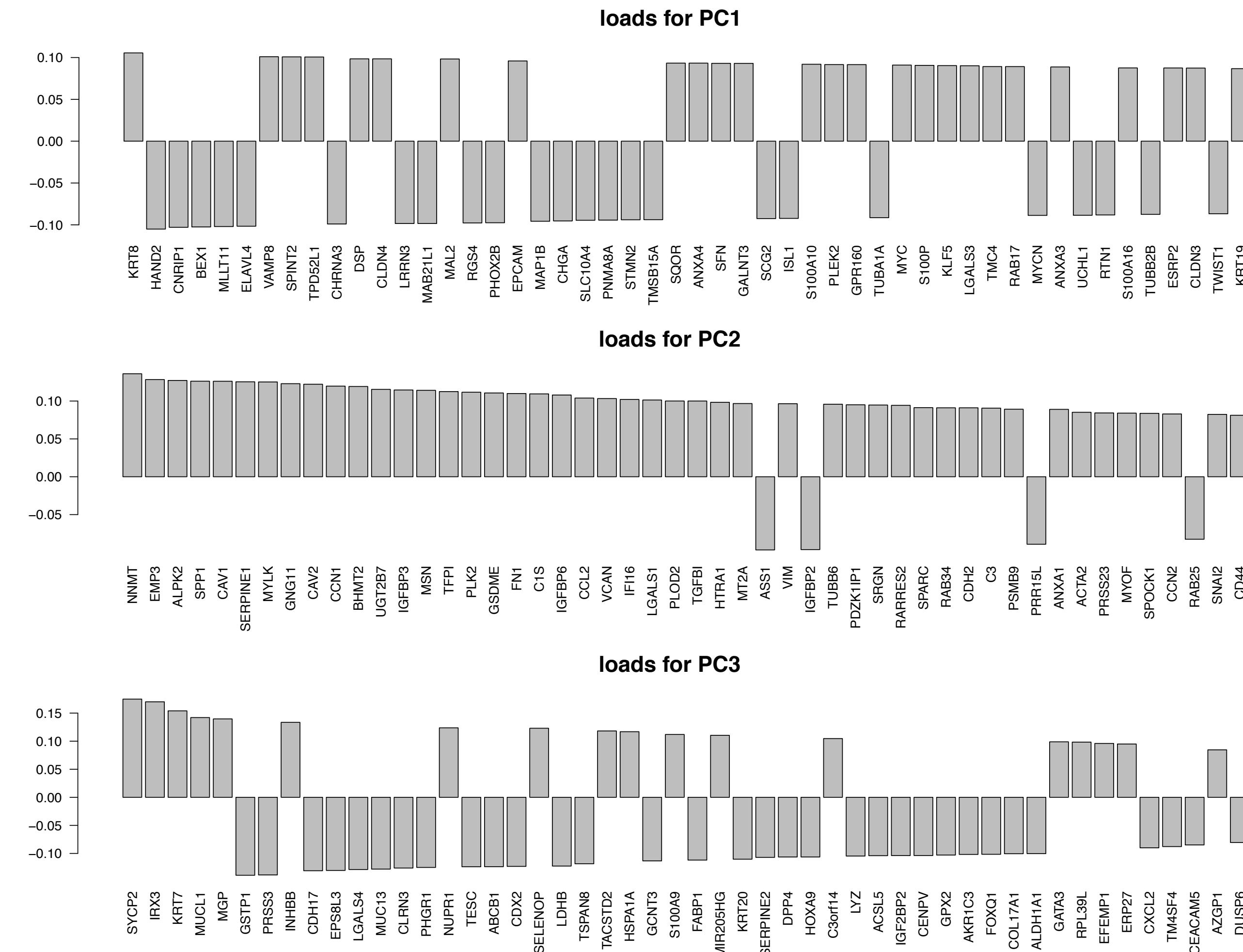
RNA expression data (244 genes) for 148 cell lines from four cancer types.



NOTE: this is an unsupervised learning problem, points are coloured *a posteriori* by cancer type

Example: GDSC dataset - loads

Loads for the first 3 principal components (only top 50 genes shown).

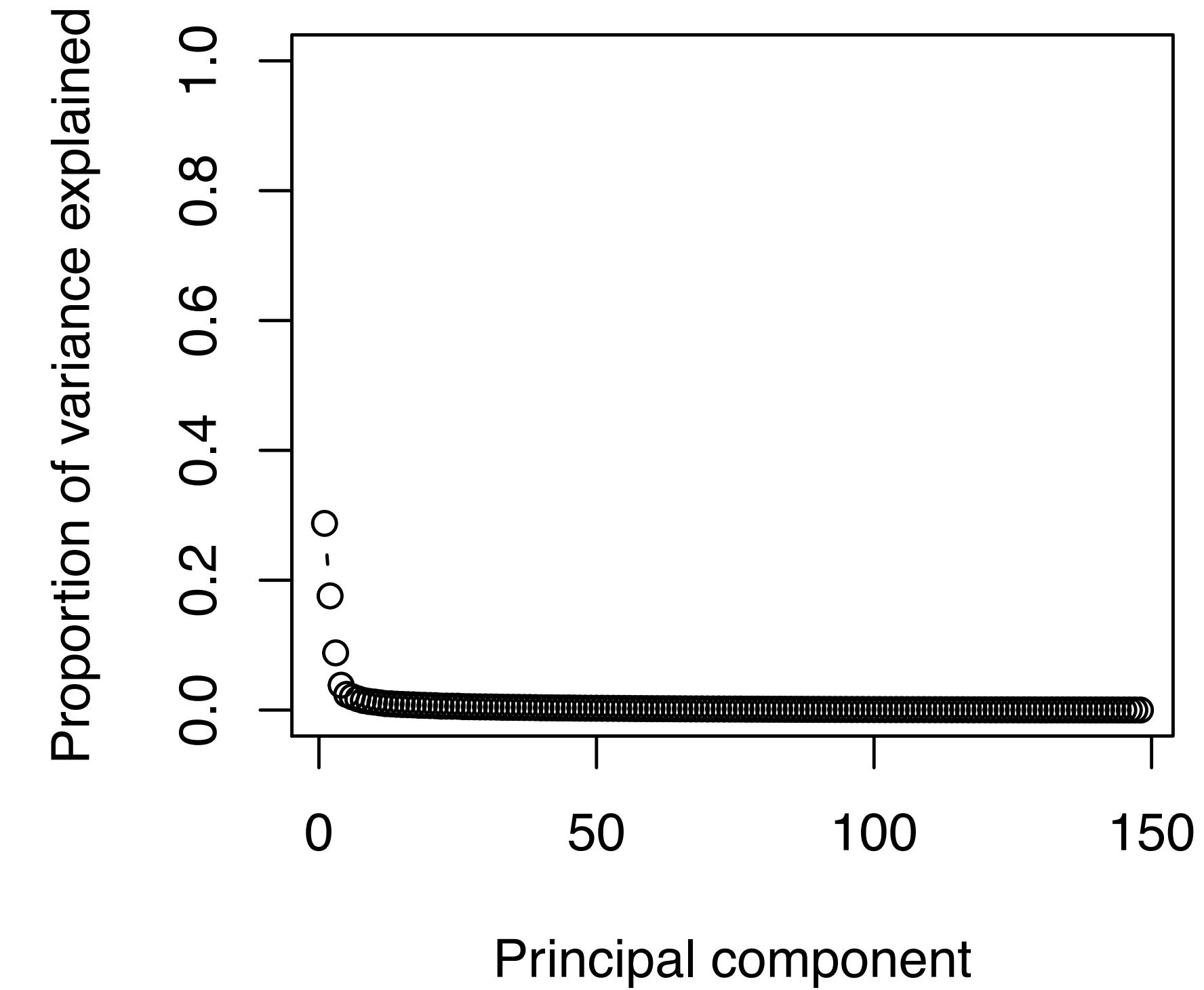


Deciding how many principal components to use

- ▶ In general an $n \times p$ data matrix \mathbf{X} has $\min(n - 1, p)$ distinct principal components.
- ▶ We want to use the smallest number of principal components that give us a good understanding of the data.
- ▶ There is no unique way to define that, but approaches are in general based on the variance explained by the principal components.

Example: GDSC dataset - proportion of variance

We can look at the **proportion of variance explained** by each principal component.

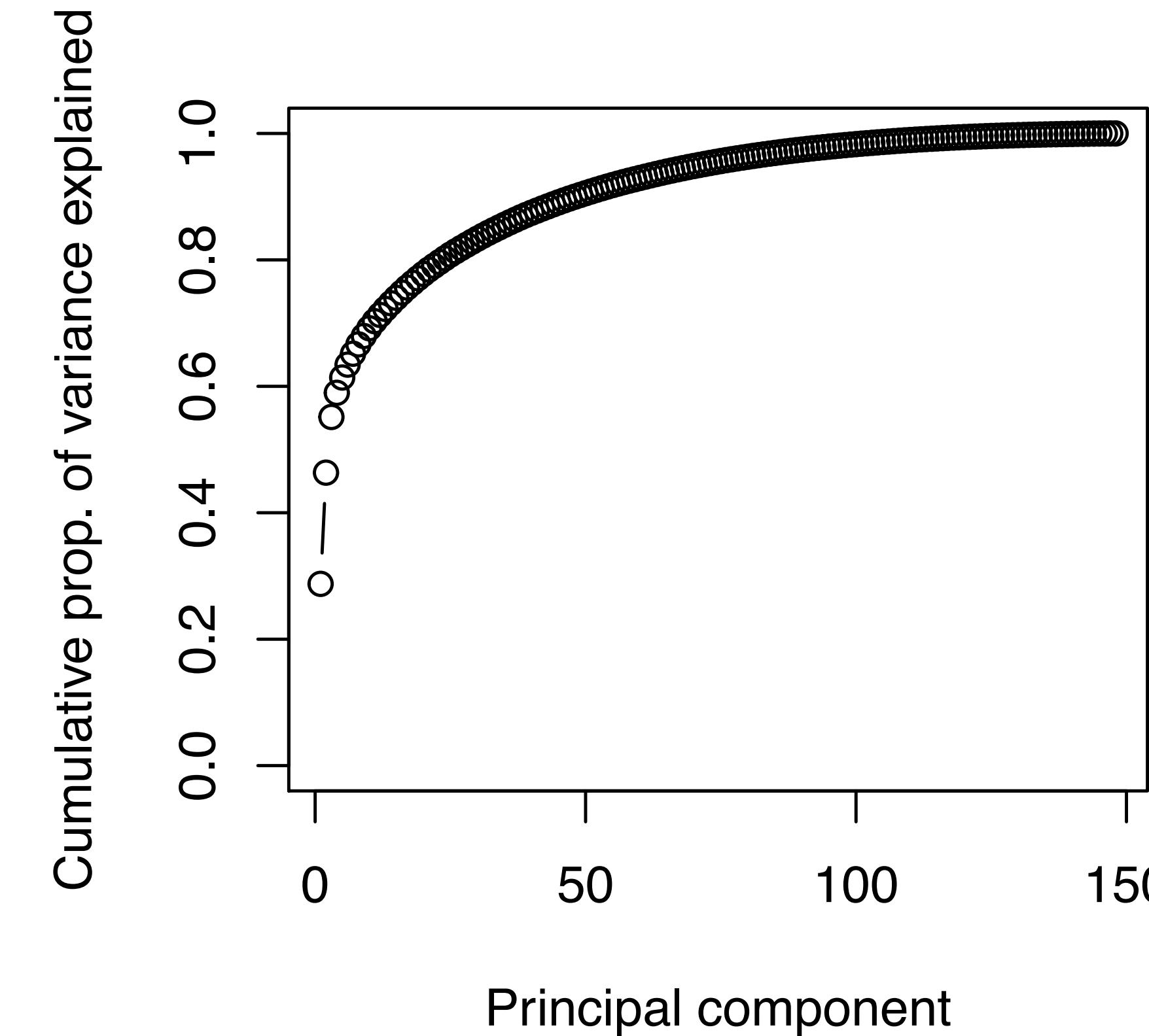


How to select the number of features?

Elbow method to select where there is a drop.

Example: GDSC dataset - cumulative proportion of variance

We can look at the **cumulative proportion of variance explained**.



How to select the number of features?

Set a threshold on what is the minimum desired cumulative variance explained.

Summary of dimensionality reduction with PCA

- ▶ **Goal:** Reduce the number of variables while retaining the most important information.
- ▶ **Key concepts:**
 - ▶ Projects data onto a smaller subspace.
 - ▶ Maximises variance in the data with fewer dimensions.
- ▶ **Interpretation:**
 - ▶ **Principal Components:** Ranked by how much variance they explain.
 - ▶ **Loadings:** Show how much each original variable contributes to each principal component.
- ▶ **Examples of biomedical Applications:**
 - ▶ Reducing dimensionality in gene expression data for cancer classification.
 - ▶ Simplifying complex patient measurements to identify dominant health factors.

Clustering

- ▶ Aim: Group observations into subsets, called clusters or segments, so that observations within a cluster are more similar to each other than observations assigned to different clusters.
- ▶ Requires a definition of *similarity* or *difference*.
- ▶ Unsupervised problem that try to find structures (clusters) based on the given data

Which of the following applications could make use of clustering?

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(Multiple choices possible)

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- ▶ Discover cancer patients subtypes based on clinical and genetic features.
- ▶ Reducing the number of variables in gene expression datasets to identify major patterns.
- ▶ Identifying groups of patients who respond similarly to a treatment based on biomarker changes.
- ▶ Categorising proteins based on structural similarities to understand functional relationships.
- ▶ Projecting high-dimensional biomarker data into 2D or 3D space for visualisation.

Note: These are all unsupervised learning problems that can be addressed either by PCA or clustering.

K-Means clustering

Given a desired number of clusters K , the K-means clustering assigns each observations to exactly one of the K clusters.

Mathematically, let C_1, \dots, C_K denote sets containing the indices of the observations in each cluster. These sets satisfy these two properties:

- ▶ $C_1 \cup C_2 \cup \dots \cup C_K = \{1, \dots, n\}$: each observation belongs to at least one of the K clusters
- ▶ $C_k \cap C_{k'} = \emptyset$ for all $k \neq k'$: the clusters are non-overlapping

K-Means clustering formulation

We want to define the clusters so that similar points are in the same cluster and dissimilar points in different clusters.

$$\underset{C_1, \dots, C_K}{\text{minimize}} \left\{ \sum_{k=1}^K W(C_k) \right\}$$

Where $W(C_k)$ is a measure of within-cluster variation for cluster C_k .

When using the squared Euclidean distance:

$$W(C_k) = \frac{1}{|C_k|} \sum_{i, i' \in C_k} \sum_{j=1}^p (x_{ij} - x_{i'j})^2$$

Where $|C_k|$ denotes the number of observations in the k th cluster

K-Means clustering formulation

The optimisation problem becomes

$$\underset{C_1, \dots, C_K}{\text{minimize}} \left\{ \sum_{k=1}^K \frac{1}{|C_k|} \sum_{i, i' \in C_k} \sum_{j=1}^p (x_{ij} - x_{i'j})^2 \right\}$$

This is equivalent to:

$$\underset{C_1, \dots, C_K}{\text{minimize}} \left\{ \sum_{k=1}^K \sum_{i \in C_k} \sum_{j=1}^p (x_{ij} - \bar{x}_{kj})^2 \right\}$$

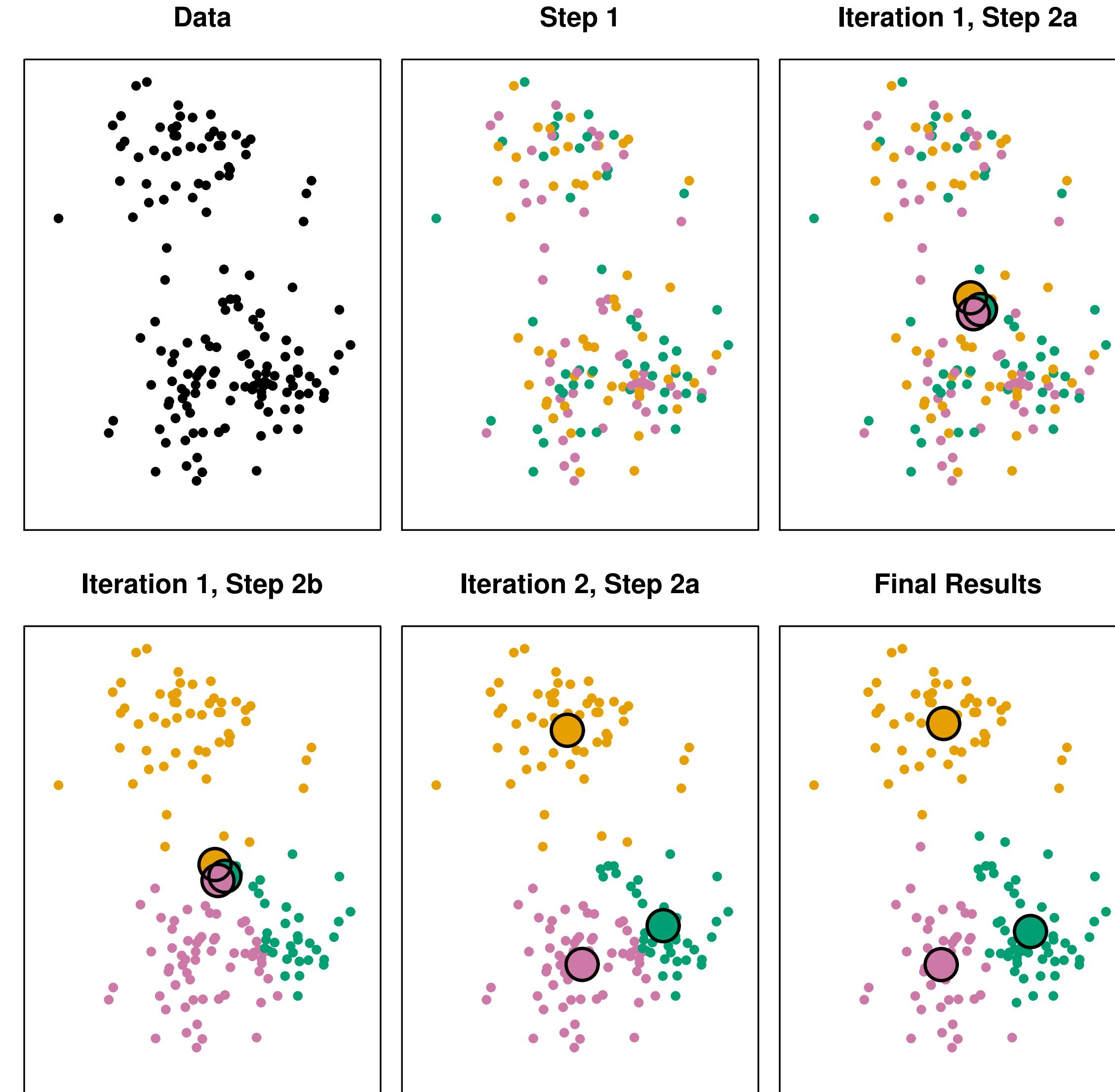
Where $\bar{x}_{kj} = \frac{1}{|C_k|} \sum_{i \in C_k} x_{ij}$ is the mean for feature j in cluster C_k

K-Means clustering algorithm

Systematic assessment of all possible partitions is unfeasible. We need a smart optimisation algorithm.

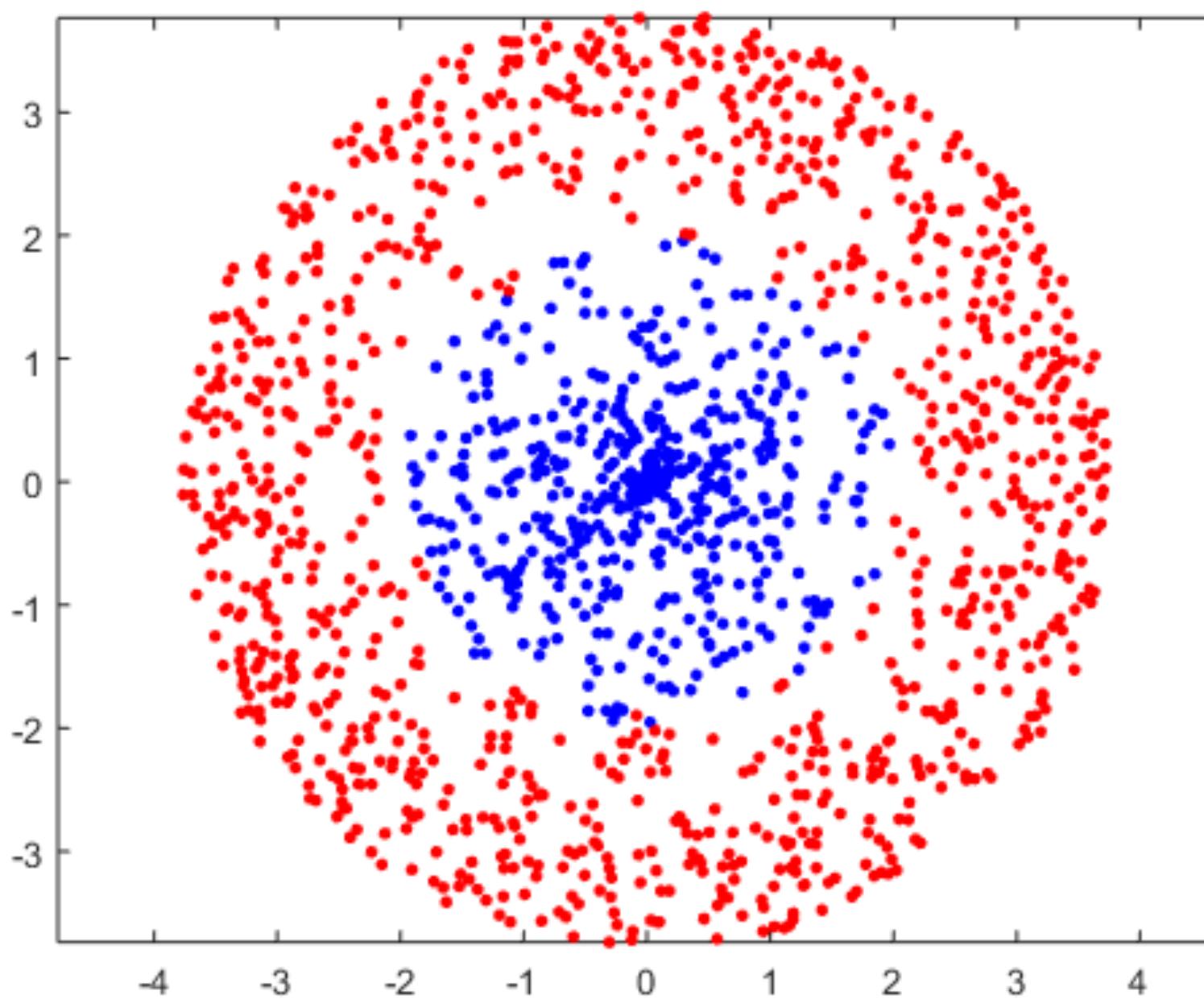
1. Randomly assign each observation to one cluster
2. Iterate the next two steps until cluster assignment stops changing
 - 2.a. For each cluster compute the mean vector \bar{x}_k (i.e. the centroid)
 - 2.b. Assign each observation to the cluster whose centroid is closest (based on Euclidian distance)

K-Means clustering algorithm



What will happen if you apply K-means to this example with K=2

- ▶ It will correctly identify the two classes
- ▶ It depends on the initialisation
- ▶ It will not be able to correctly identify the two classes

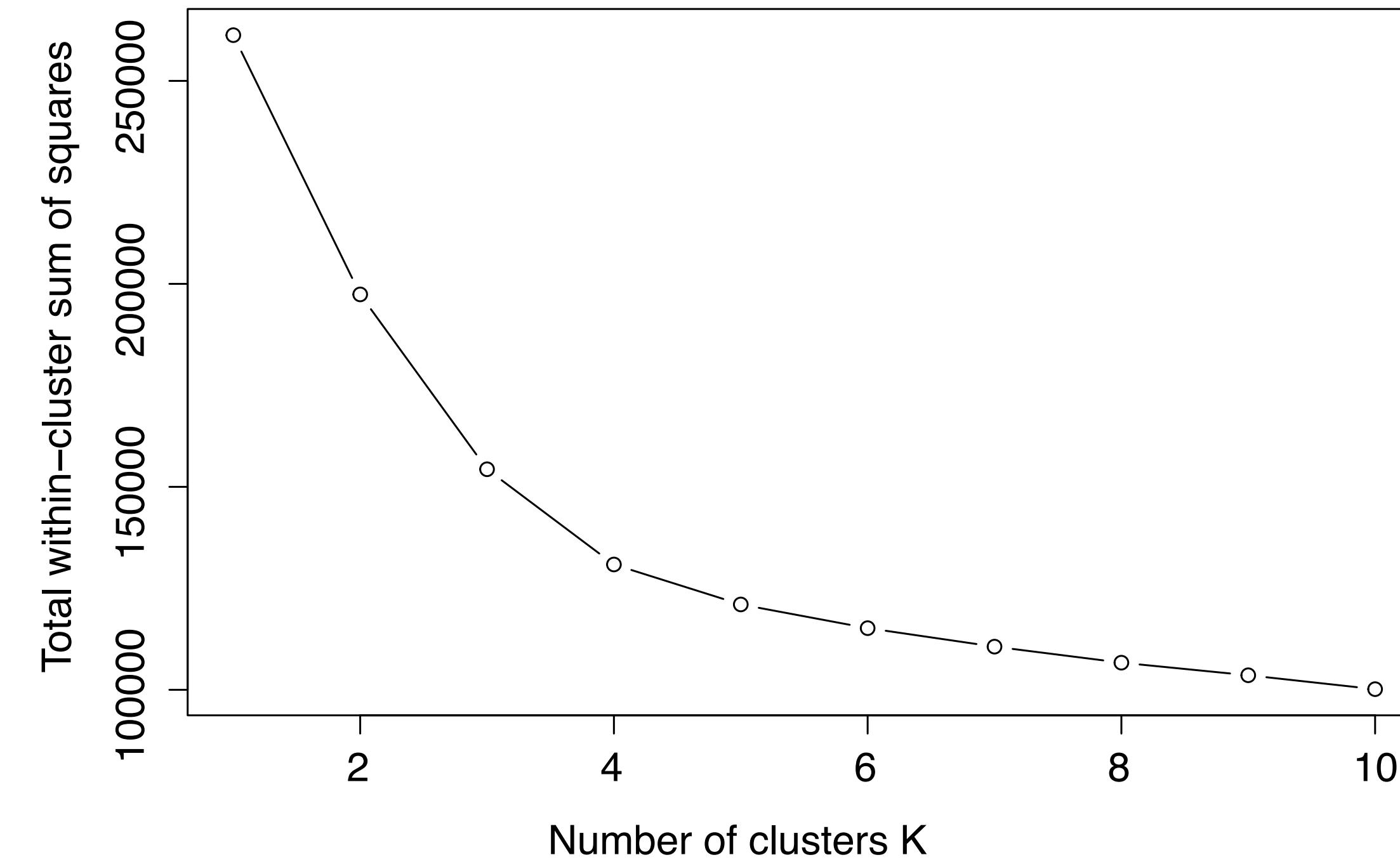


K-means algorithm problems and solutions

- ▶ Problem: Does not guarantee the global minimum. Solution: Check if switching single observations to a different group decreases the objective function.
- ▶ Problem: Different random initialisations can provide different solutions. Solution: Run it multiple times and select the solution with minimum objective function.
- ▶ We need to define the number of clusters.

K-means example: GDSC data

Compute K-means for different number of clusters and look at the total within-cluster sum of squares for each clustering.



Look at the elbow to define the optimal number of clusters.

K-means example: GDSC data

Number of cases of each cancer type (columns) in each cluster (rows), when using K=4.

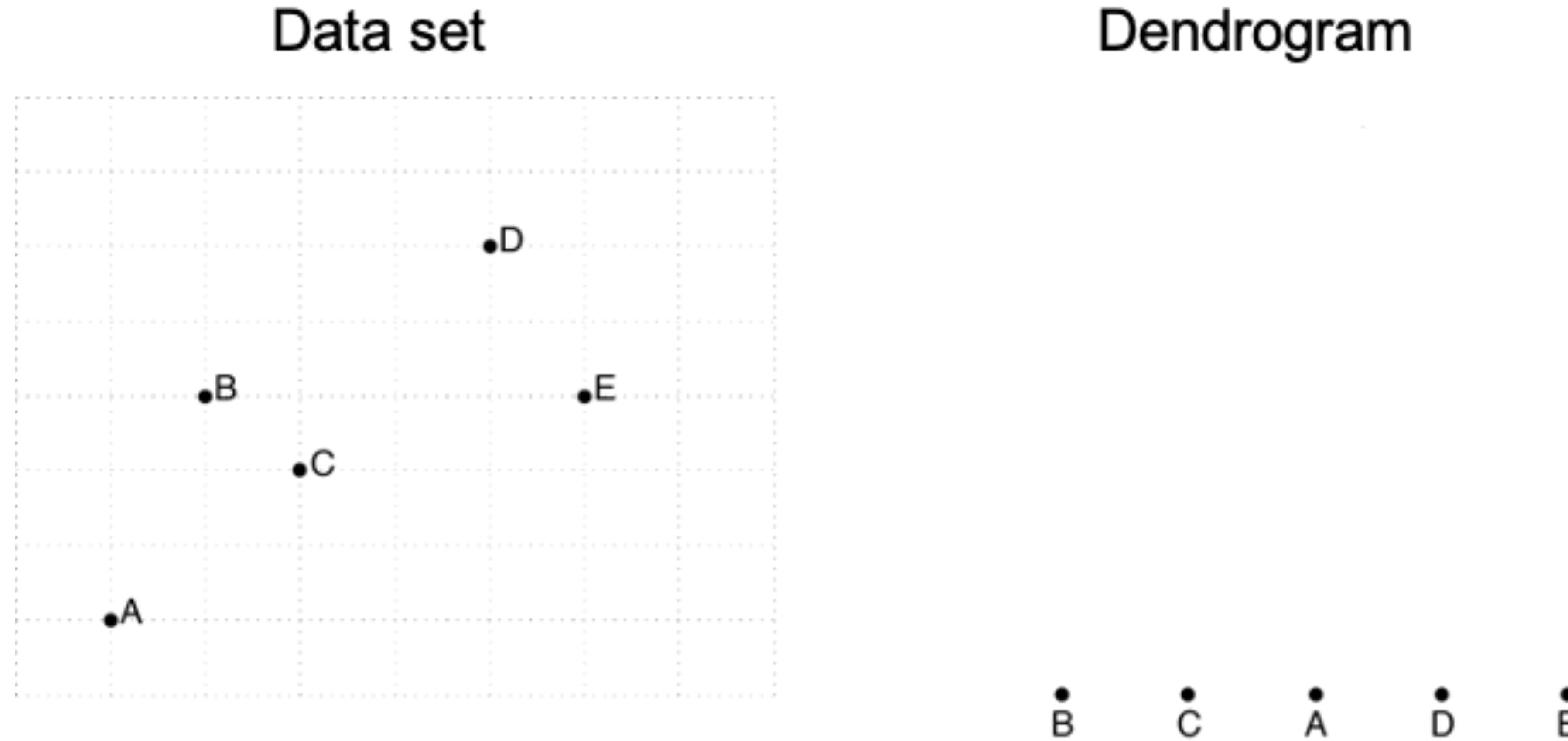
	breast	colorectal	kidney	neuroblastoma
1	6	0	28	1
2	41	7	0	0
3	0	37	0	0
4	0	1	0	27

Hierarchical clustering

- ▶ Differently from K-means, *hierarchical clustering* does not require to specify the number of clusters.
- ▶ It organises observations in a hierarchy, where clusters at each level of the hierarchy are created merging clusters at the next lower level.
- ▶ The approach that we will see is *bottom-up*:
 - ▶ It starts from the lowest level, where each observation is a singleton cluster.
 - ▶ For $N-1$ steps it merges a selected pair of clusters (i.e. the most similar) in a single cluster.
- ▶ The process can be visualised using a *dendrogram*.

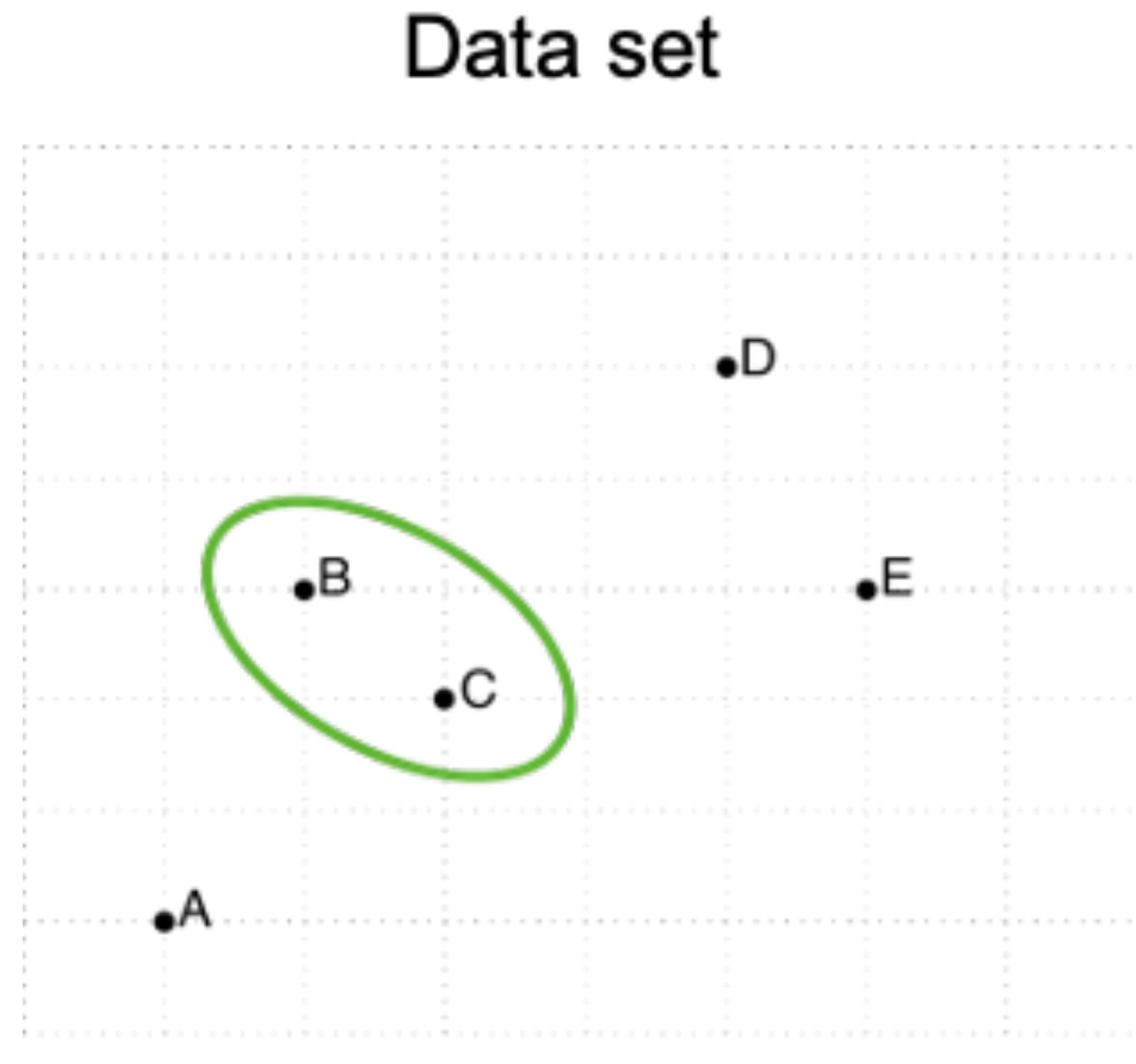
Hierarchical clustering - bottom-up approach

Each *leaf* of the dendrogram is an observation.



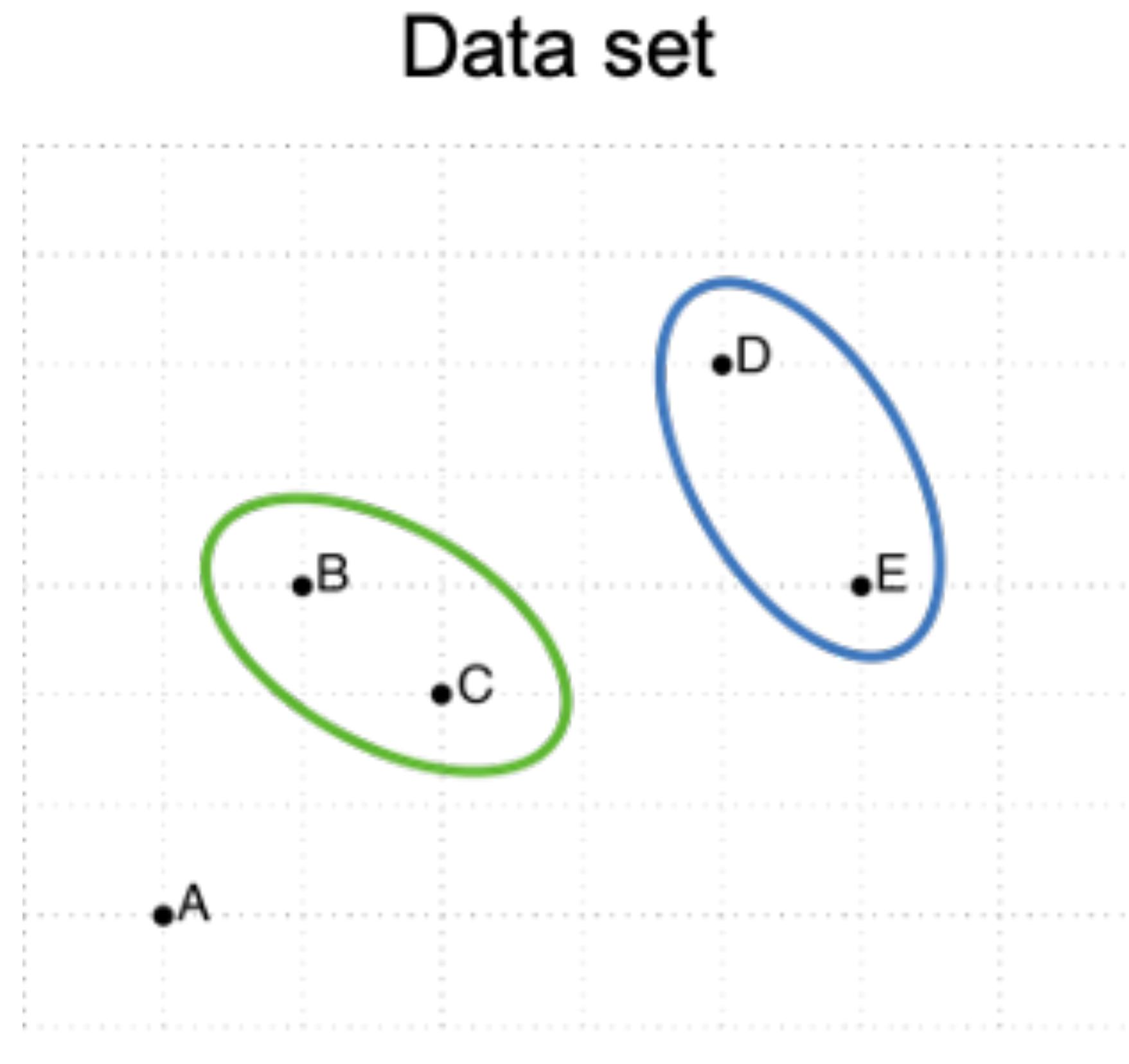
Hierarchical clustering - bottom-up approach

Leaves begin to fuse into *branches* for observations which are similar.



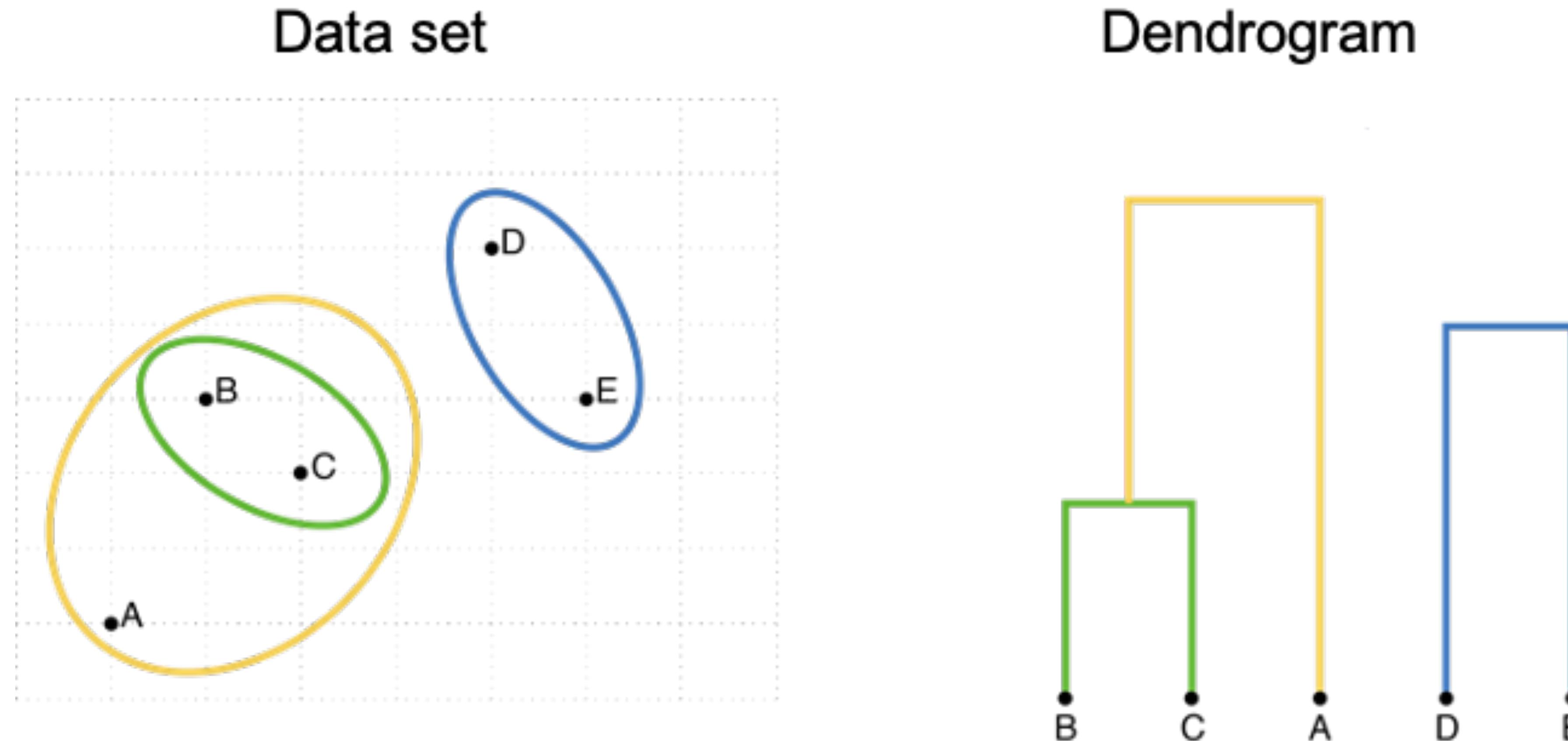
Hierarchical clustering - bottom-up approach

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Hierarchical clustering - bottom-up approach

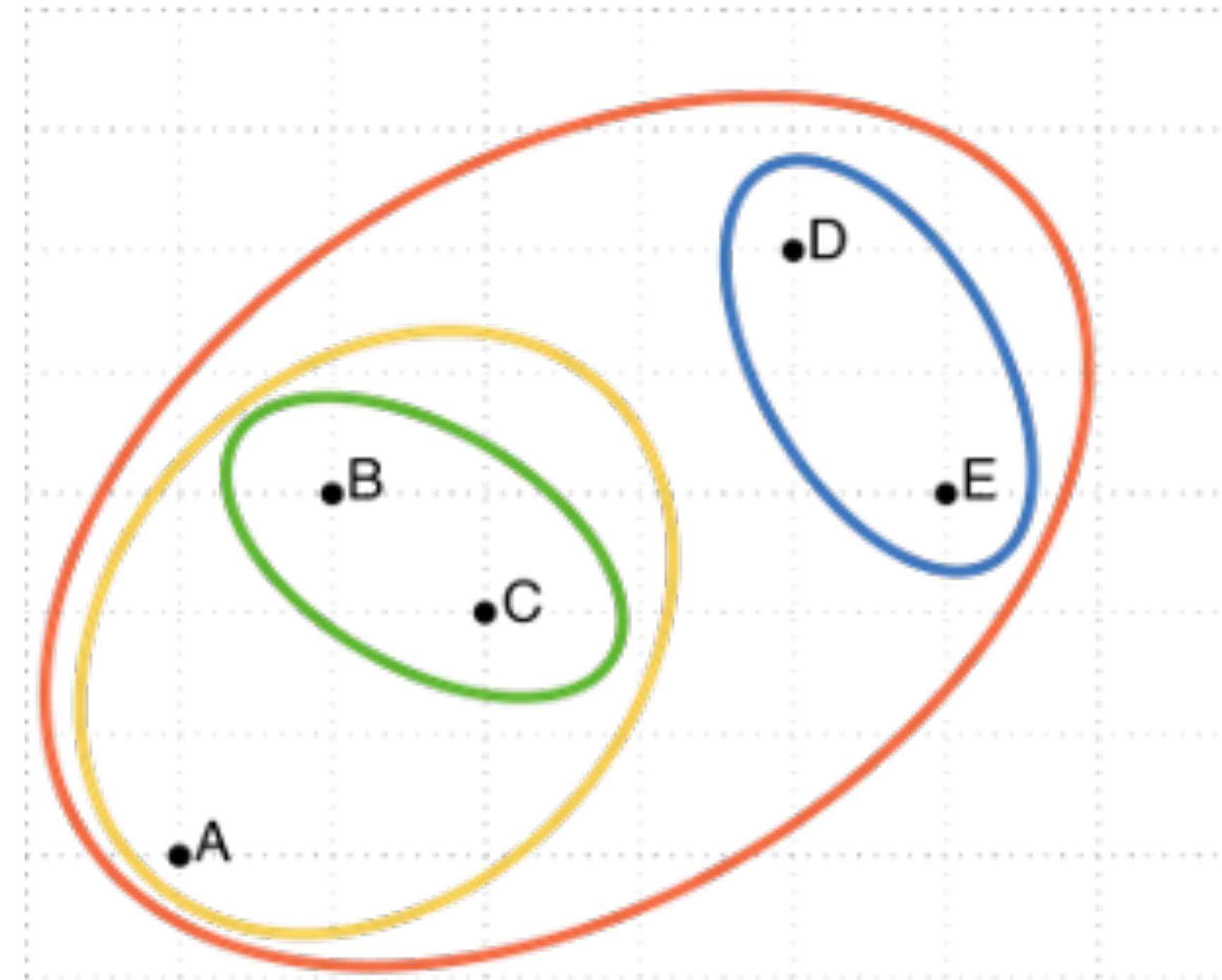
Branches start to fuse either with leaves...



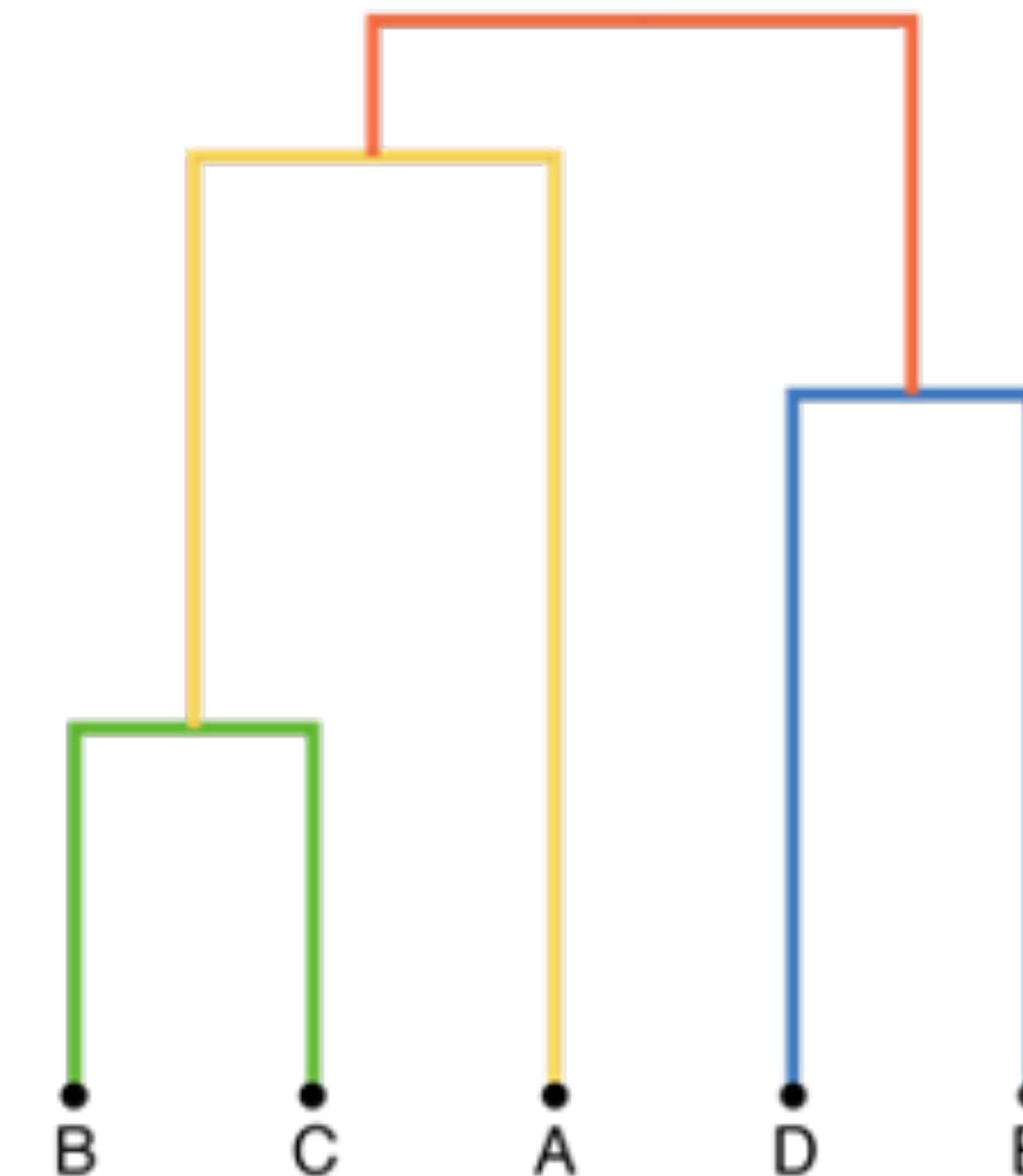
Hierarchical clustering - bottom-up approach

Branches start to fuse either with leaves or with other branches.

Data set



Dendrogram



Hierarchical clustering - Linkage

Given a metric of dissimilarity $d_{i,i'}$ between a pair of observations i and i' (e.g. Euclidean distance), we also need to define a measure of dissimilarity between clusters, i.e. the *linkage*.

Considering two clusters G and H , the most used types of linkage are:

- ▶ *Single linkage*

$$d_{SL}(G, H) = \min_{i \in G, i' \in H} d_{i,i'}$$

- ▶ *Complete linkage*

$$d_{CL}(G, H) = \max_{i \in G, i' \in H} d_{i,i'}$$

- ▶ *Average linkage*

$$d_{AL}(G, H) = \frac{1}{n_G n_H} \sum_{i \in G} \sum_{i' \in H} d_{i,i'}$$

Where n_G and n_H are the number of observations in cluster G and H

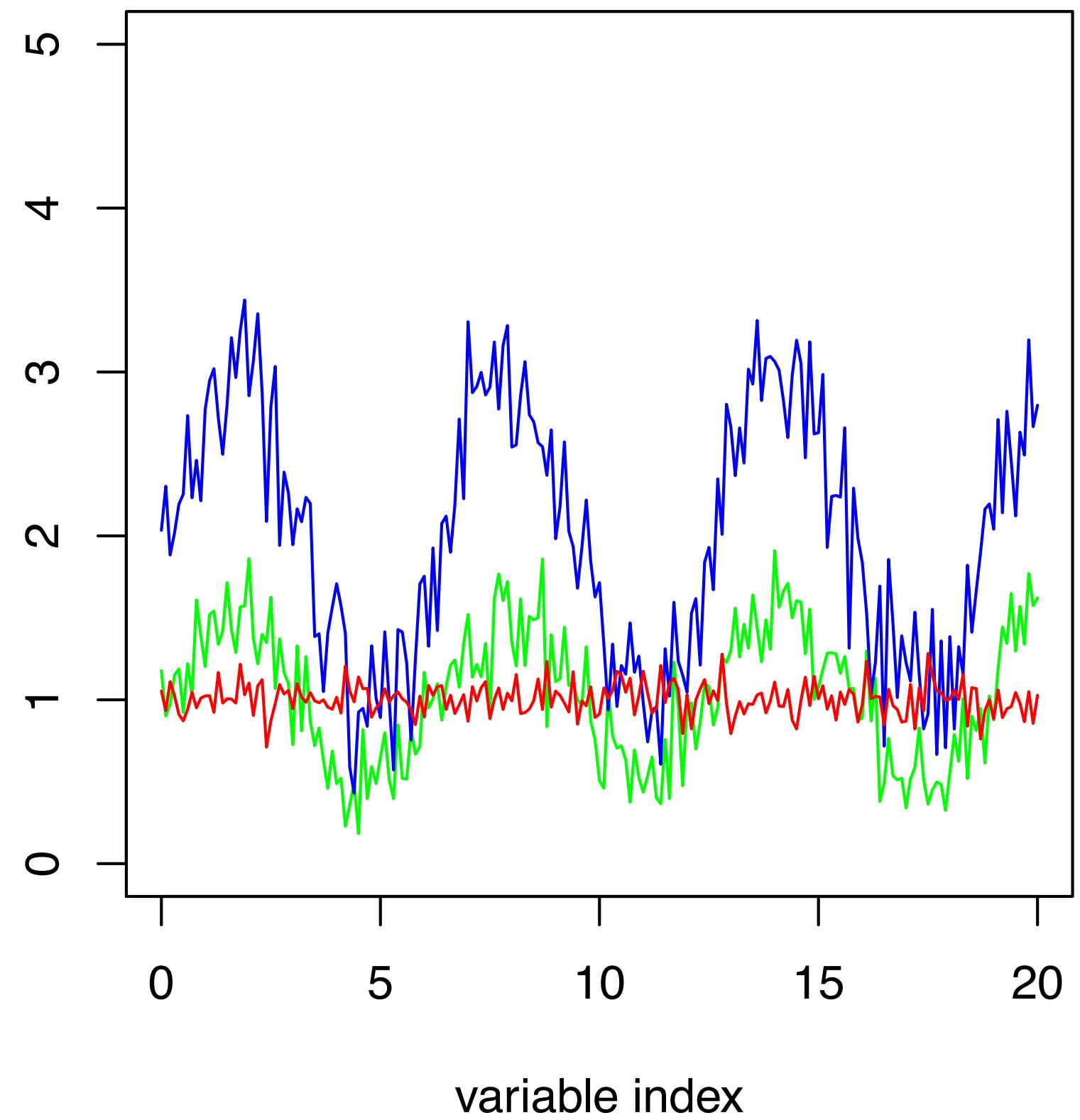
Hierarchical clustering - Dissimilarity metric

As metric of dissimilarity $d_{i,i'}$ between a pair of observations i and i' the most common approaches used are:

- ▶ Euclidean distance
- ▶ Correlation (of observations across features)

Which metric of dissimilarity between observations would you use?

These are time series data where each time point is a feature and we want to focus on similarity of the temporal profile.



- ▶ Euclidean distance
- ▶ Correlation

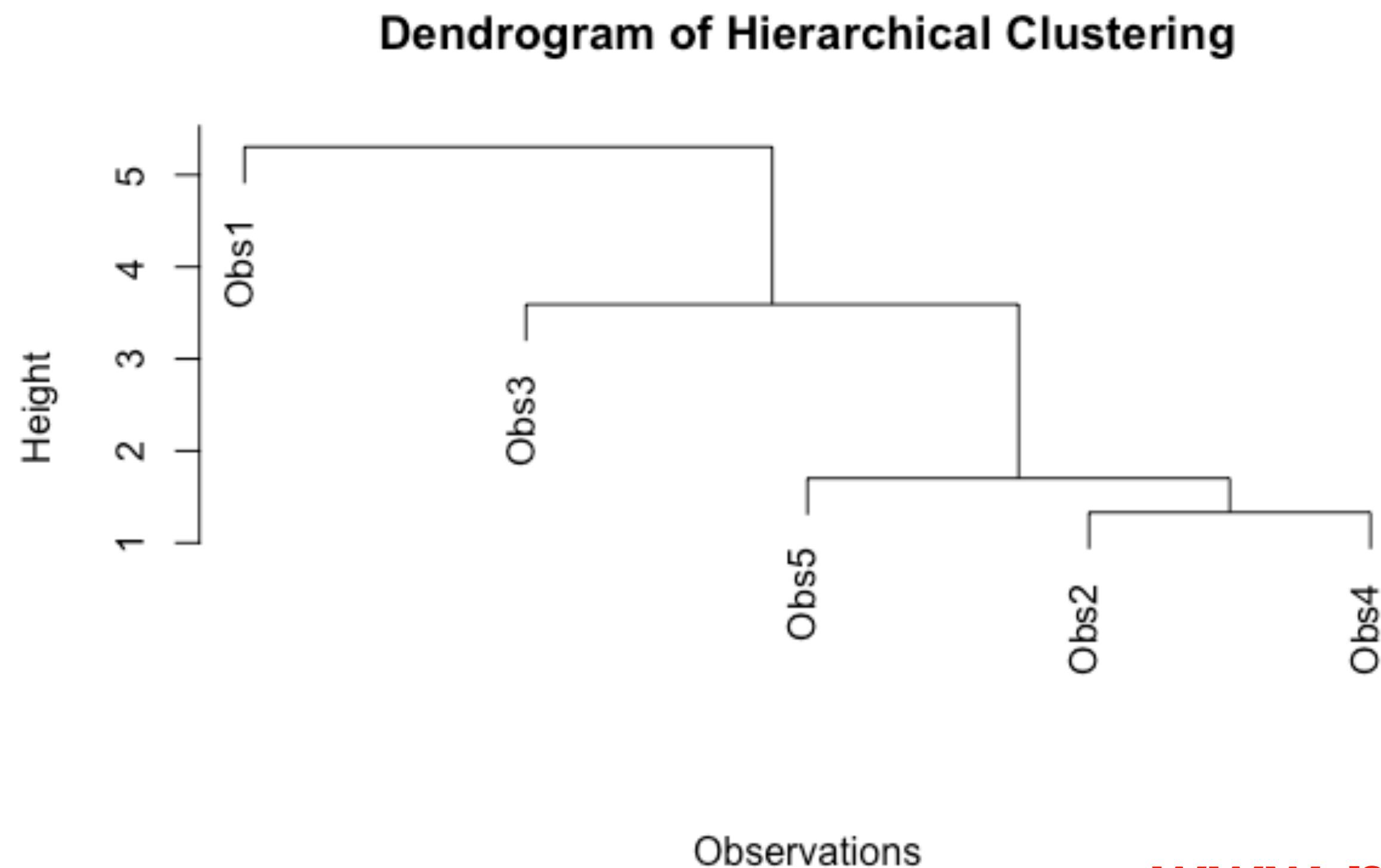
Hierarchical clustering - Interpreting the dendrogram

- ▶ Branches' height is proportional to the similarity between nodes
- ▶ Observations that fuse at the bottom of the dendrogram are similar to each other
- ▶ Observations that fuse at the top of the dendrogram are different from each other
- ▶ We can cut the dendrogram at a certain height to obtain clusters

Which of the following two pairs of observations is more similar?

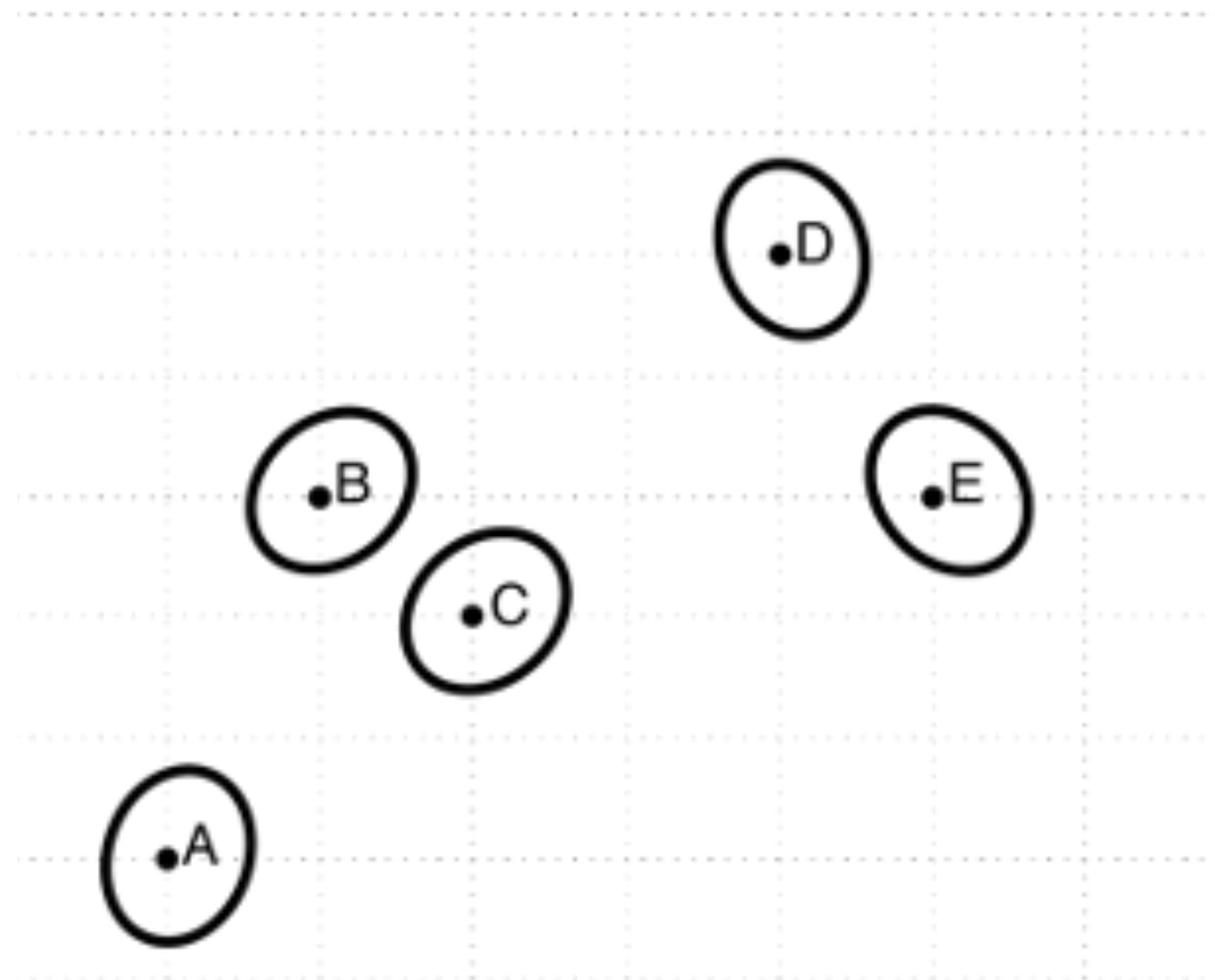
(Multiple choices possible)

- ▶ Observations 1 and 2
- ▶ Observations 1 and 3
- ▶ Observations 3 and 5
- ▶ Observations 3 and 2
- ▶ Observations 3 and 4

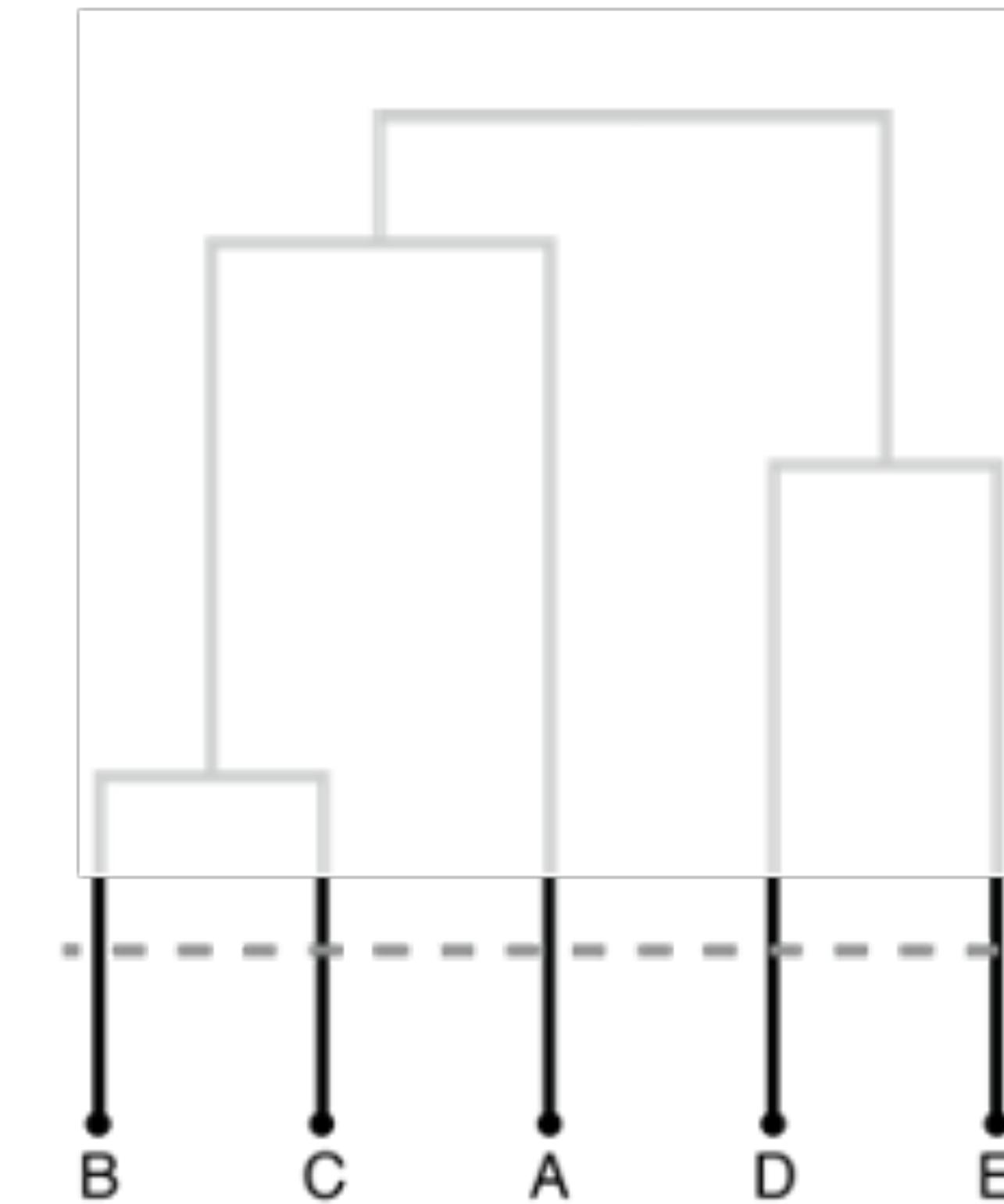


Hierarchical clustering - How to obtain clusters

Data set

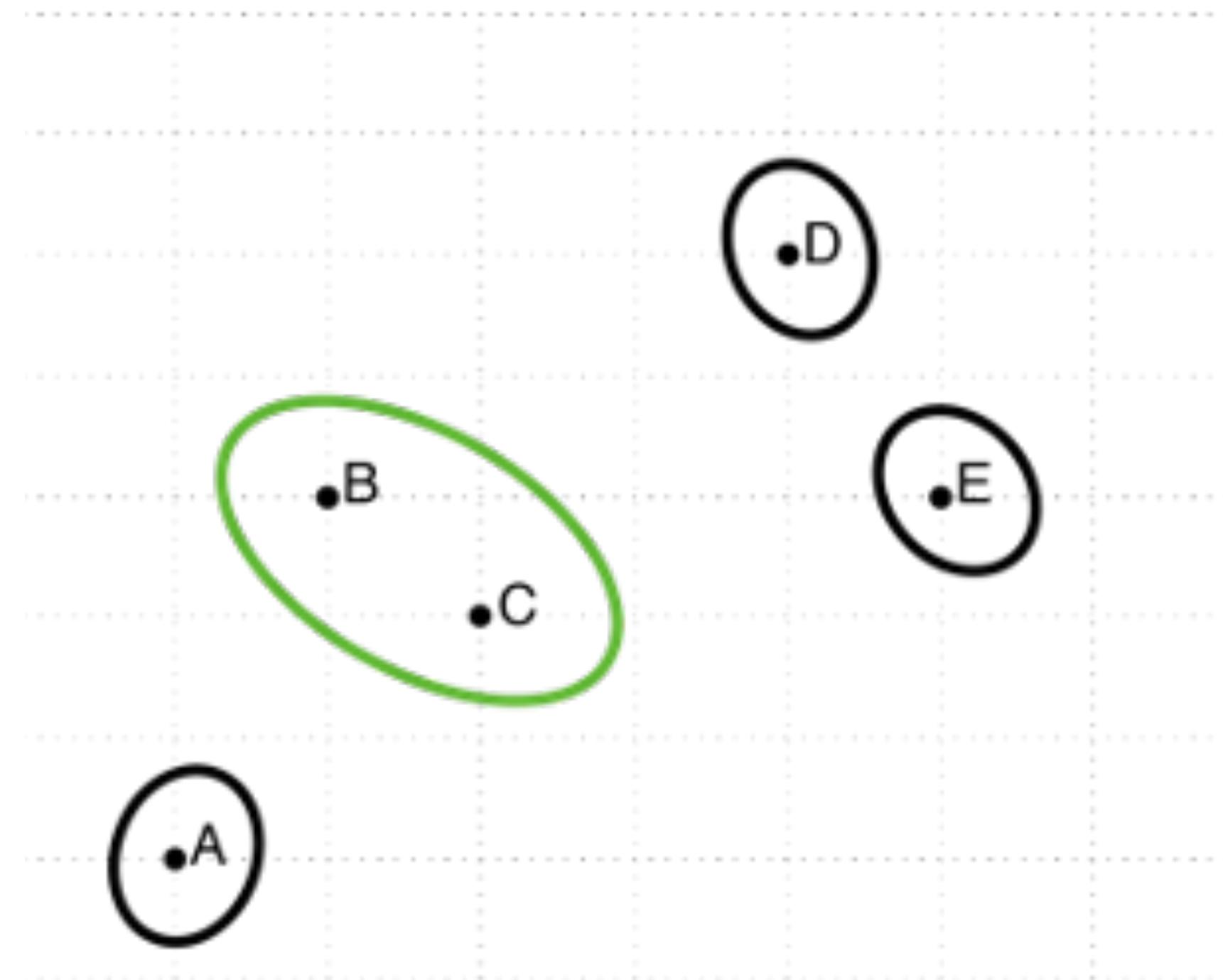


Dendrogram

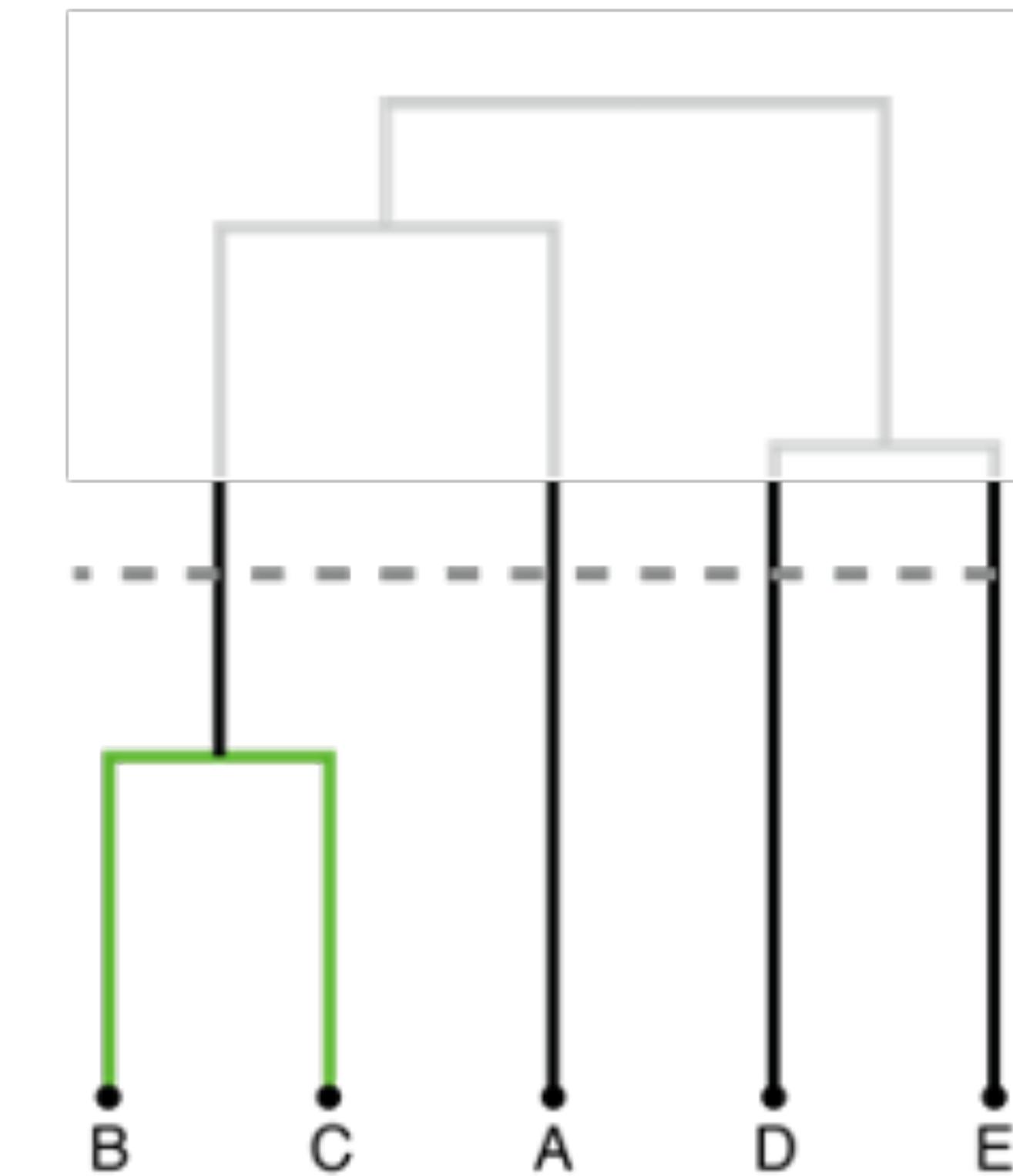


Hierarchical clustering - How to obtain clusters

Data set

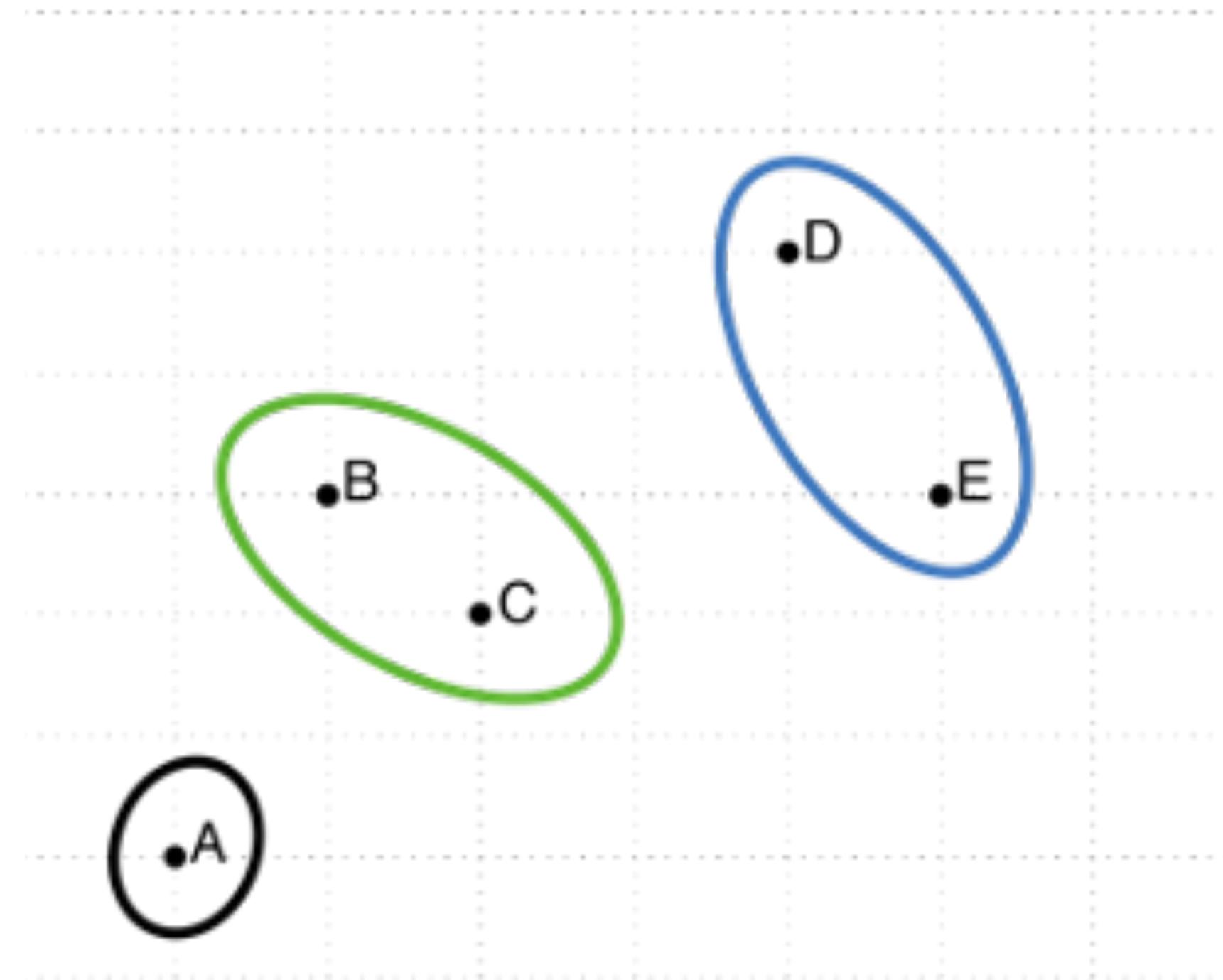


Dendrogram

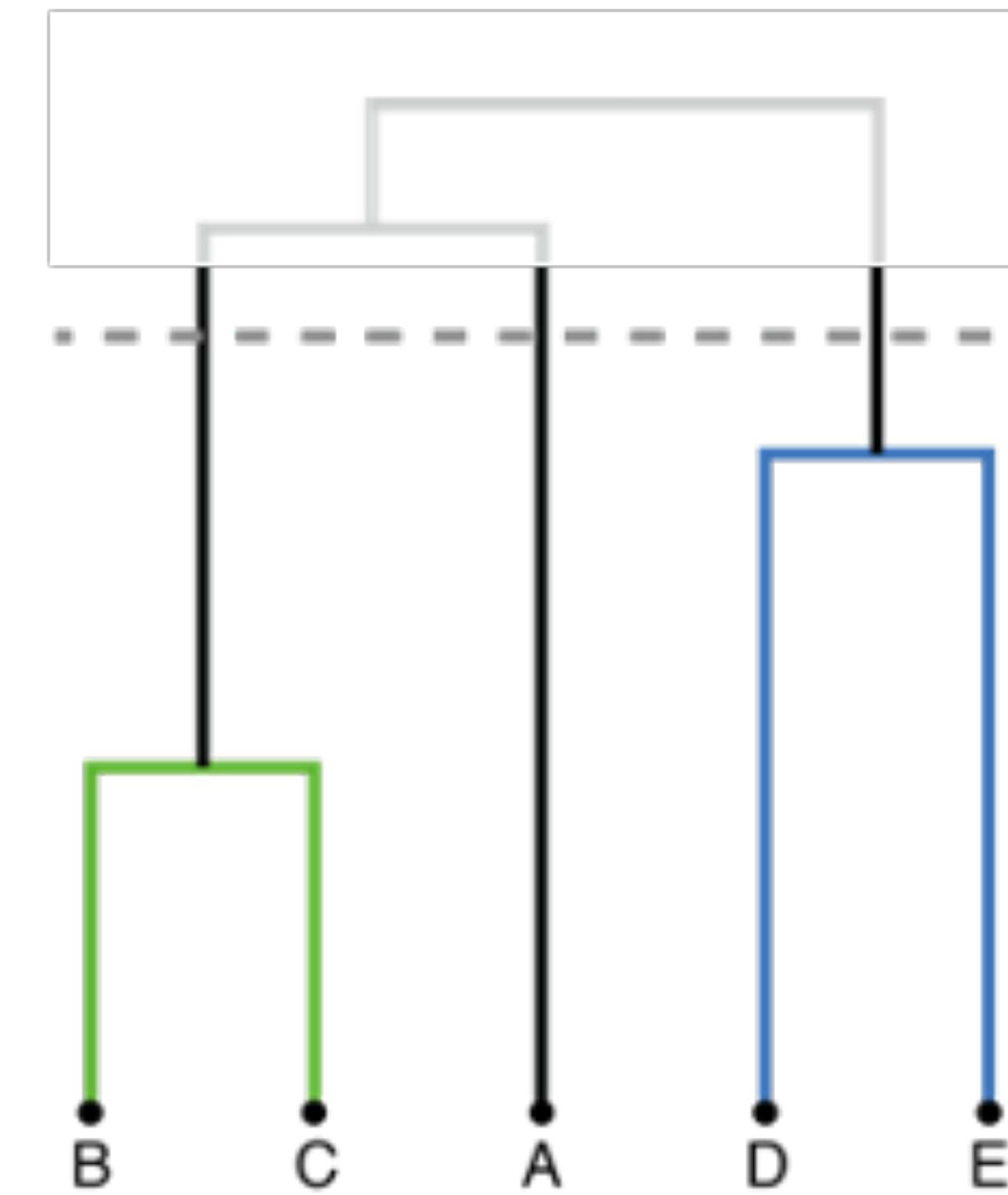


Hierarchical clustering - How to obtain clusters

Data set

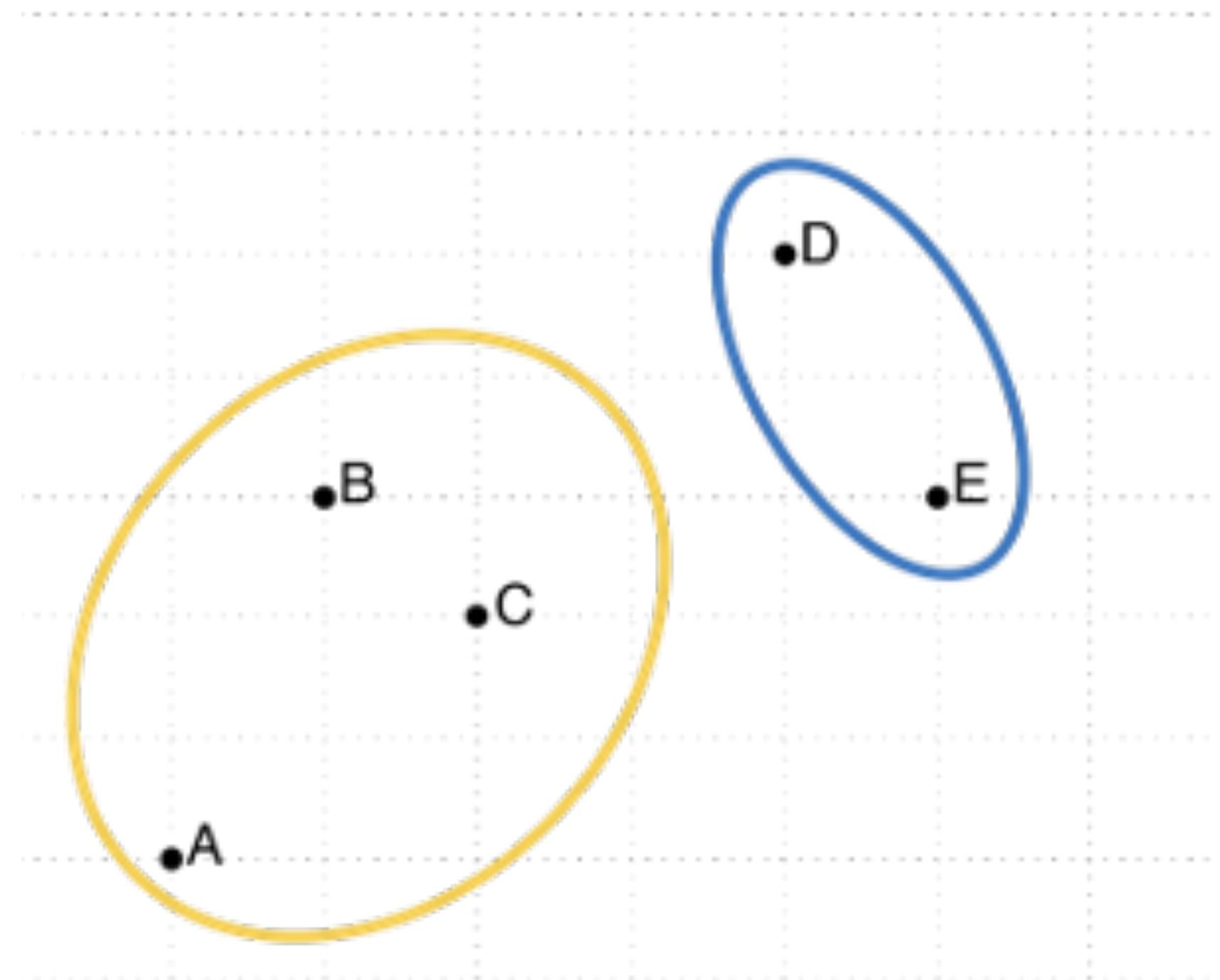


Dendrogram

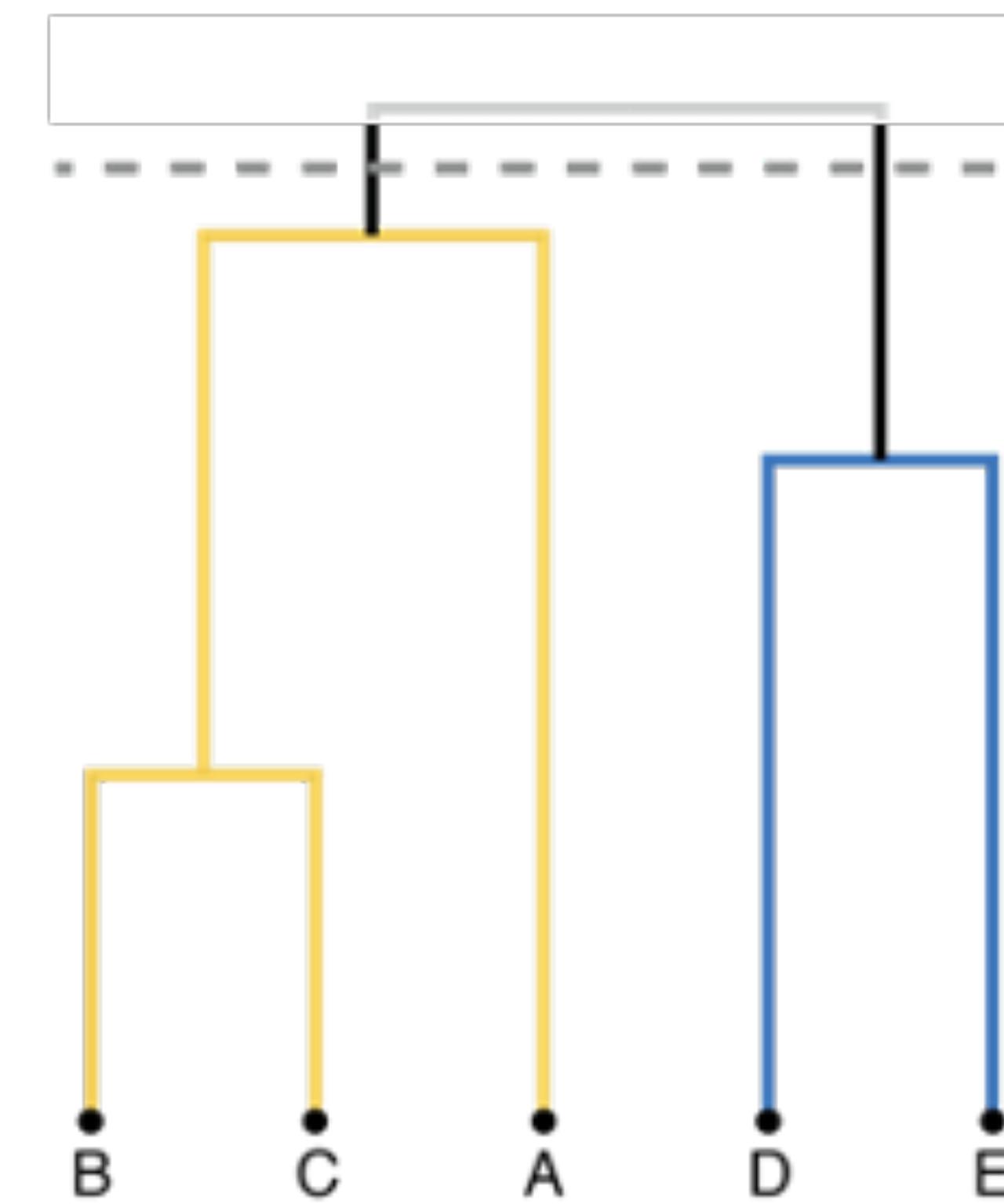


Hierarchical clustering - How to obtain clusters

Data set

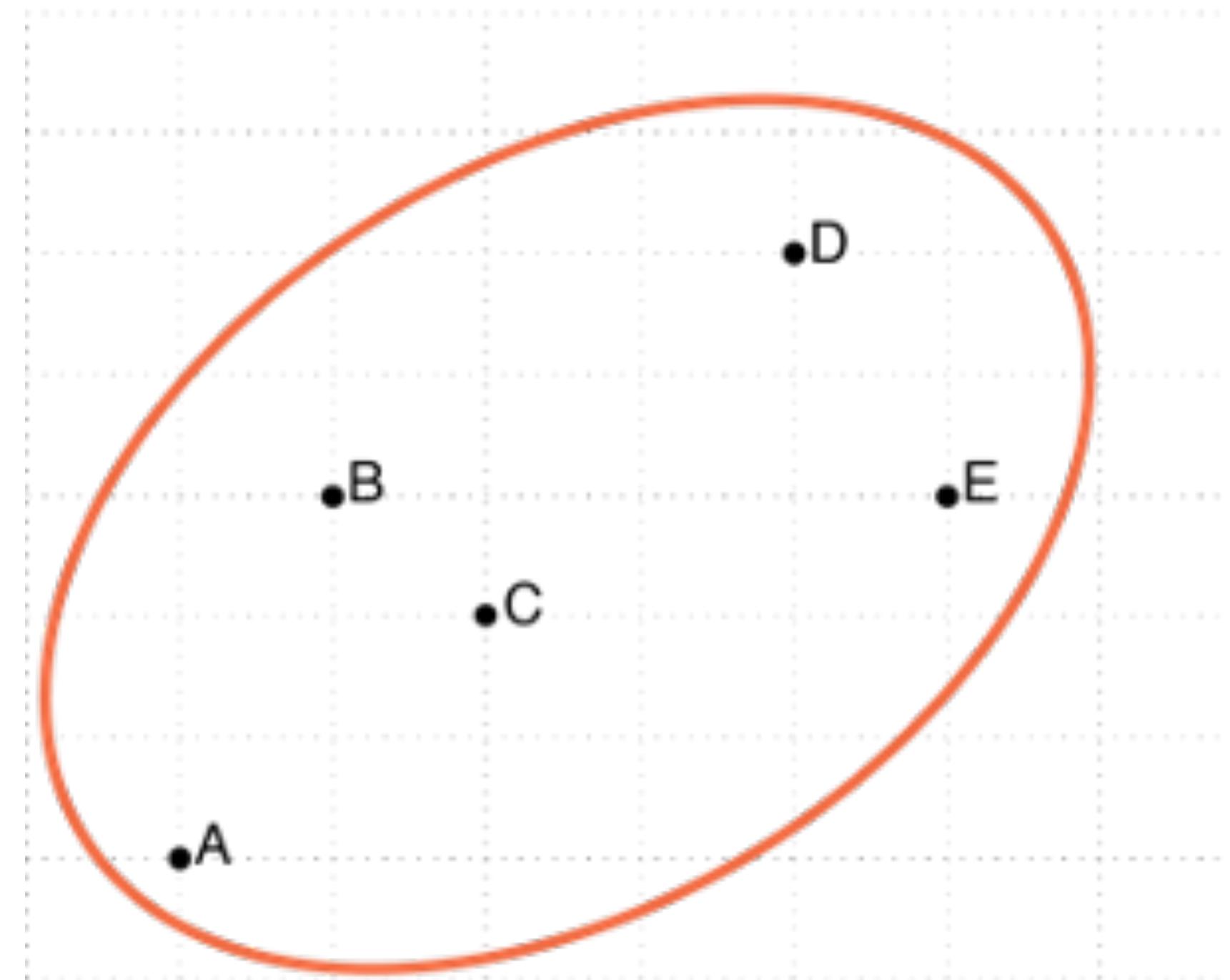


Dendrogram

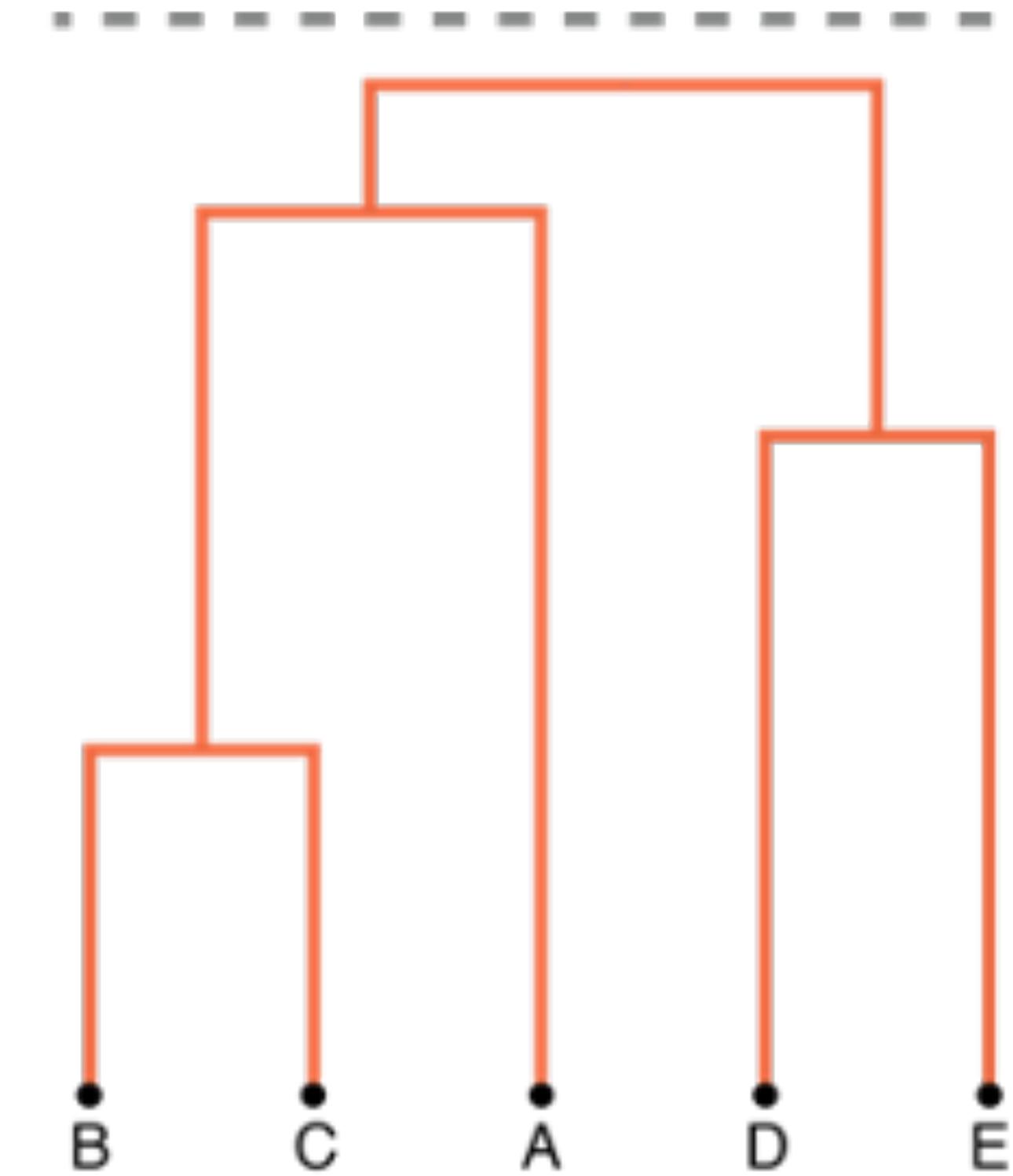


Hierarchical clustering - How to obtain clusters

Data set



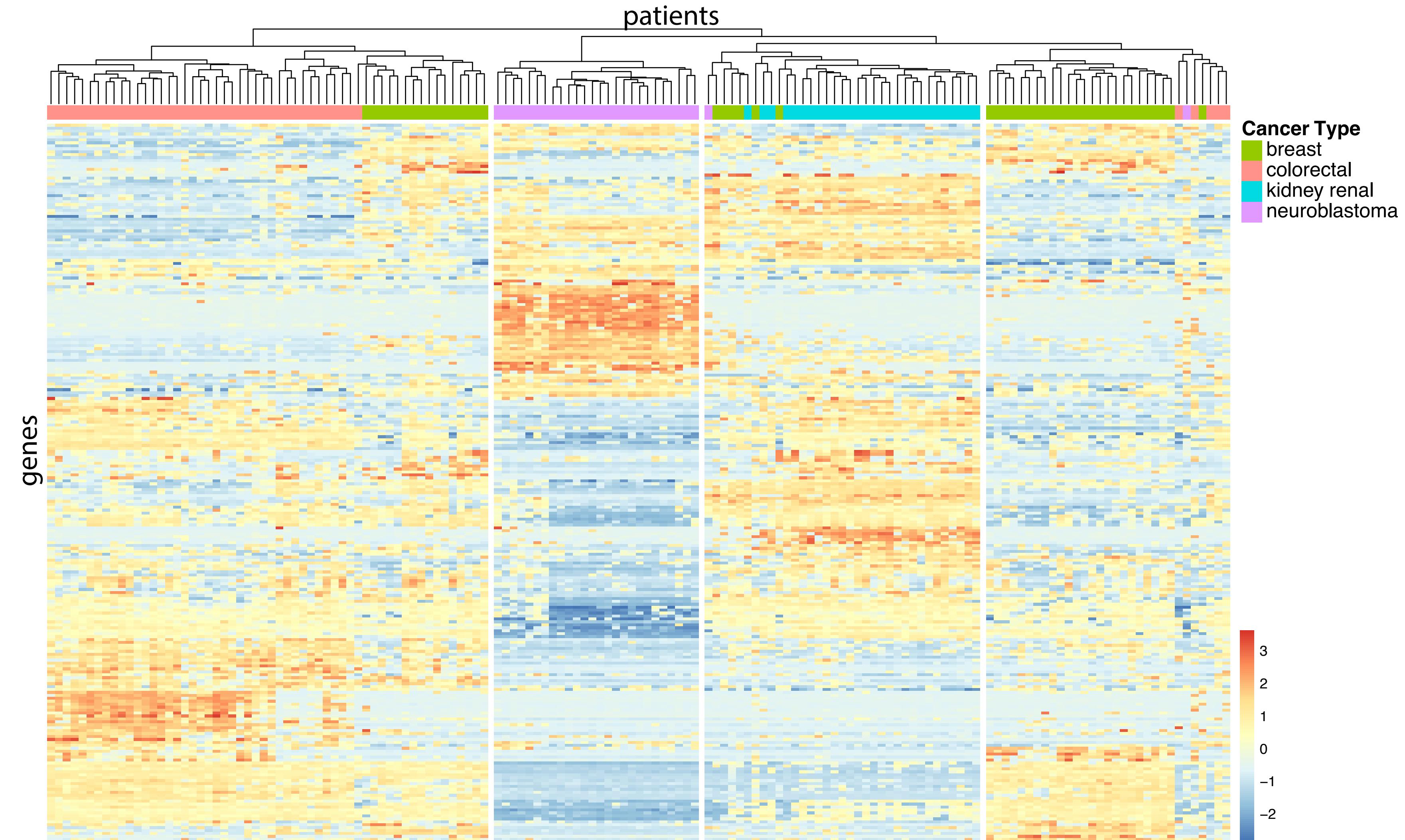
Dendrogram



Hierarchical clustering - examples with GDSC data

Linkage: Complete linkage

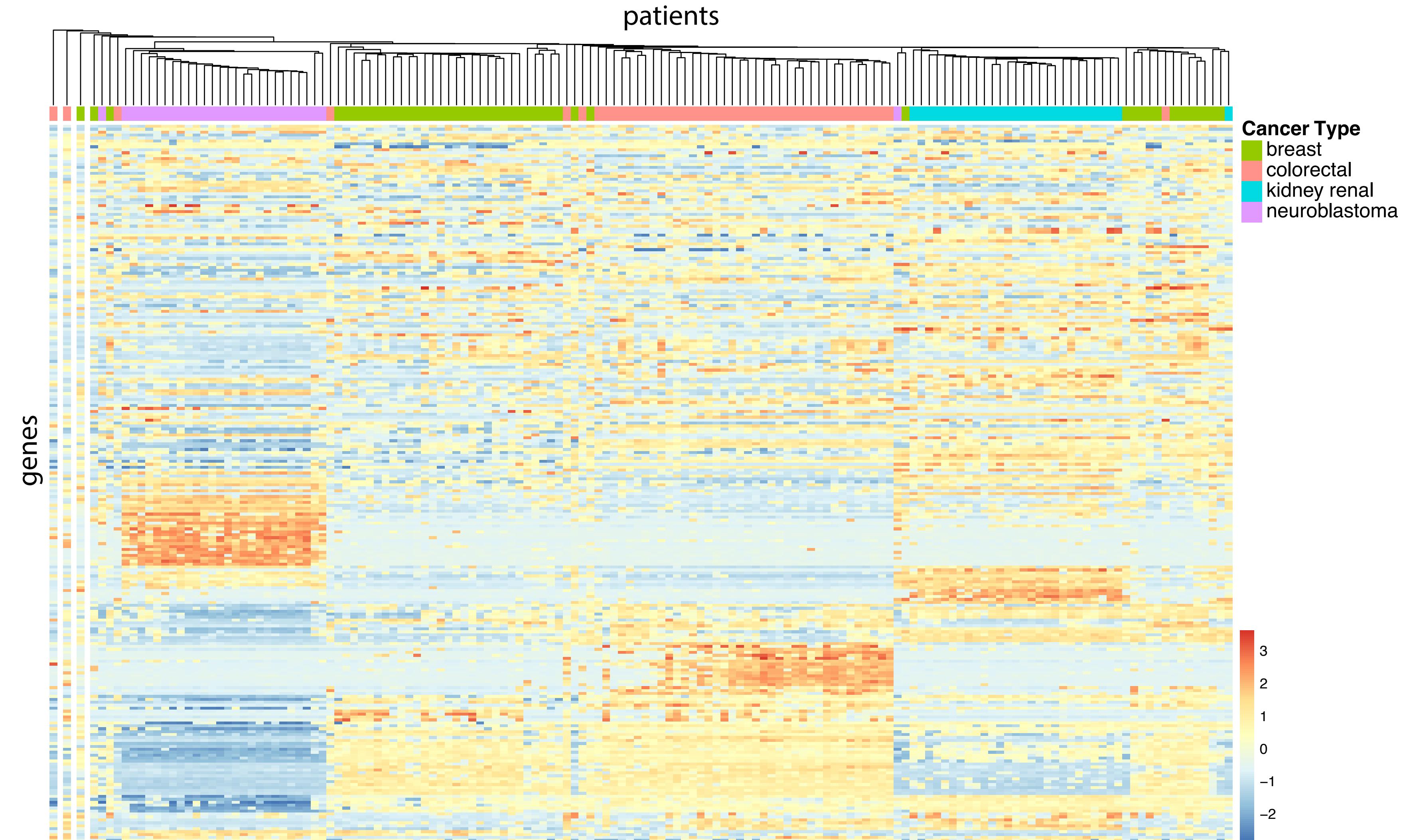
Dissimilarity metric: Euclidean distance



Hierarchical clustering - examples with GDSC data

Linkage: Single linkage

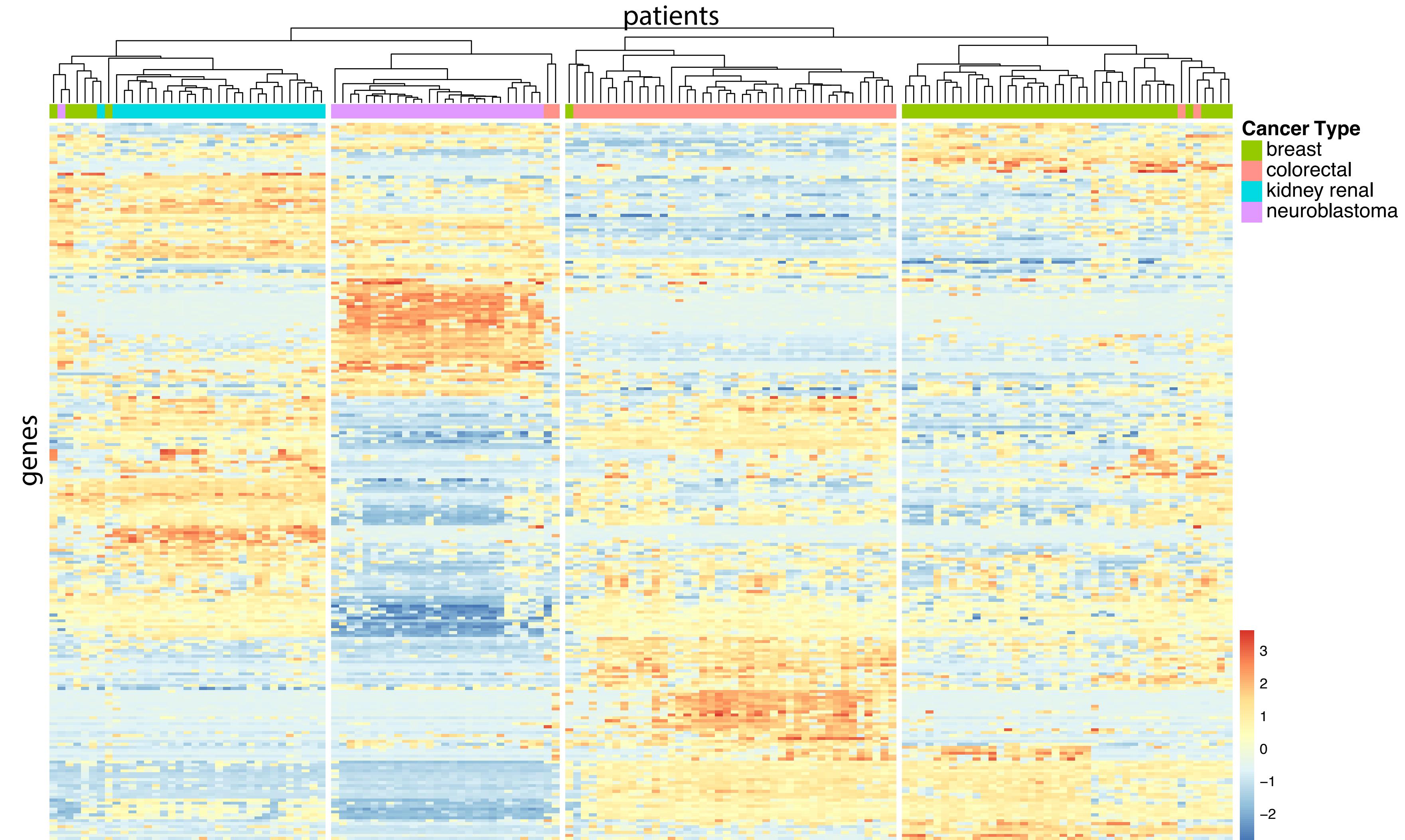
Dissimilarity metric: Euclidean distance



Hierarchical clustering - examples with GDSC data

Linkage: Complete linkage

Dissimilarity metric: Correlation



Summary of clustering

- ▶ **Goal:** Group data points based on similarity.
- ▶ **Key techniques:**
 - ▶ **K-means:**
 - ▶ Partition data into K clusters by minimising within-cluster variation.
 - ▶ Requires predefining the number of clusters.
 - ▶ **Hierarchical Clustering (bottom-up):**
 - ▶ Starts with each point as its own cluster and merges them step by step.
 - ▶ Produces a dendrogram that shows the merging process.
- ▶ **Examples of biomedical Applications:**
 - ▶ Grouping patients based on clinical measurements to identify subgroups with similar conditions.
 - ▶ Clustering genes based on expression patterns to reveal functional relationships.

Questions?