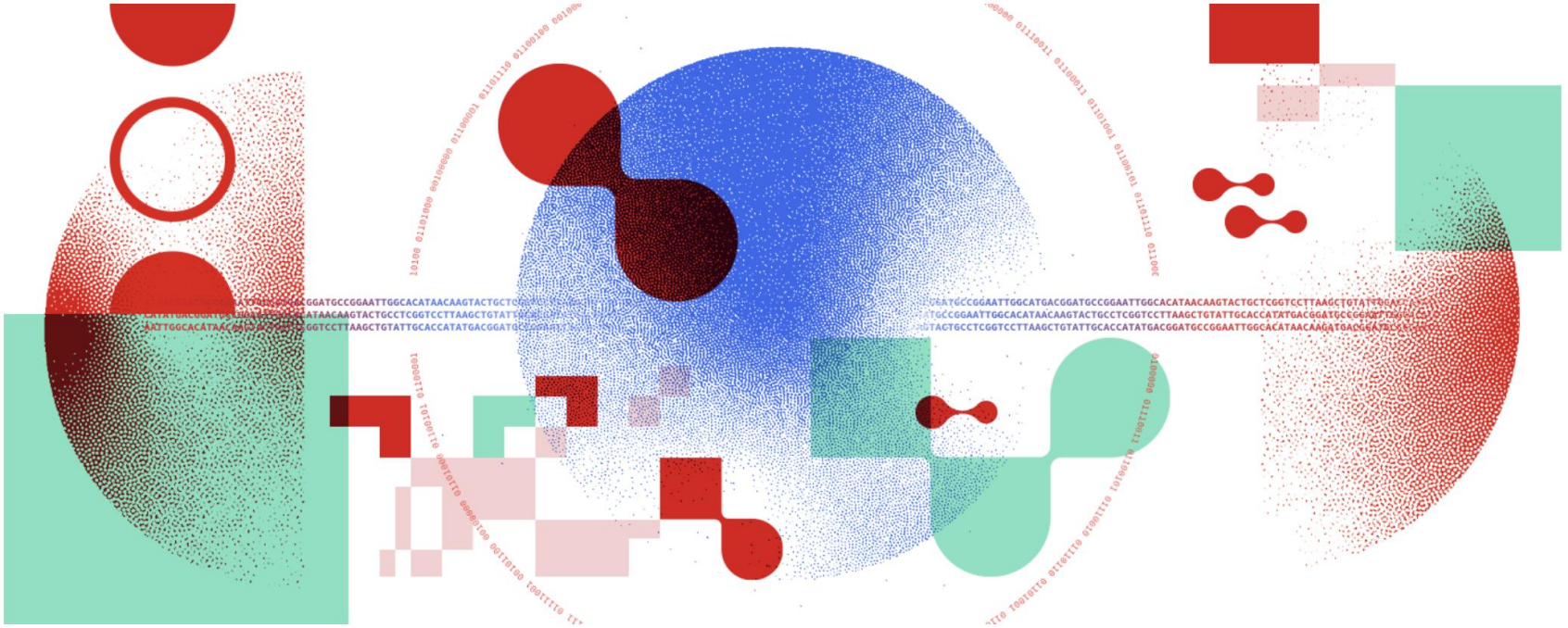


Introduction to Statistics and Data Visualisation with R

Lausanne, January 2026

Joao Lourenço and Rachel Marcone

Clustering, Heatmap and PCA

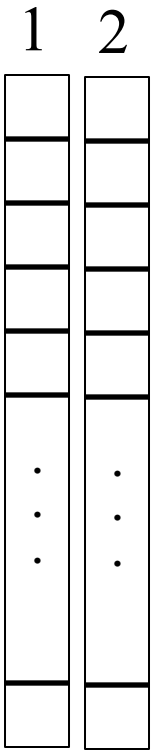
Visual representation

Dimension

Dimension:
the number of coordinates we need to locate a point in a given space.

2-dimension

	Coordinate1	Coordinate2
x1	1	2
x2	1	4
…	…	…
xN	5	2



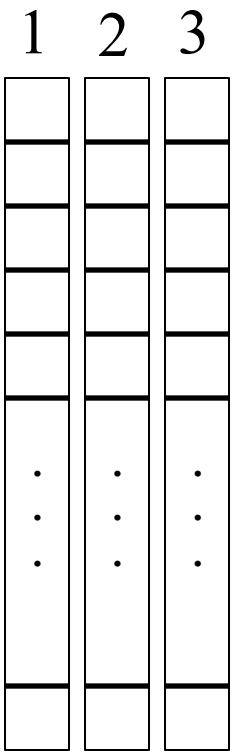
Two dimensions: latitude and longitude

Latitude

Longitude

3-dimension

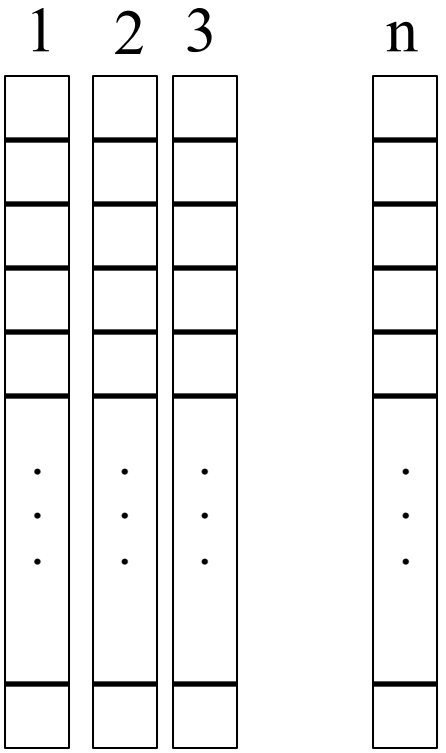
	Coordinate1	Coordinate2	Coordinate3
x1	1	2	5
x2	1	4	7
…	…	…	…
xN	5	2	1



Three dimensions: latitude, longitude and altitude

n-dimension

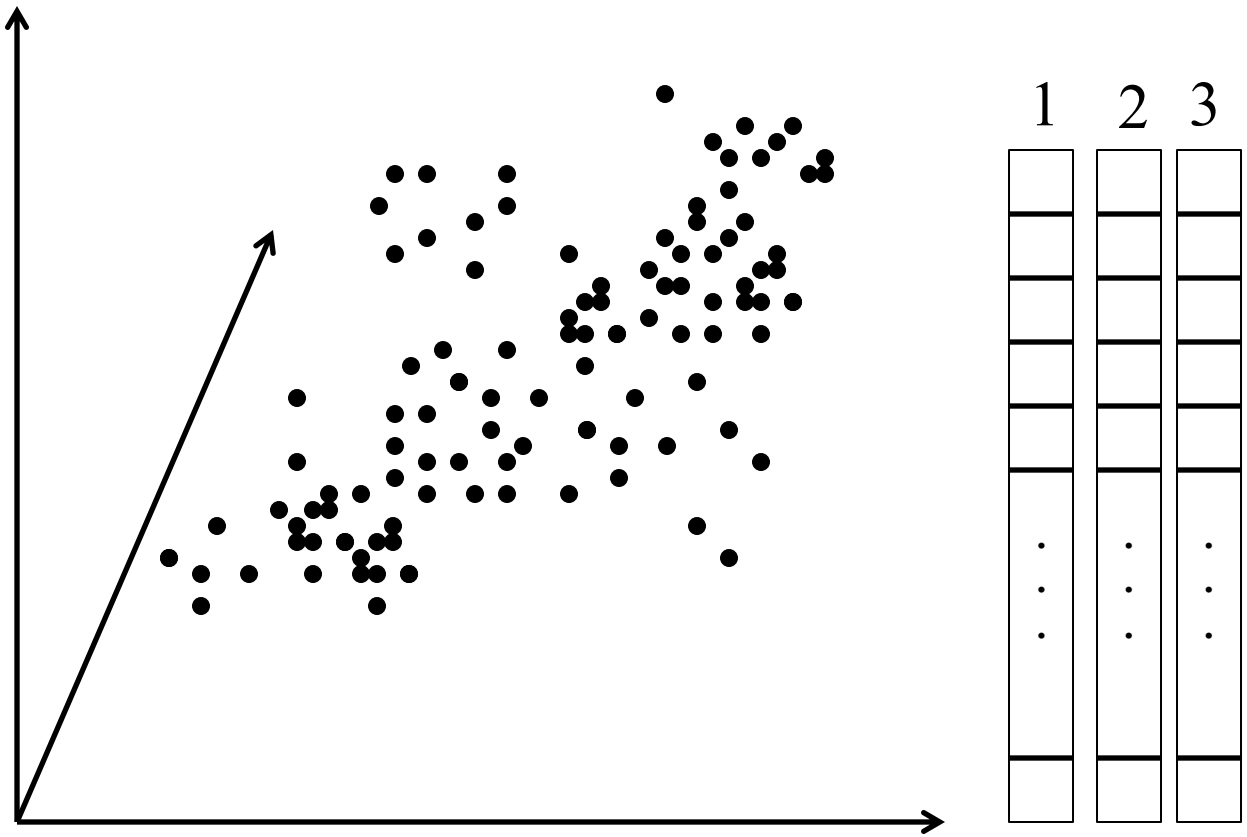
	Coordinate1	Coordinate2	Coordinate3	…	Coordinate-n
x1	1	2	5	…	18
x2	1	4	7	…	2
…	…	…	…	…	…
xN	5	2	1	…	4



Dimension in biology?

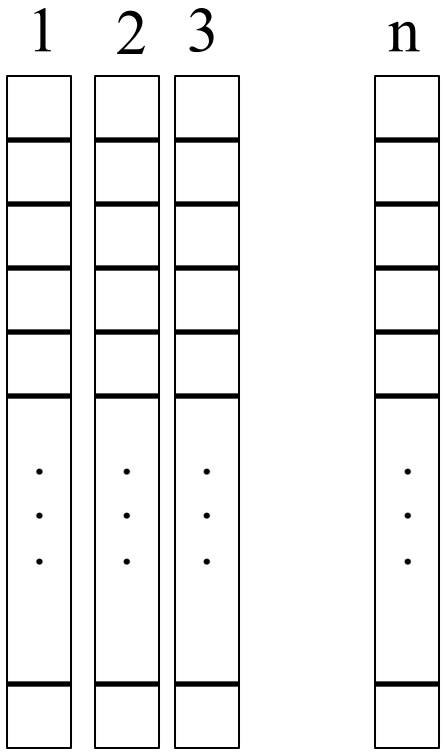
Genes

3-dimension



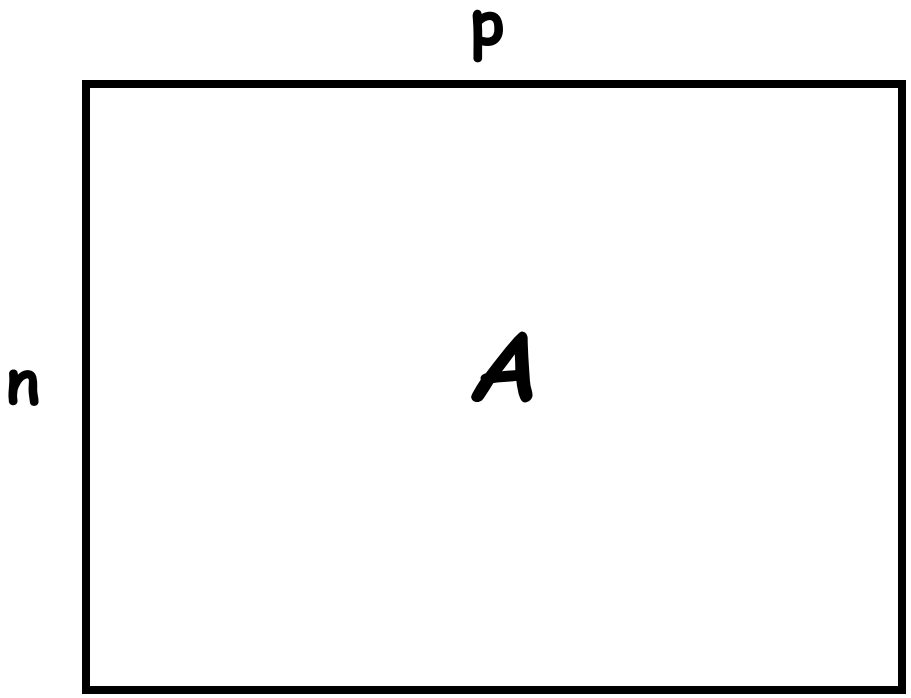
n-dimension

?

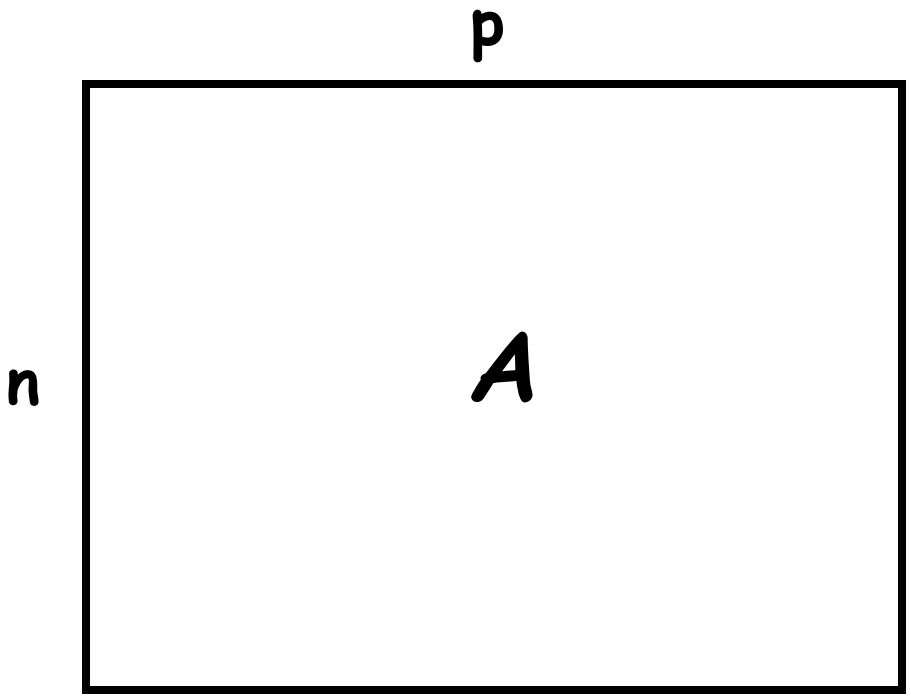


Dimension reduction

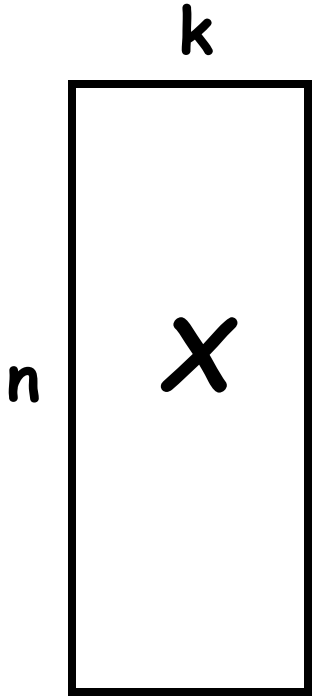
Starting point: Big Data

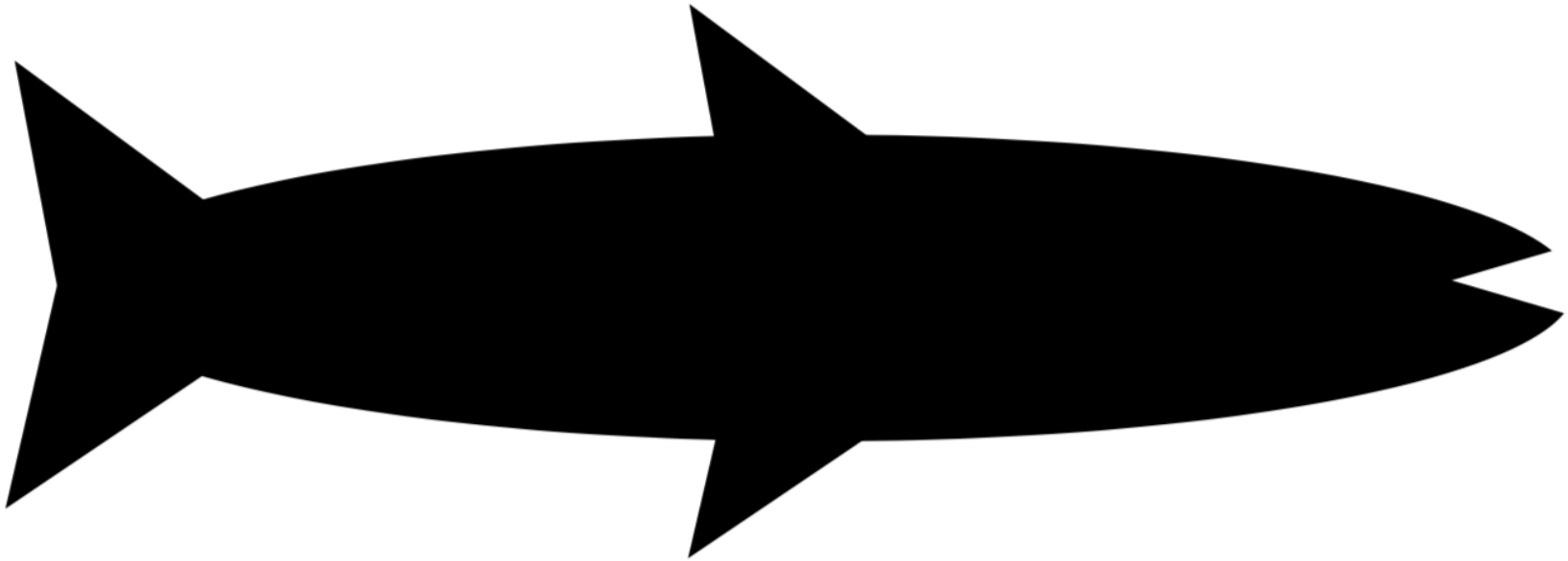


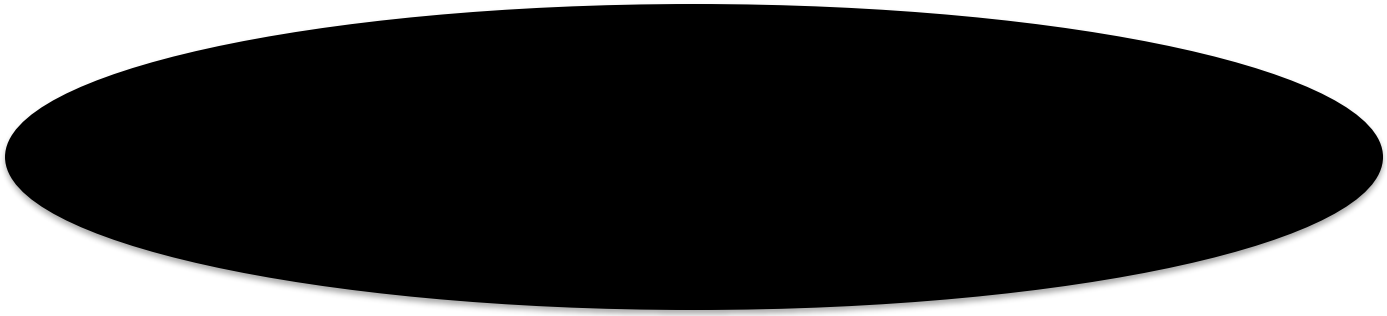
Starting point: Big Data



End result: human readable





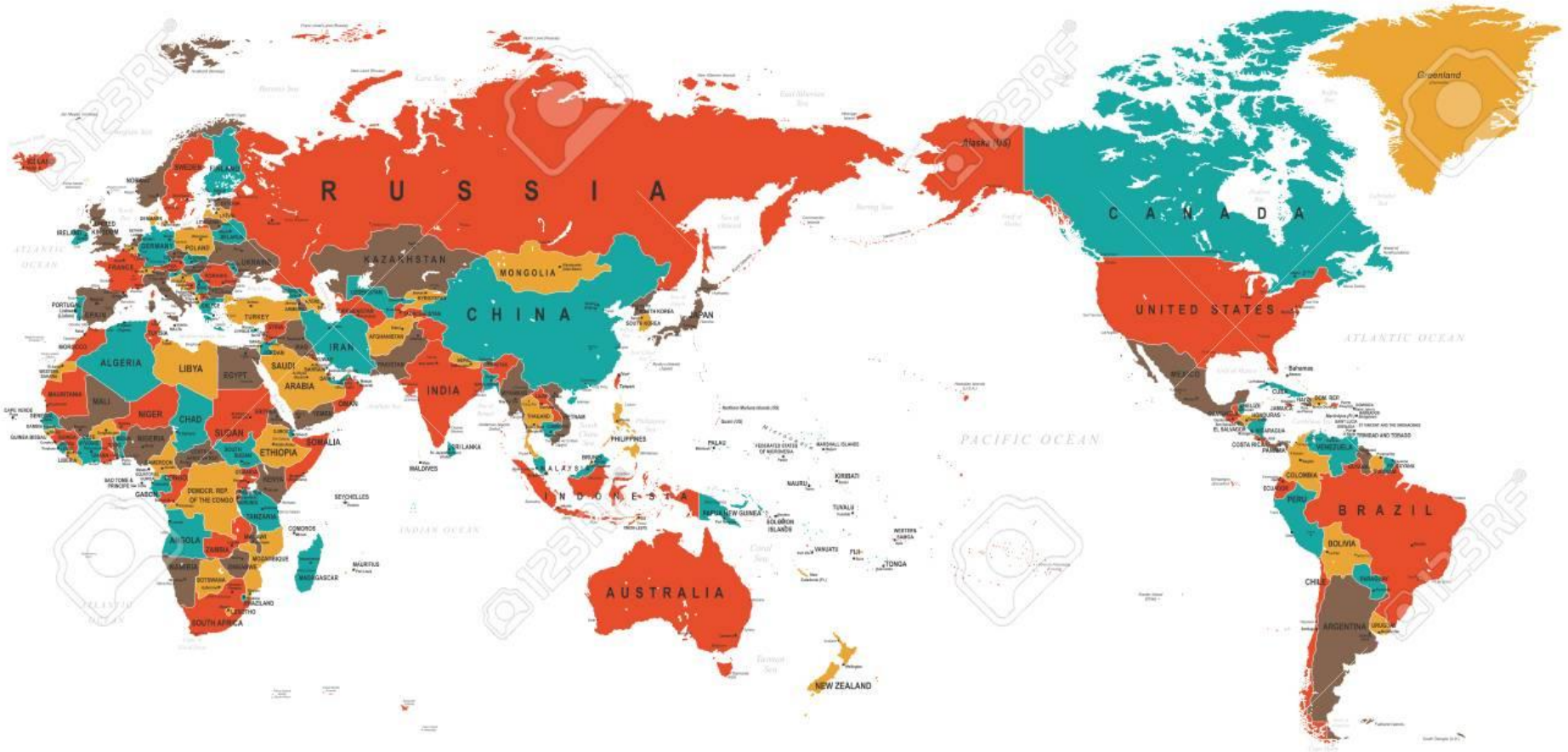


clarity of representation

Over-simplification

There are many possibilities and there is not a « better » one than another. It depends on what you want to show.

https://ontheworldmap.com/



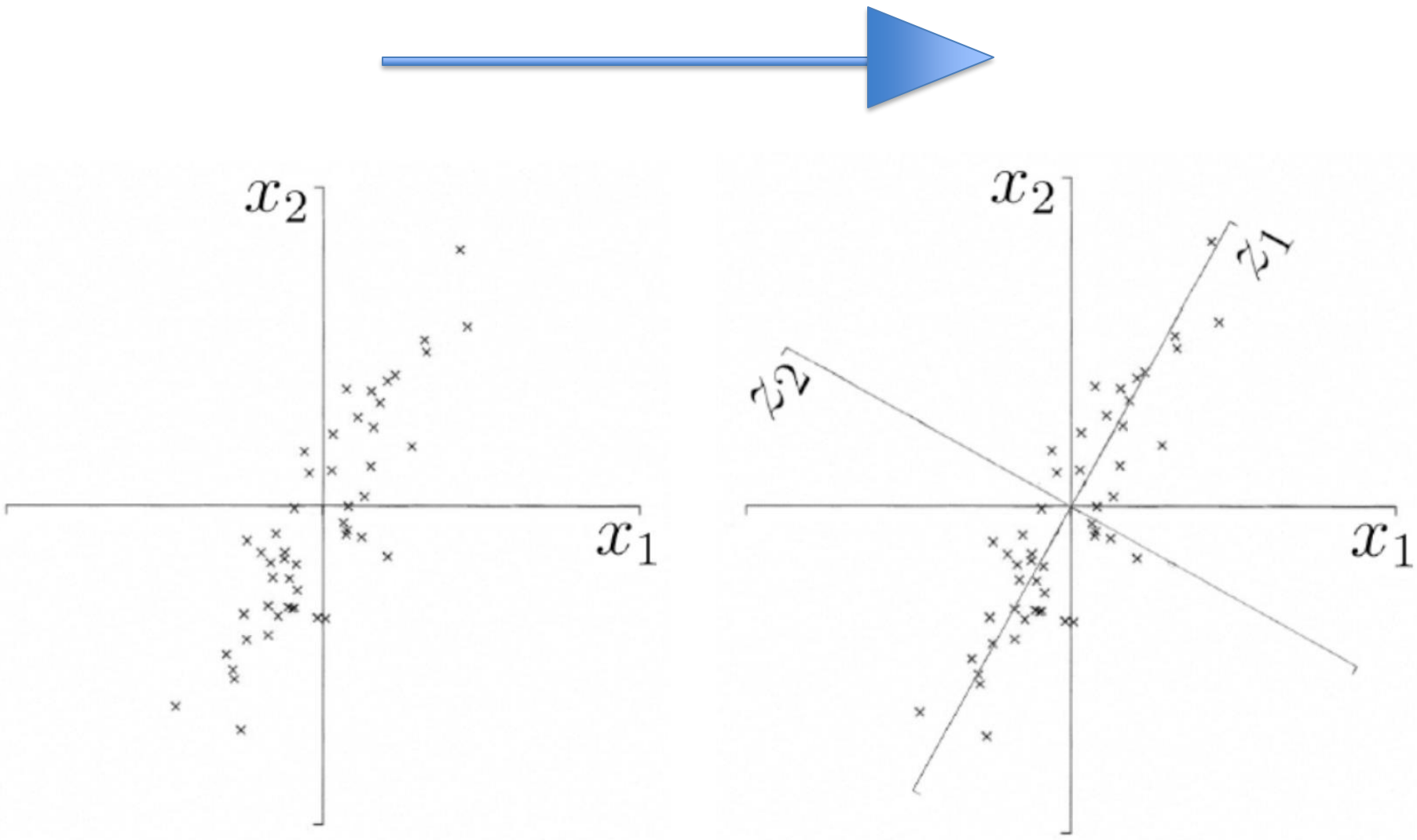
https://www.shutterstock.com/fr/image-vector/world-map-pacific-china-asia-centered-1731018682

Principal Component Analysis (PCA)

Pearson (1901) and Hotelling (1933)

-PCA is based on variance
-PCA is the best angle to see and evaluate the data

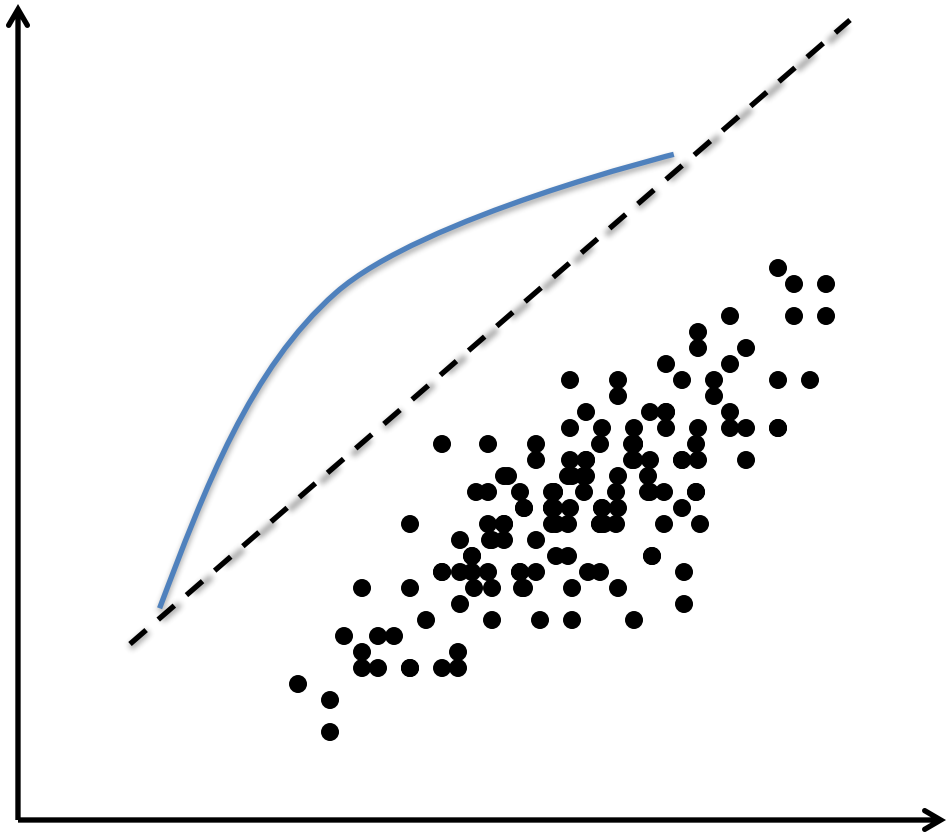
Which and how?


x_2
x_1
x_2
z_1
z_2
x_1

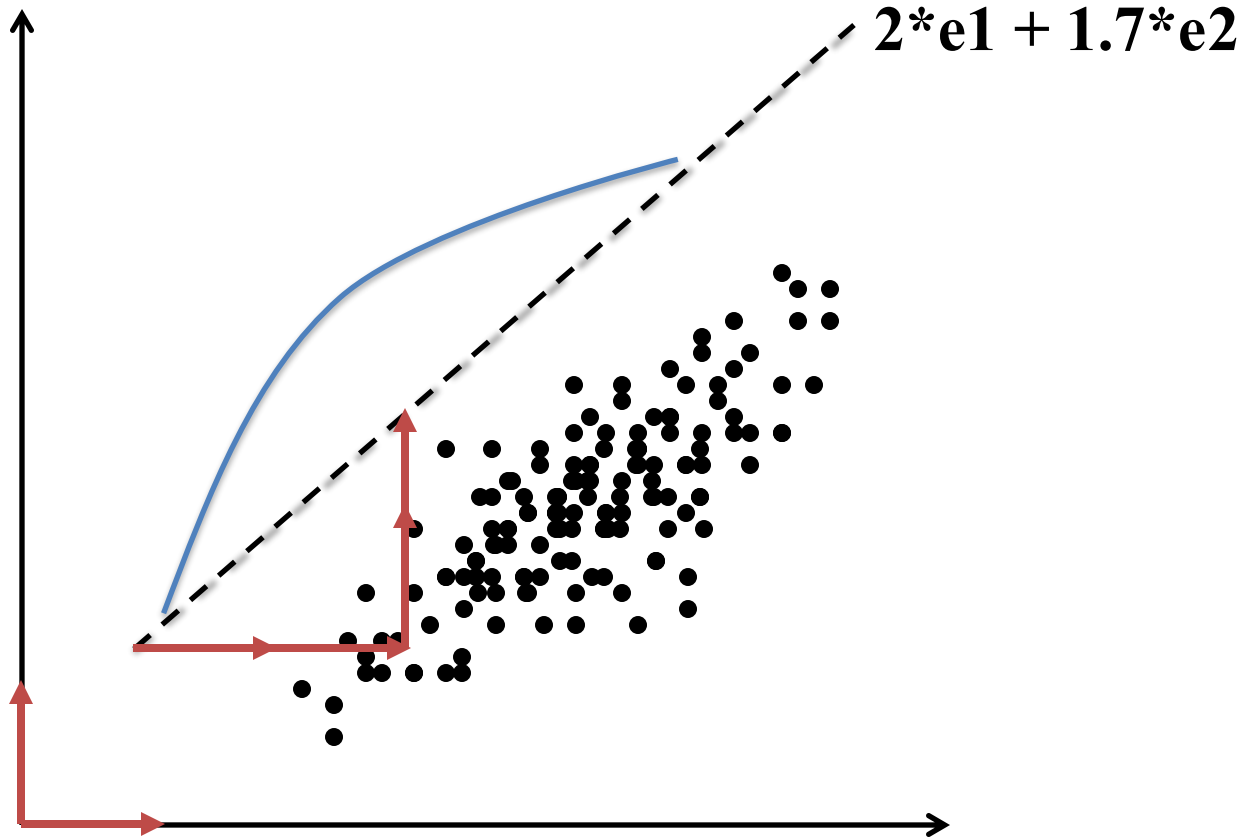
PCA- Principal component analysis

1. Largest variance first

PCA- Principal component analysis



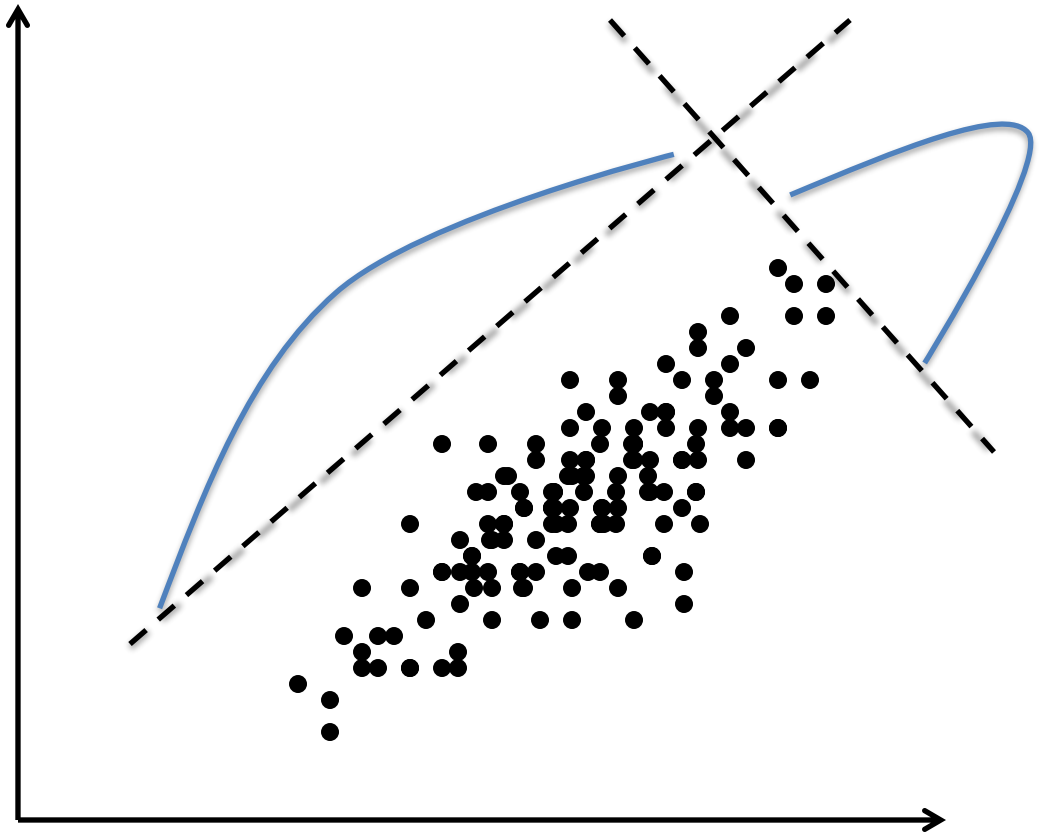
PCA- Principal component analysis



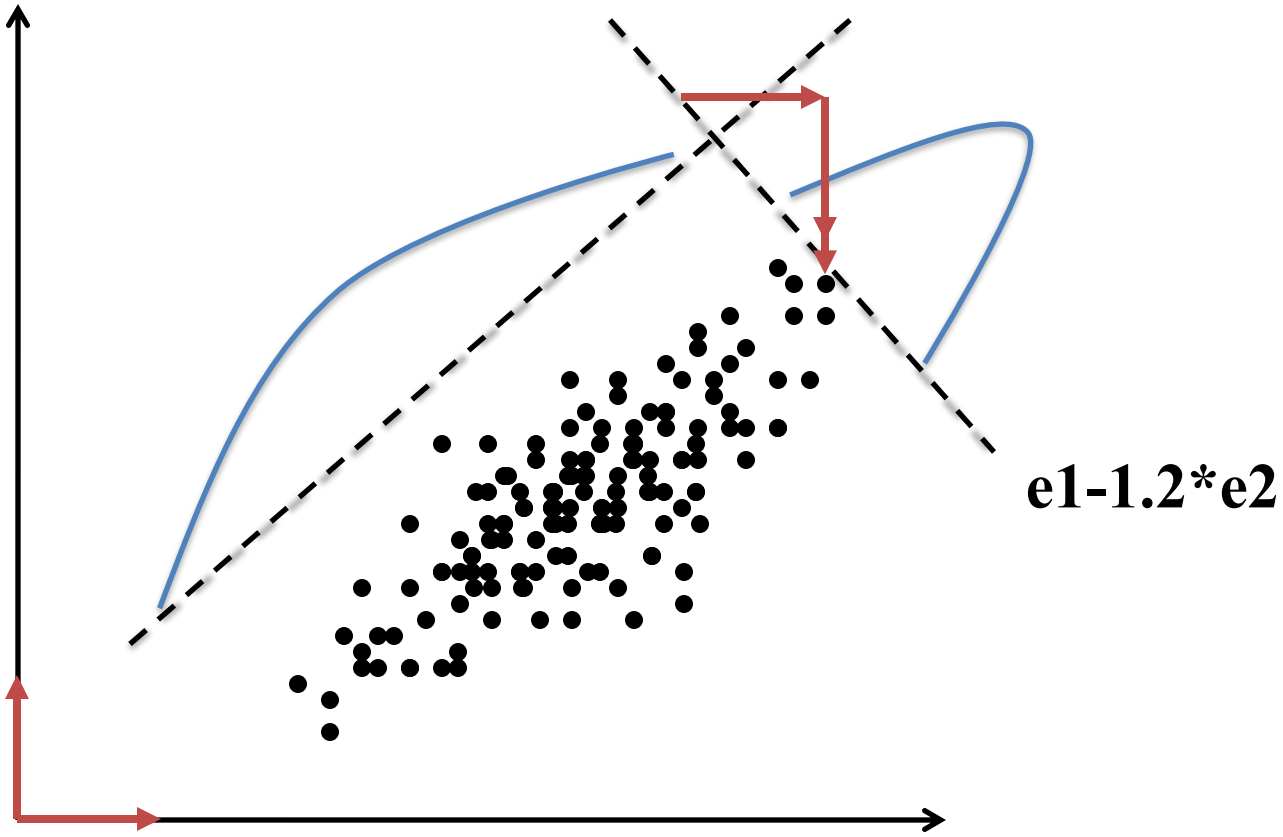
PCA- Principal Component Analysis

2. Select uncorrelated principal axis (orthogonal)

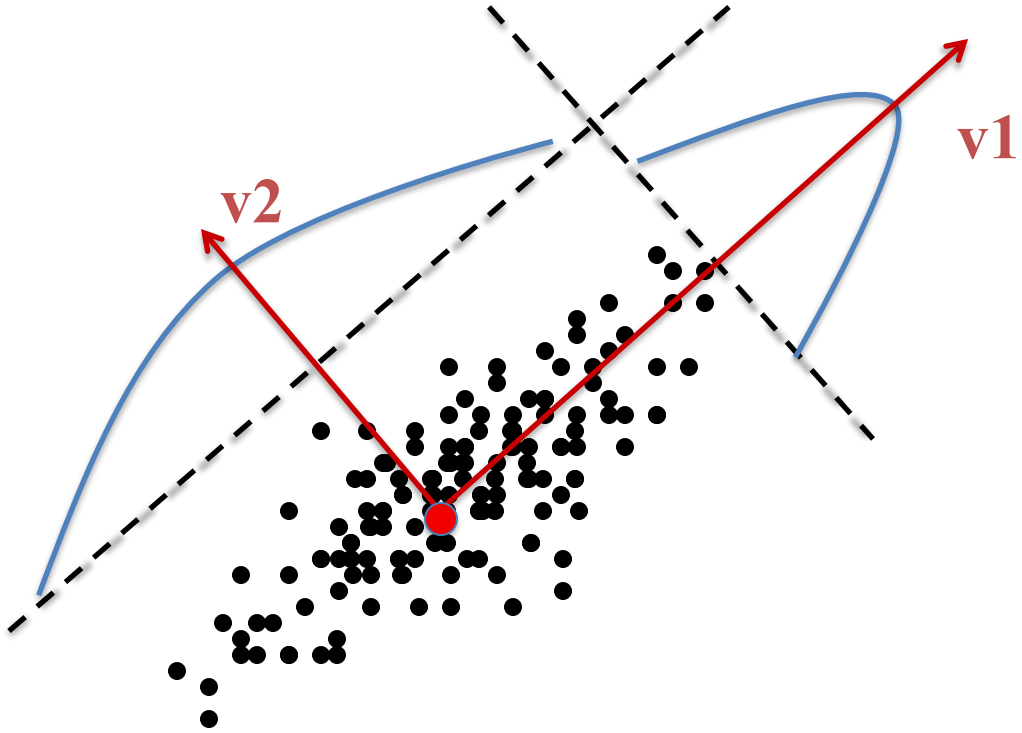
PCA- Principal Component Analysis



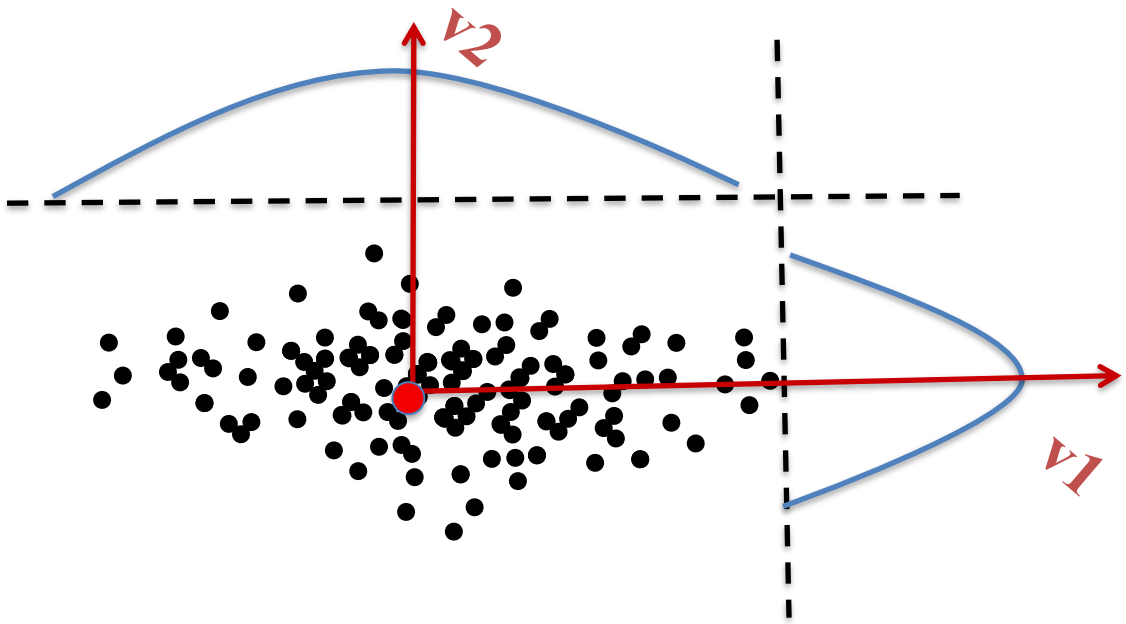
PCA- Principal Component Analysis

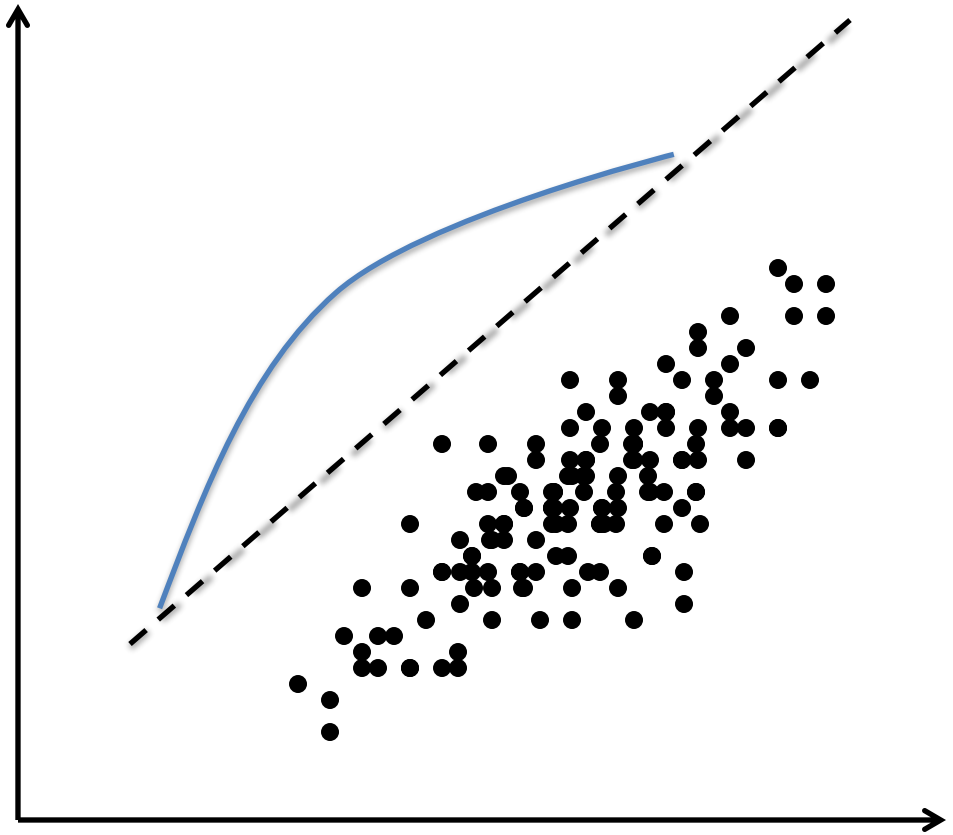


PCA- Principal Component Analysis

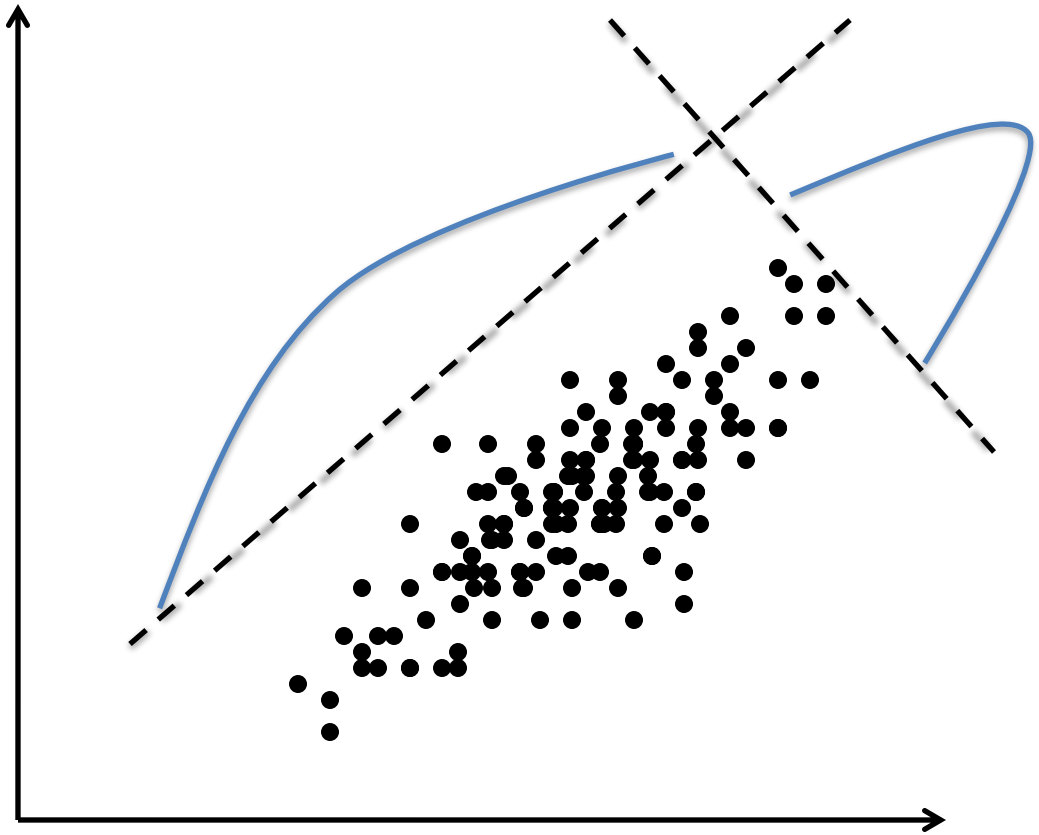


PCA- Principal Component Analysis

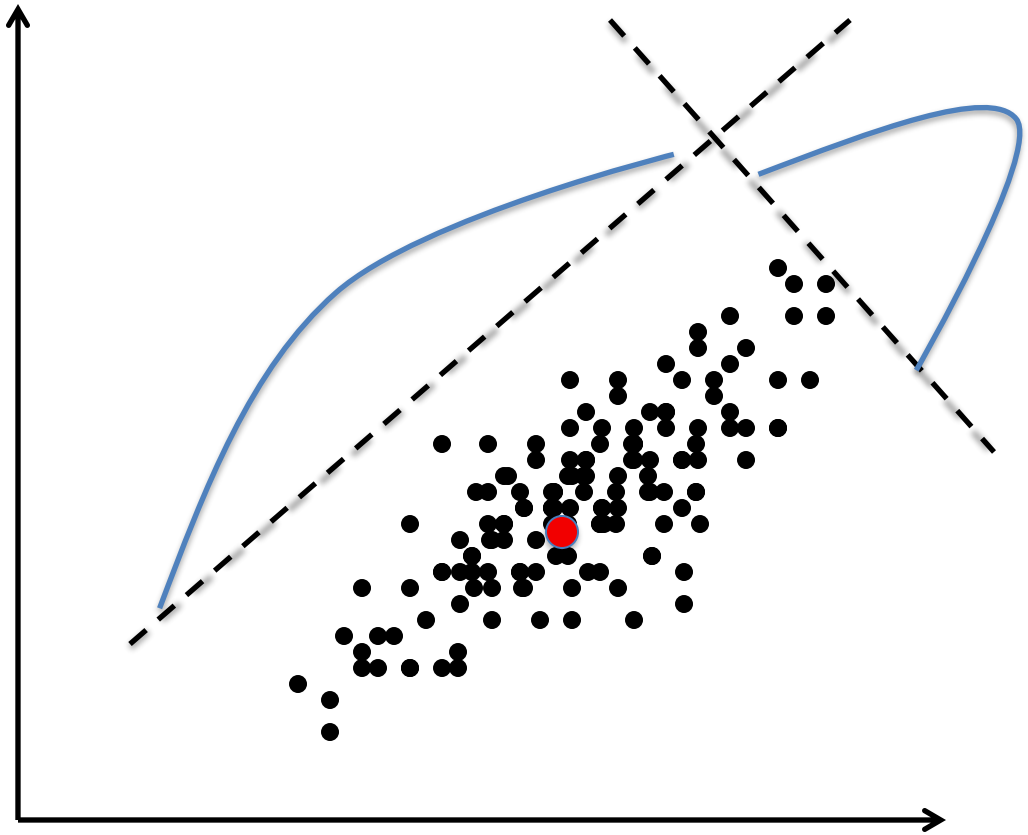


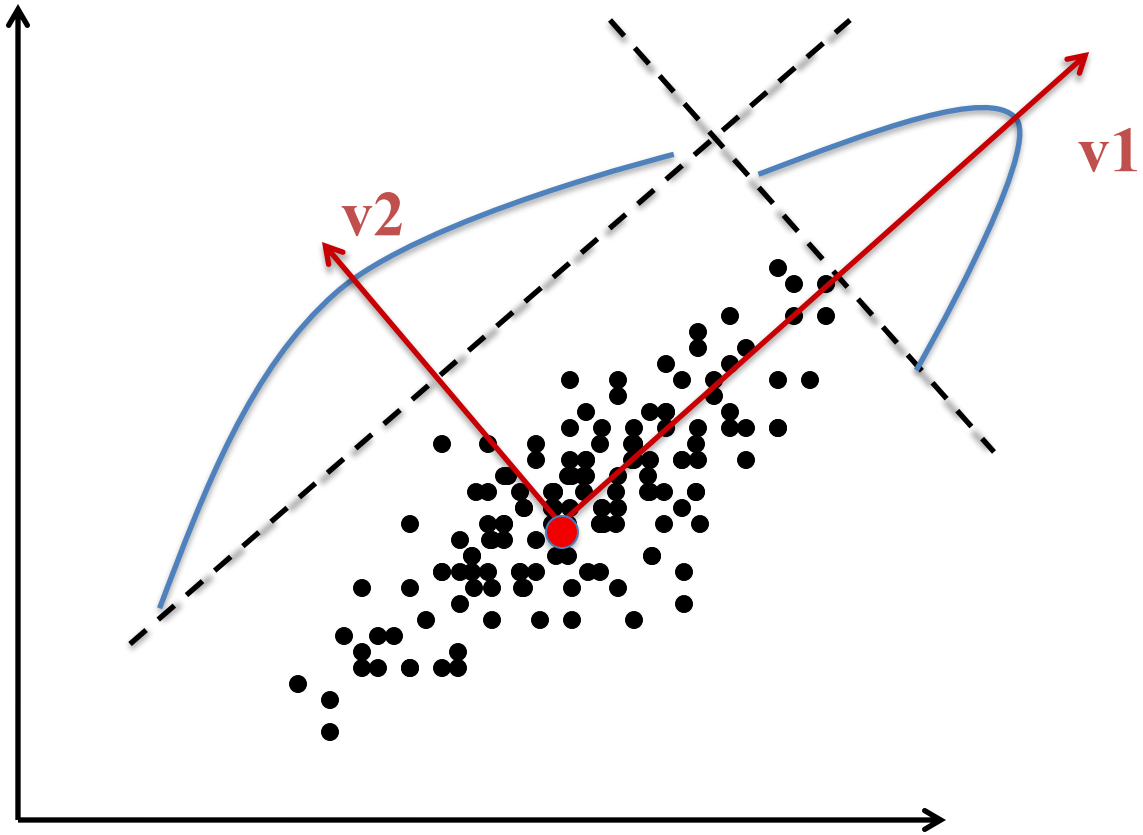


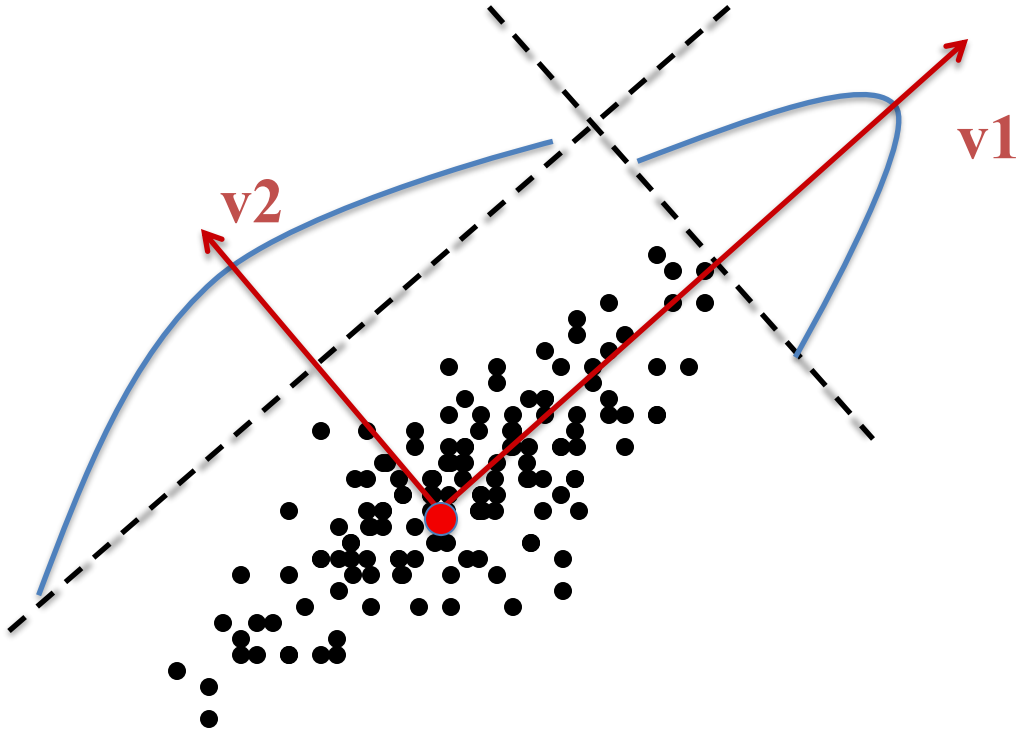
2. Select uncorrelated principal axis (orthogonal)

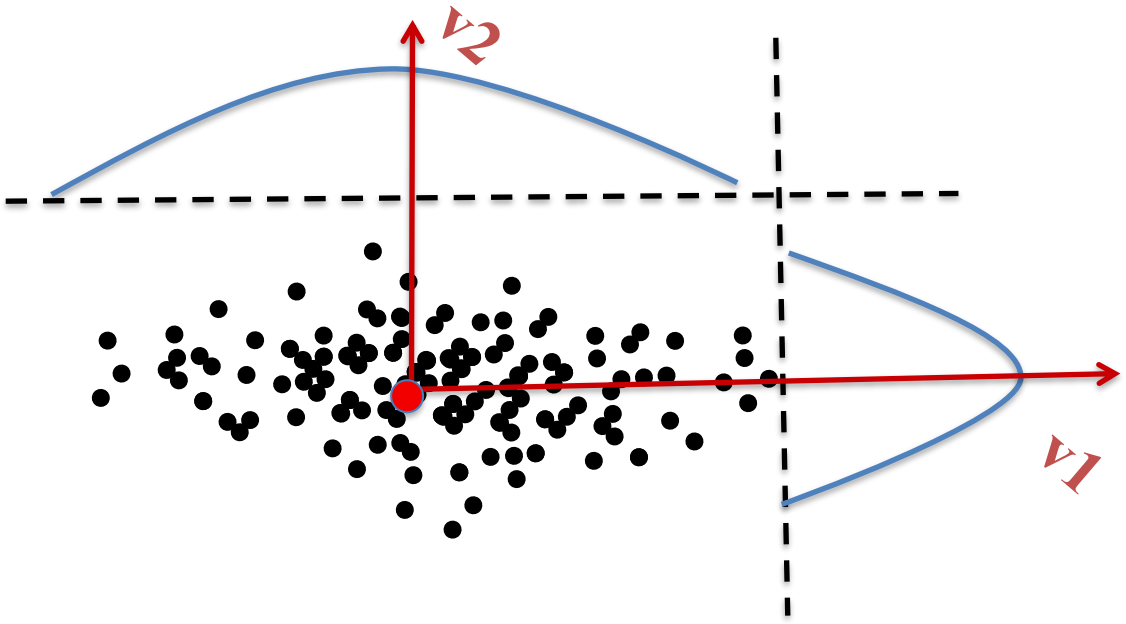


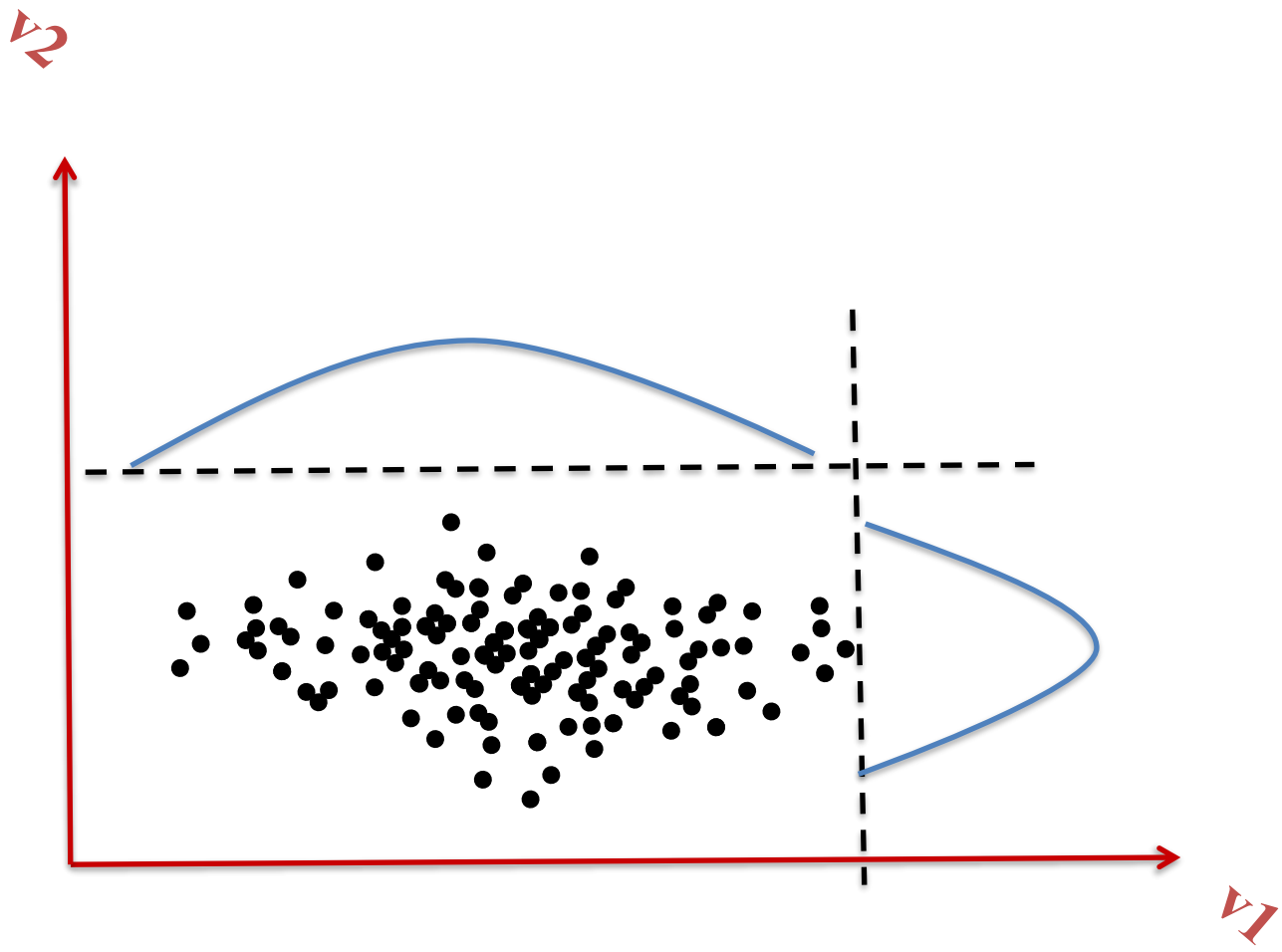
centroid



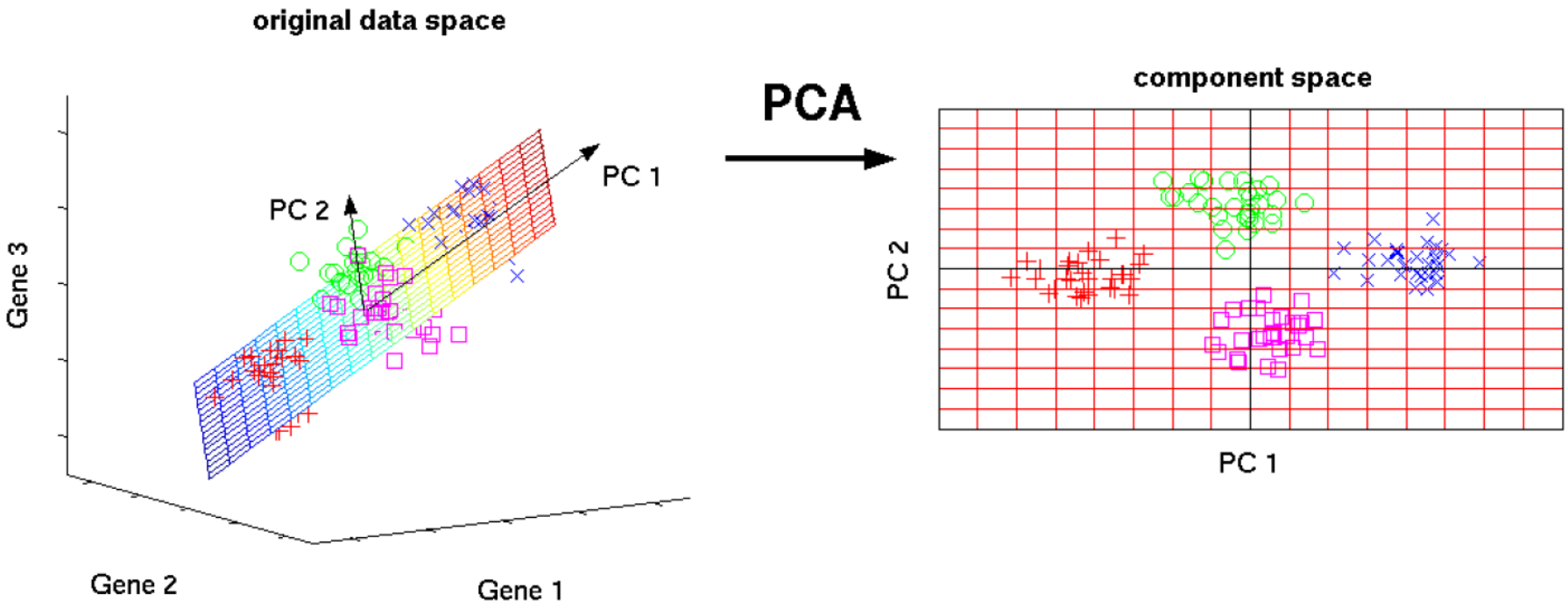

v2
v1


v2
v1


v2
v1



Without centroid


original data space
PC 2
PC 1
Gene 3
Gene 2
Gene 1
PCA
component space
PC 2
PC 1

The PCA axis

- The PC are linear combination of the original axis.
- The estimated parameters of the linear combination is known and therefore we can know positively or negatively how much it goes into one direction or the other one.
- Indeed as the original axis are g1,g2,g3… and the new axis are a1g1 +a2g2…, one takes the ai that are the highest, positively and negatively and therefore knows which features are mostly representing the axis you see.

- Observation : **Scaling** is important, if one variable is on a different scale than another, it will dominate the PCA procedure as the largest variance might be observed there, and the low dimension plot will really just be visualizing that dimension.

Mathematically

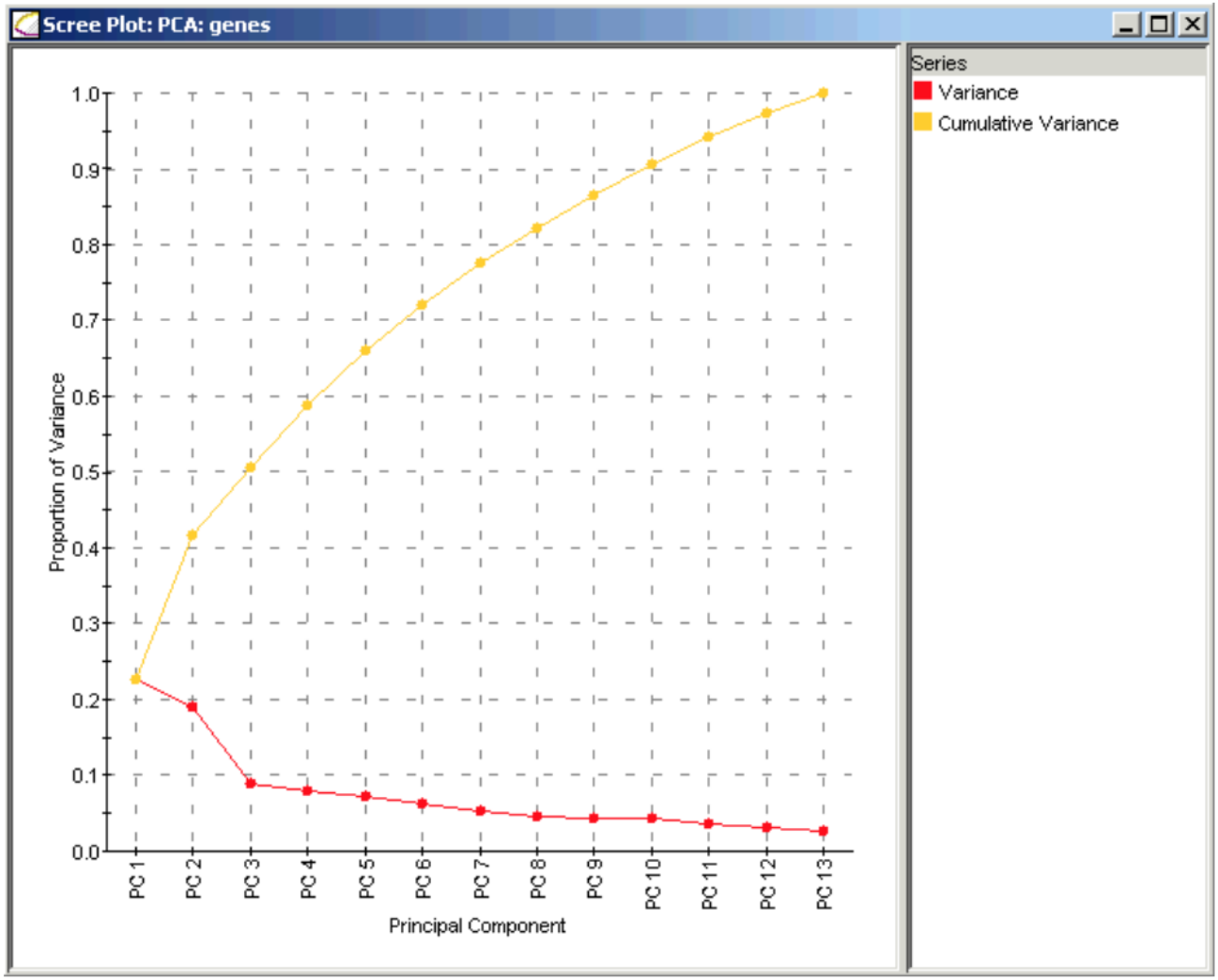
You calculate the covariance matrix meaning a matrix containing two-by-two covariances

- if positive then : the two variables increase or decrease together (correlated)
- if negative then : One increases when the other decreases (Inversely correlated)

- (Corr(X,Y) = Cov(X,Y)/sd(x)*sd(y))
- Eigenvectors and eigenvalues are the linear algebra concepts that we need to compute from the covariance matrix in order to determine the ***principal components*** of the data.

Mathematically

- eigenvectors of the Covariance matrix
  are *the directions of the axes where there is the most variance (this is something you can prove mathematically!)*
- eigenvalues are the coefficients attached to eigenvectors, which give the *amount of variance carried in each Principal Component*.
- After having the principal components, to compute the percentage of variance (information) accounted for by each component, we divide the eigenvalue of each component by the sum of eigenvalues.



Scree Plot for Genetic Data. (Source.)

https://towardsdatascience.com/a-one-stop-shop-for-principal-component-analysis-5582fb7e0a9c

How many PCs?

- Method 1: We **arbitrarily select** a number of principal components to include. Suppose I wanted to keep five principal components in my model. In the genetic data case above, these five principal components explains about 66% of the total variability that would be explained by including all 13 principal components.
- Method 2: Suppose I wanted to include **enough principal components to explain 90%** of the total variability explained by all 13 principal components. In the genetic data case above, I would include the first 10 principal components and drop the final three variables from ***Z****.
- Method 3: Here, we want to "**find the elbow**." In the scree plot above, we see there's a big drop in proportion of variability explained between principal component 3 and the following. In this case, we'd likely include the first three features and drop the remaining features. As you can see, this method is a bit subjective as "elbow" doesn't have a mathematically precise definition and, in this case, we'd include a model that explains only about 42% of the total variability.

In R

```
>pca<-prcomp(data, center = TRUE, scale. = FALSE)
#coordinate of sample on components were identified

#Importance of components
>summary(pca)
```

In R

```
>pca<-prcomp(data, center = TRUE, scale. = FALSE)
#coordinate of sample on components were identified

#Importance of components
>summary(pca)

>pca$x
>plot(pca$x)
```

```
> pca.iris.cov$x
             PC1         PC2         PC3          PC4
  [1,] -2.684125626 -0.319397247 0.027914828  0.0022624371
  [2,] -2.714141687  0.177001225 0.210464272  0.0990265503
  [3,] -2.888990569  0.144949426 -0.017900256 0.0199683897
  [4,] -2.745342856  0.318298979 -0.031559374 -0.0755758166
  [5,] -2.728716537 -0.326754513 -0.090079241 -0.0612585926
  [6,] -2.280859633 -0.741330449 -0.168677658 -0.0242008576
  [7,] -2.820537751  0.089461385 -0.257892158 -0.0481431065
  [8,] -2.626144973 -0.163384960 0.021879318  -0.0452978706
  [9,] -2.886382732  0.578311754 -0.020759570 -0.0267447358
 [10,] -2.672755798  0.113774246 0.197632725  -0.0562954013
 [11,] -2.506947091 -0.645068899 0.075318009  -0.0150199245
 [12,] -2.612755231  0.014729939 0.102150260  -0.1563792078
```

Center, Scale

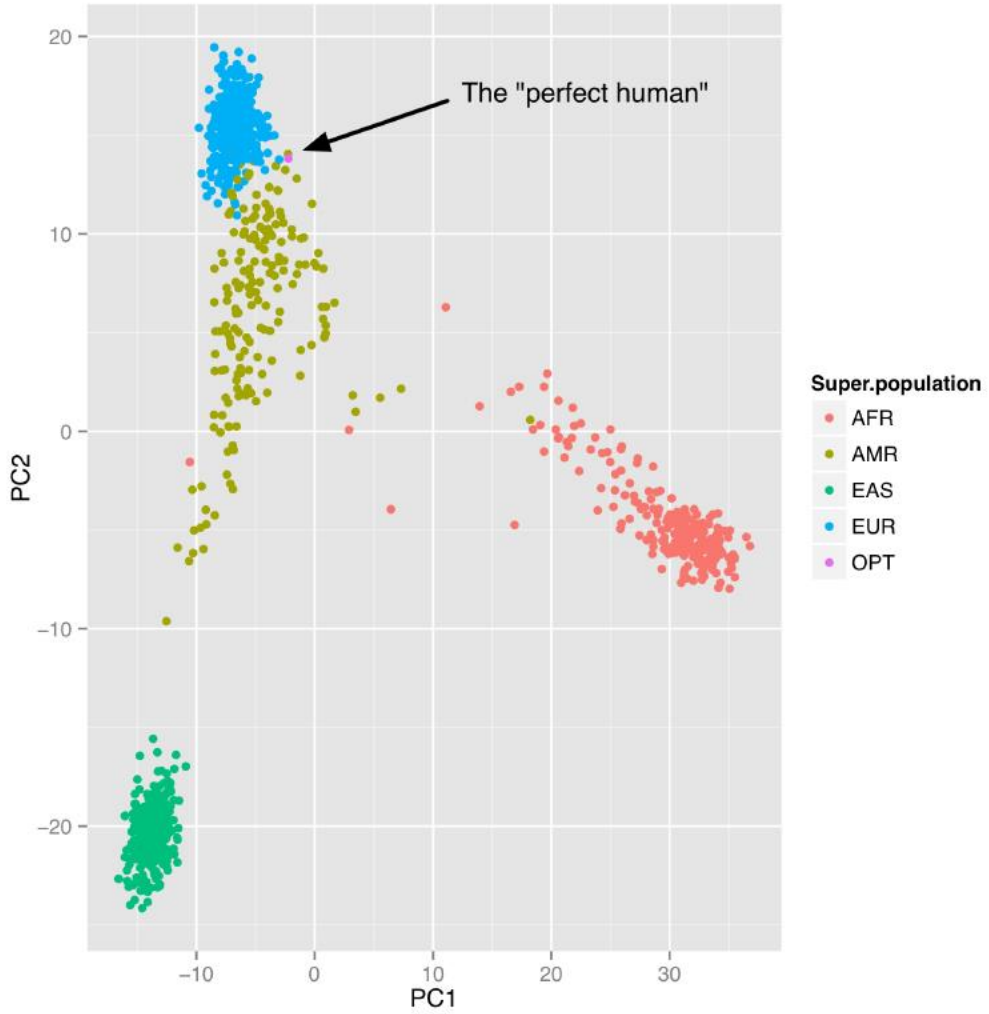
- Why should we center, why should we scale ?
- Will see this through the exercises
- In principal if one considers PCA to be the Eigenvalue of the covariance matrix, then centering yes or no will not change the result.
- In prcomp, however, "PCA" is defined as computing the eigenvalues of the X^TX / (n-1) matrix, which in a centered data is exactly the covariance matrix, otherwise not.

Center, Scale

- Although proven not to be exactly true *, this will result in first PCs capturing the mean of the data as this "explains" most of the variance in the model.
- The scaling will determine if you compute eigenvectors on the covariance matrix (if unscaled) or on the correlation matrix (if scaled).
- This again (mostly) means that what you will capture in the first PCs is mostly what is in a bigger scale.

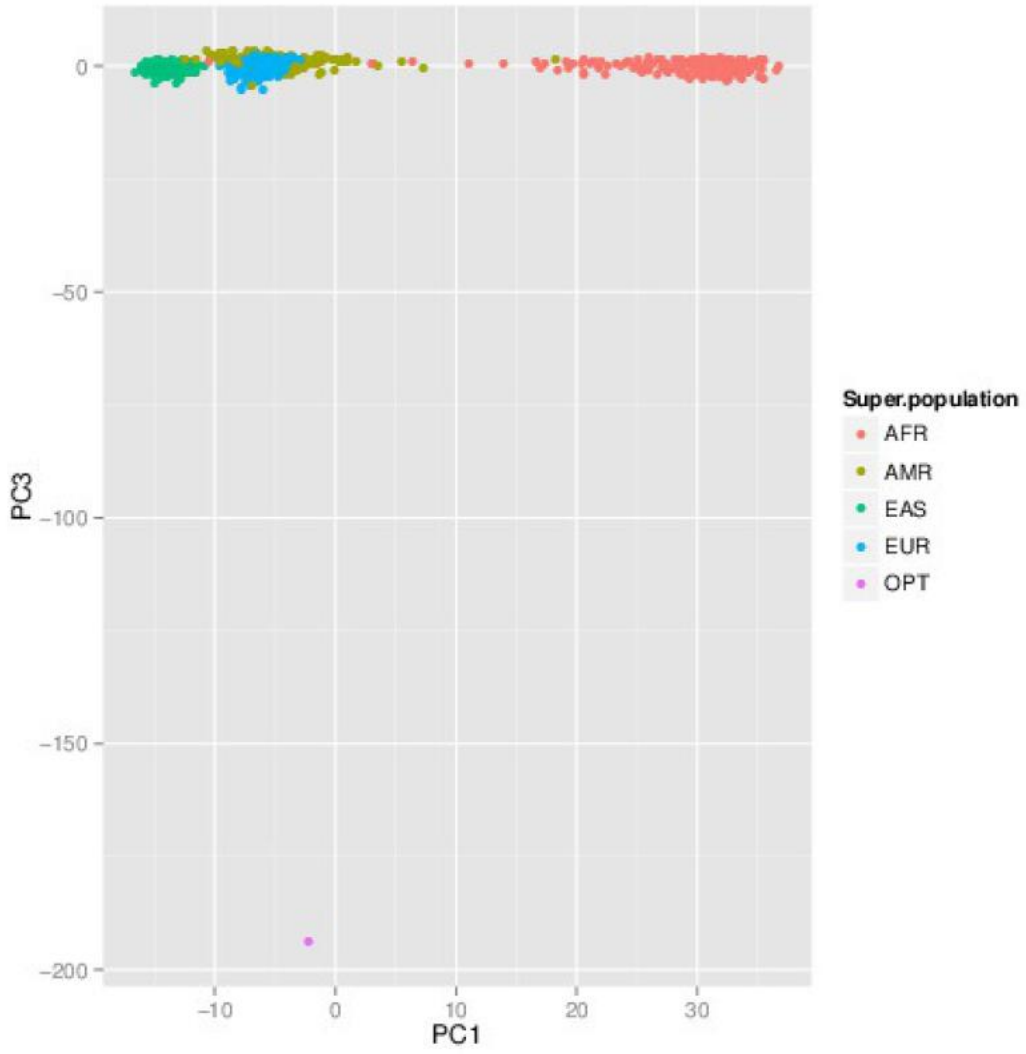
- *The Effect of Data Centering on PCA Models Neal B. Gallagher, Donal O'Sullivan, Manuel Palacios

The perfect human is Puerto Rican…



https://liorpachter.wordpress.com/2014/12/02/the-perfect-human-is-puerto-rican/

… or an alien ?

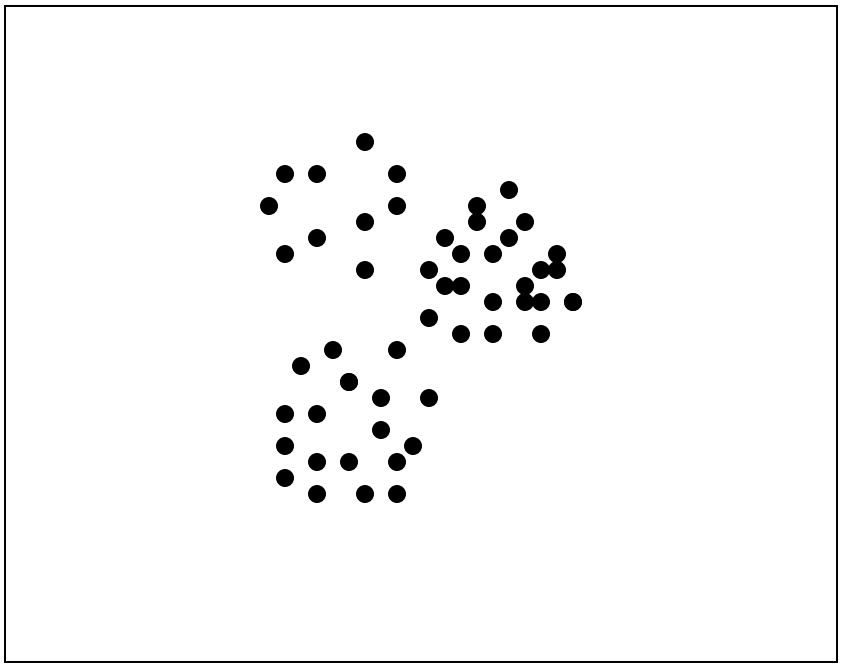

0
−50
−100
−150
−200
PC3
−10
0
10
20
30
PC1
Super.population
AFR
AMR
EAS
EUR
OPT

What's new ?

PCA	linear	Matrix Factorization		
ICA	linear	Matrix Factorization		
MDS	non-linear	Matrix Factorization		
Sparce NNMF	non-linear	Matrix Factorization	2010	https://pdfs.semanticscholar.org/664d/40258f12ad28ed0b7d4c272935ad72a150db.pdf
cPCA	non-linear	Matrix Factorization	2018	https://doi.org/10.1038/s41467-018-04608-8
ZIFA	non-linear	Matrix Factorization	2015	https://doi.org/10.1186/s13059-015-0805-z
ZINB-WaVE	non-linear	Matrix Factorization	2018	https://doi.org/10.1038/s41467-017-02554-5
Diffusion maps	non-linear	graph-based	2005	https://doi.org/10.1073/pnas.0500334102
Isomap	non-linear	graph-based	2000	10.1126/science.290.5500.2319
t-SNE	non-linear	graph-based	2008	https://lvdmaaten.github.io/publications/papers/JMLR_2008.pdf
- BH t-SNE	non-linear	graph-based	2014	https://lvdmaaten.github.io/publications/papers/JMLR_2014.pdf
- Flt-SNE	non-linear	graph-based	2017	arXiv:1712.09005
LargeVis	non-linear	graph-based	2018	arXiv:1602.00370
UMAP	non-linear	graph-based	2018	arXiv:1802.03426
PHATE	non-linear	graph-based	2017	https://www.biorxiv.org/content/biorxiv/early/2018/06/28/120378.full.pdf
scvis	non-linear	Autoencoder (MF)	2018	https://doi.org/10.1038/s41467-018-04368-5
VASC	non-linear	Autoencoder (MF)	2018	https://doi.org/10.1016/j.gpb.2018.08.003

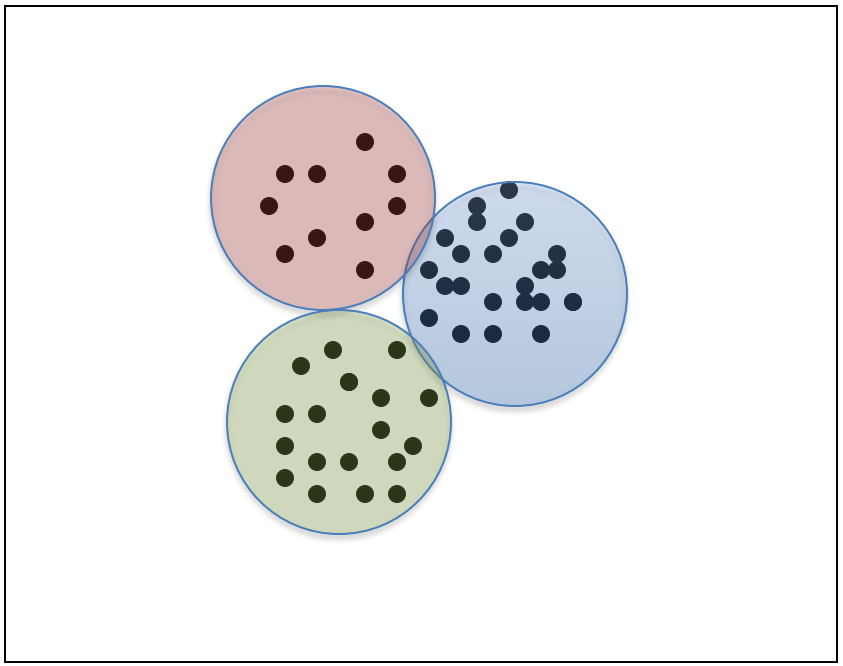
Clustering

Clustering



Point cloud

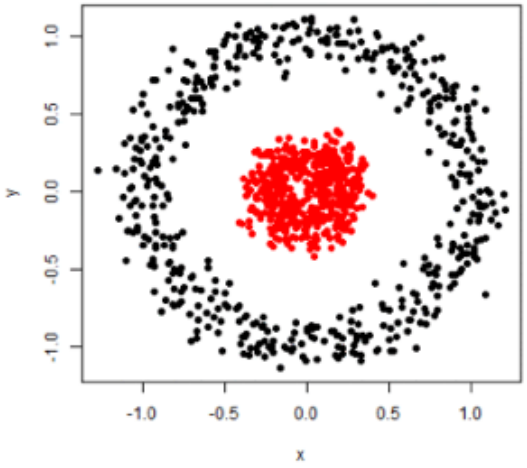
Clustering

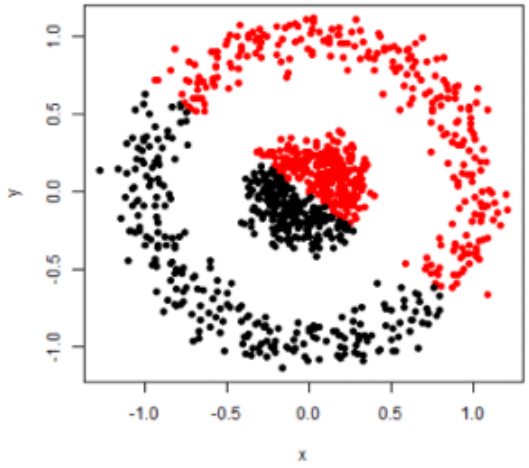


Clustering

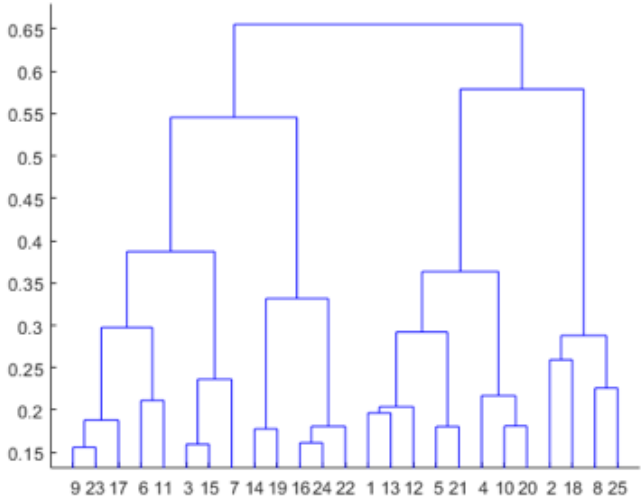
Clustering method are divided into two categories :

Partitioning clustering





Hierarchical clustering



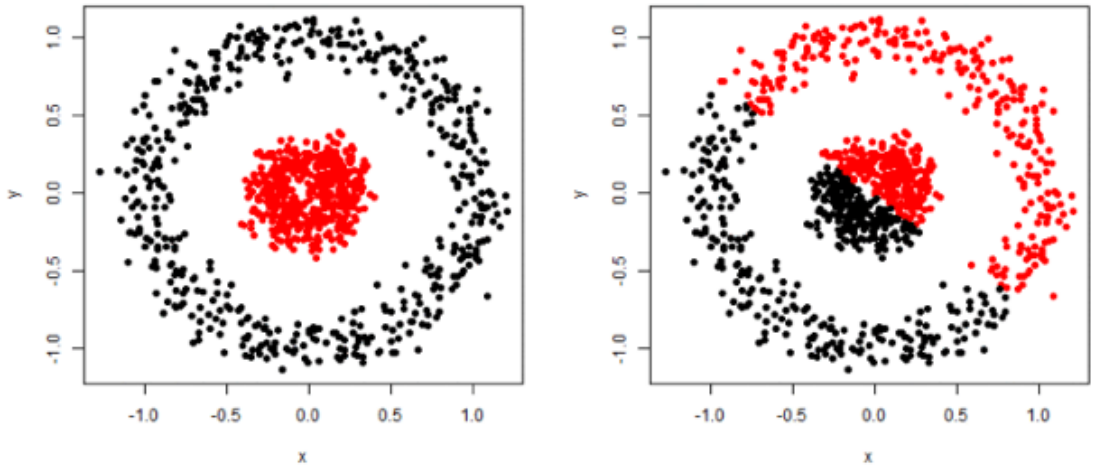
***Handbook of cluster analysis, Hennig C. et al.**

Partitioning clustering

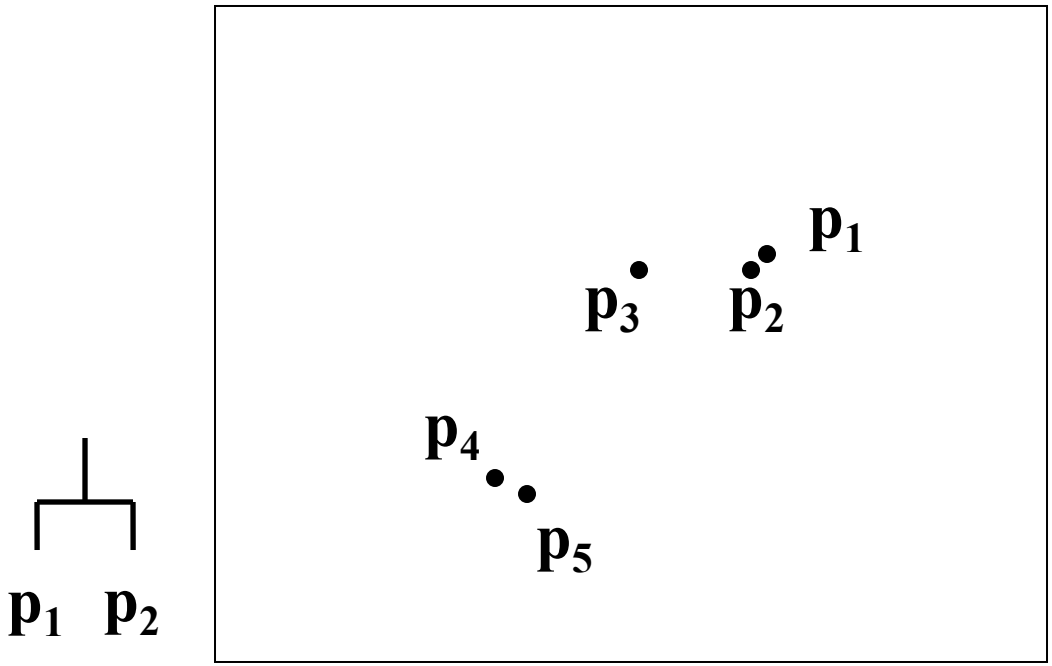
Convex partitioning. Example: K-means

Density based approaches. Example: DBSCAN

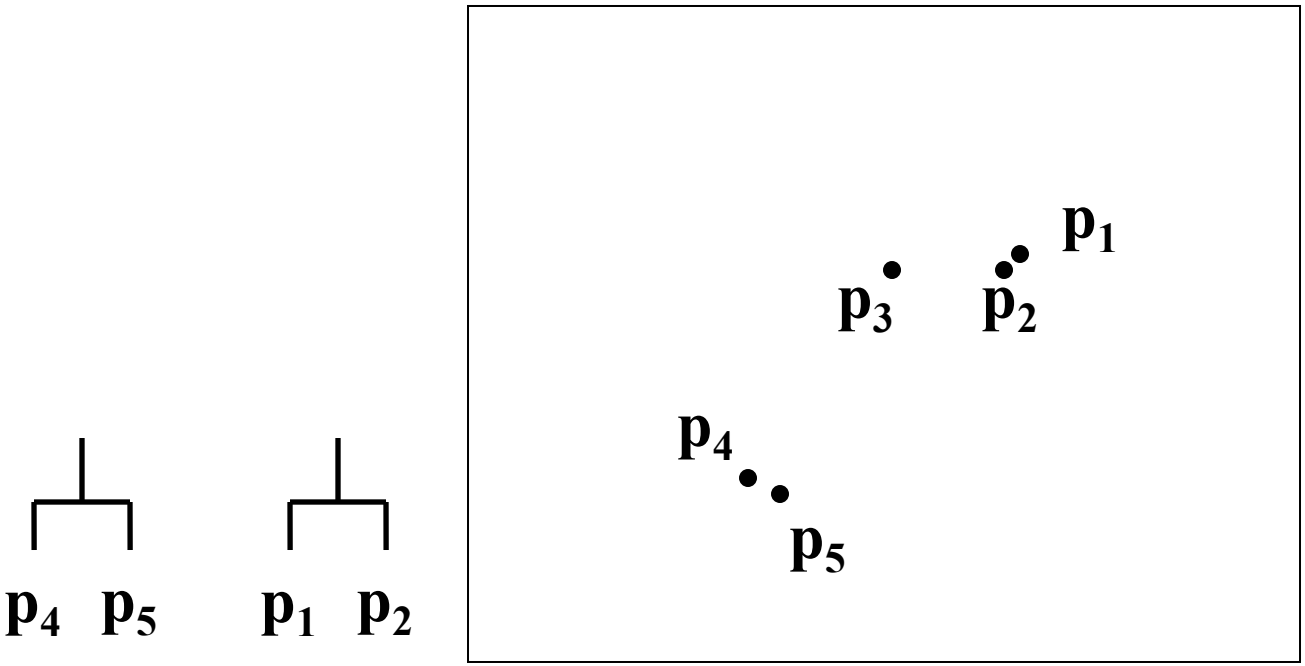
Model-based approaches. Example: Mclust



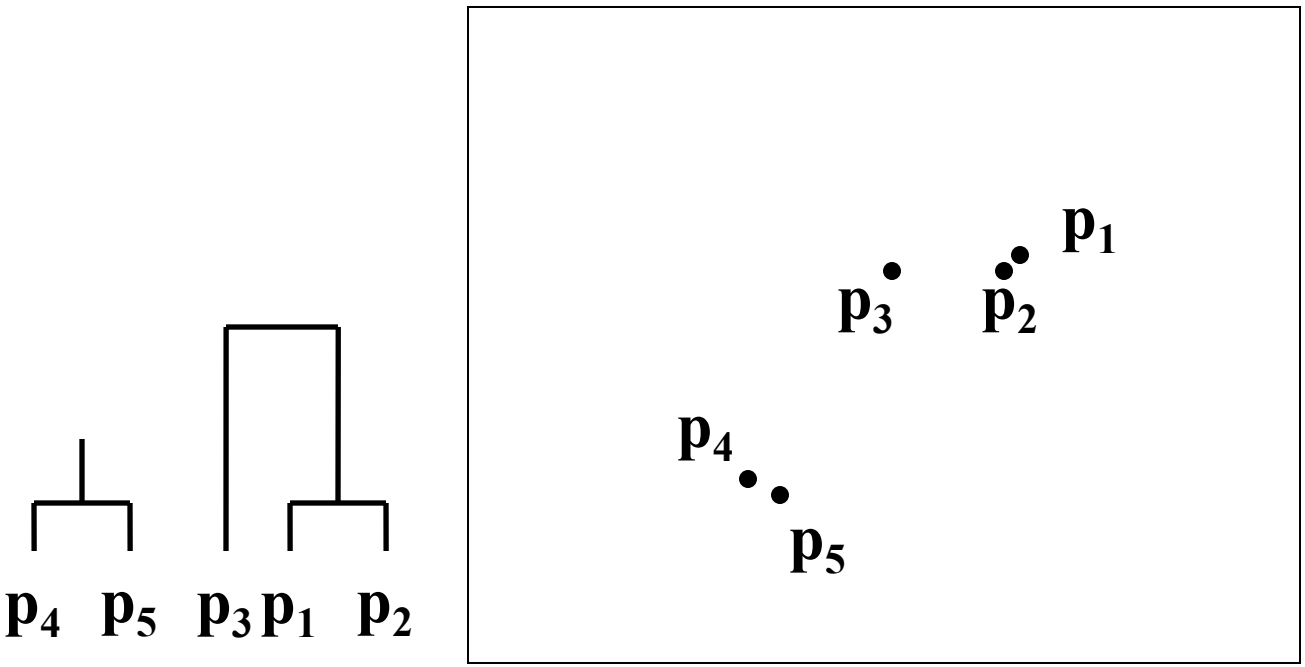
Hierarchical Clustering



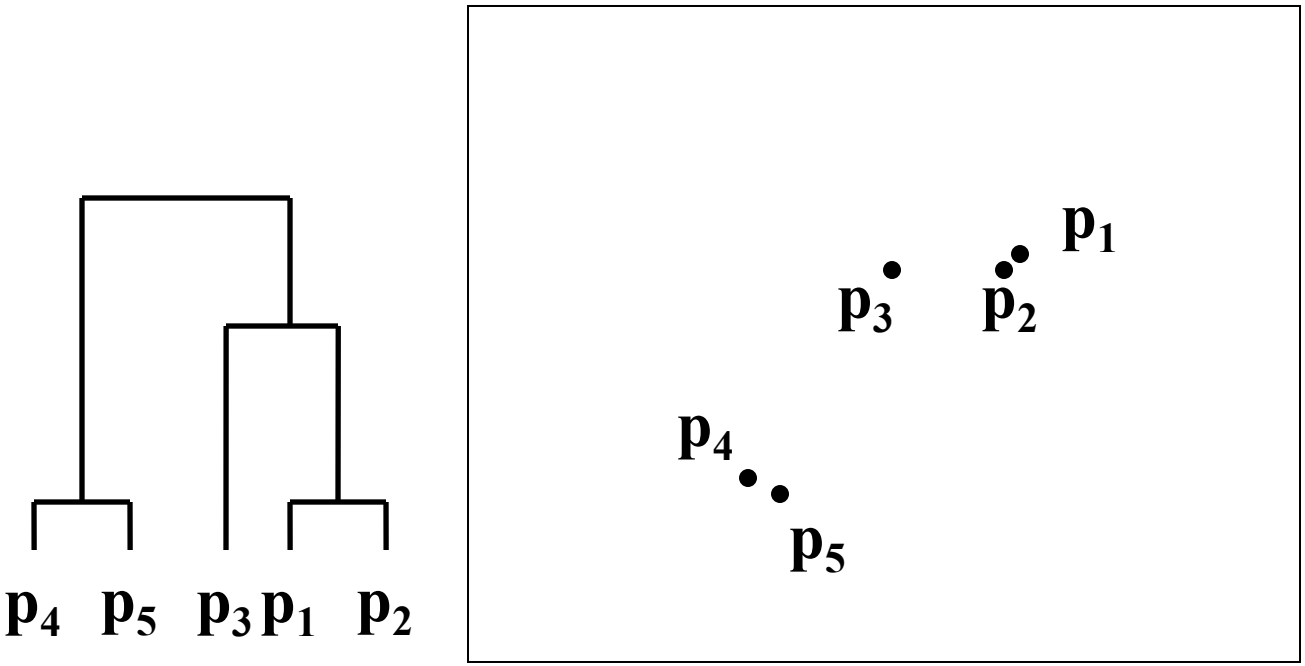
Hierarchical Clustering



Hierarchical Clustering



Hierarchical Clustering



Distance

Euclidean

X = (2, 0)

Y = (-2, -2)

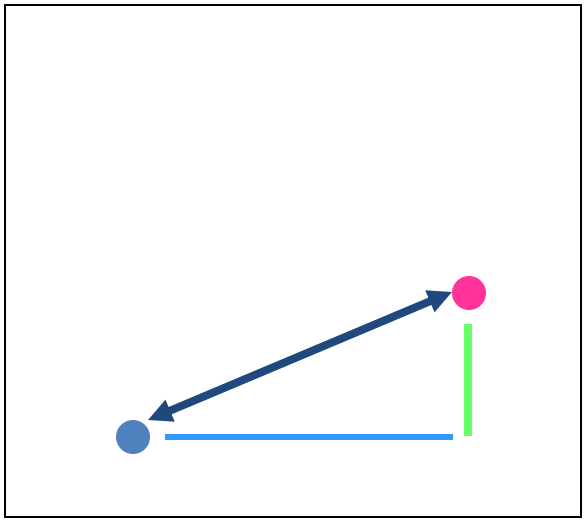
$\sqrt{\ [\Sigma\ (y - x)^2\ ]}$

$= \sqrt{([-2 - 2]^2 + [-2 - 0]^2)}$

$= \sqrt{(4^2 + 2^2)}$

$= \sqrt{20}$

$= 4.47$



It represents the “multivariate dissimilarity” of X & Y

Squared Euclidean

$X = (2, 0)$

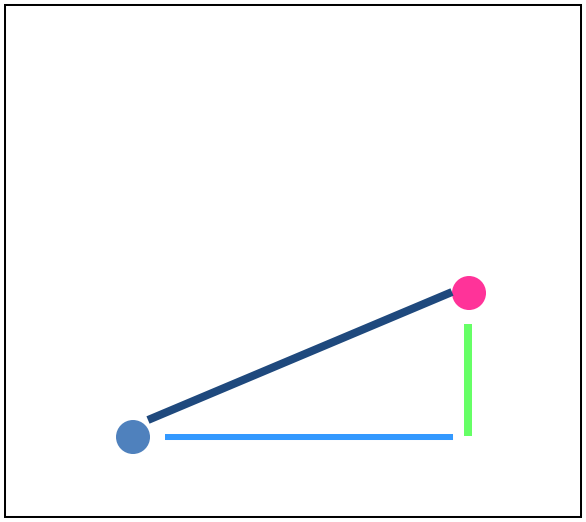
$Y = (-2, -2)$

$\Sigma (y - x)^2$

$= ([-2 - 2]^2 + [-2 - 0]^2)$

$= (4^2 + 2^2)$

$= 20$



It represents the “multivariate dissimilarity” of X & Y

City Block

X = (2, 0)

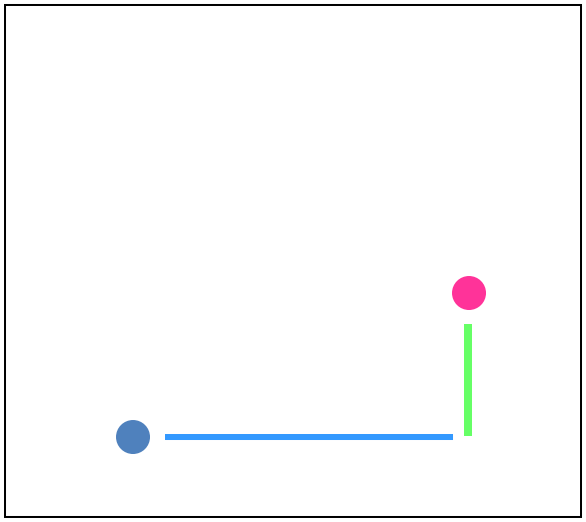
Y = (-2, -2)

$\Sigma |y - x|$

= (|-2 – 2| + |-2 – 0|)

= |-4| + |-2|

= 6



Distance Measures in 2D

- Euclidean $\sqrt{[\Sigma (y - x)^2]}$
- Squared Euclidean $\Sigma (y - x)^2$
- City-Block $\Sigma | y - x |$

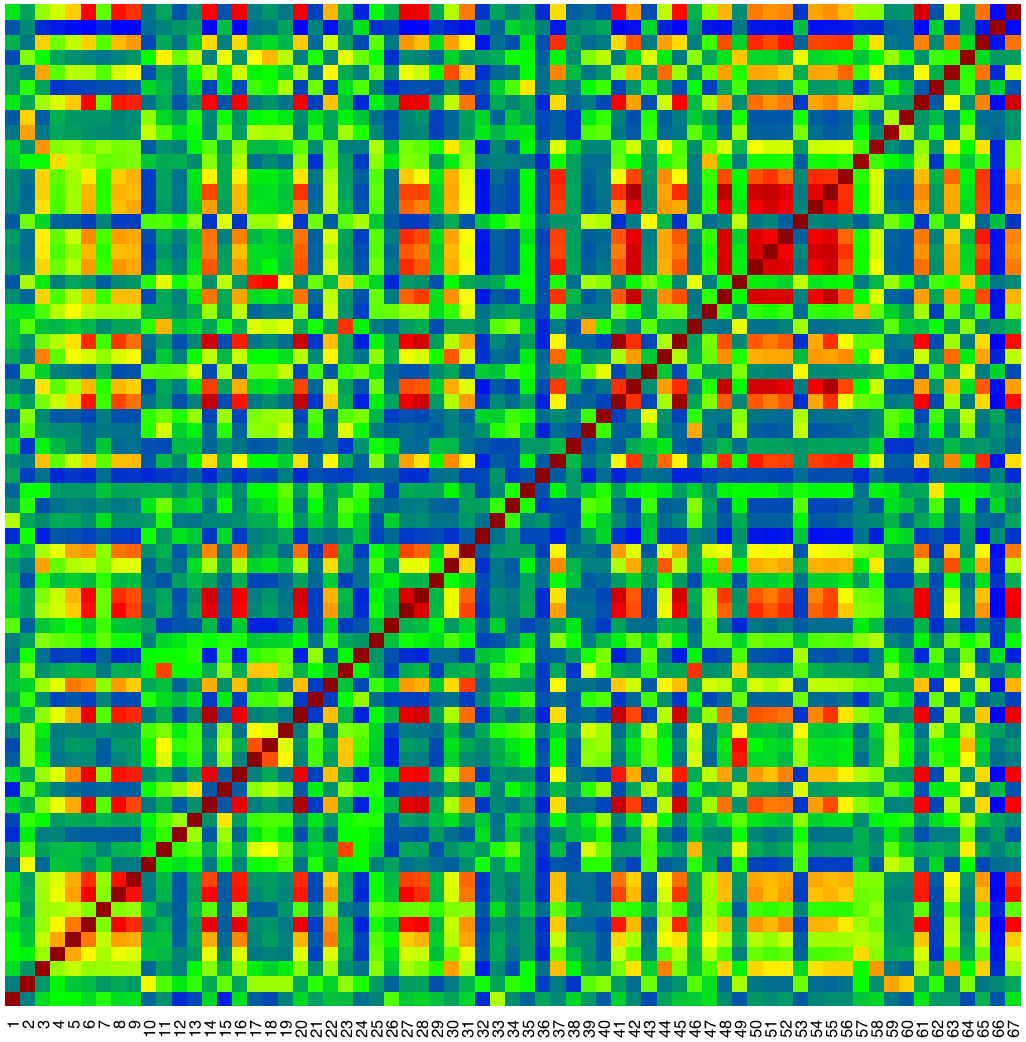
Distance Measures in nD

- Euclidean $$d = \sqrt{(a_1 - b_1)^2 + (a_2 - b_2)^2 + \ldots + (a_n - b_n)^2}$$
- Squared Euclidean $$d = (a_1 - b_1)^2 + (a_2 - b_2)^2 + \ldots + (a_n - b_n)^2$$
- City-Block $$d = |a_1 - b_1| + |a_2 - b_2| + \ldots + |a_n - b_n|$$

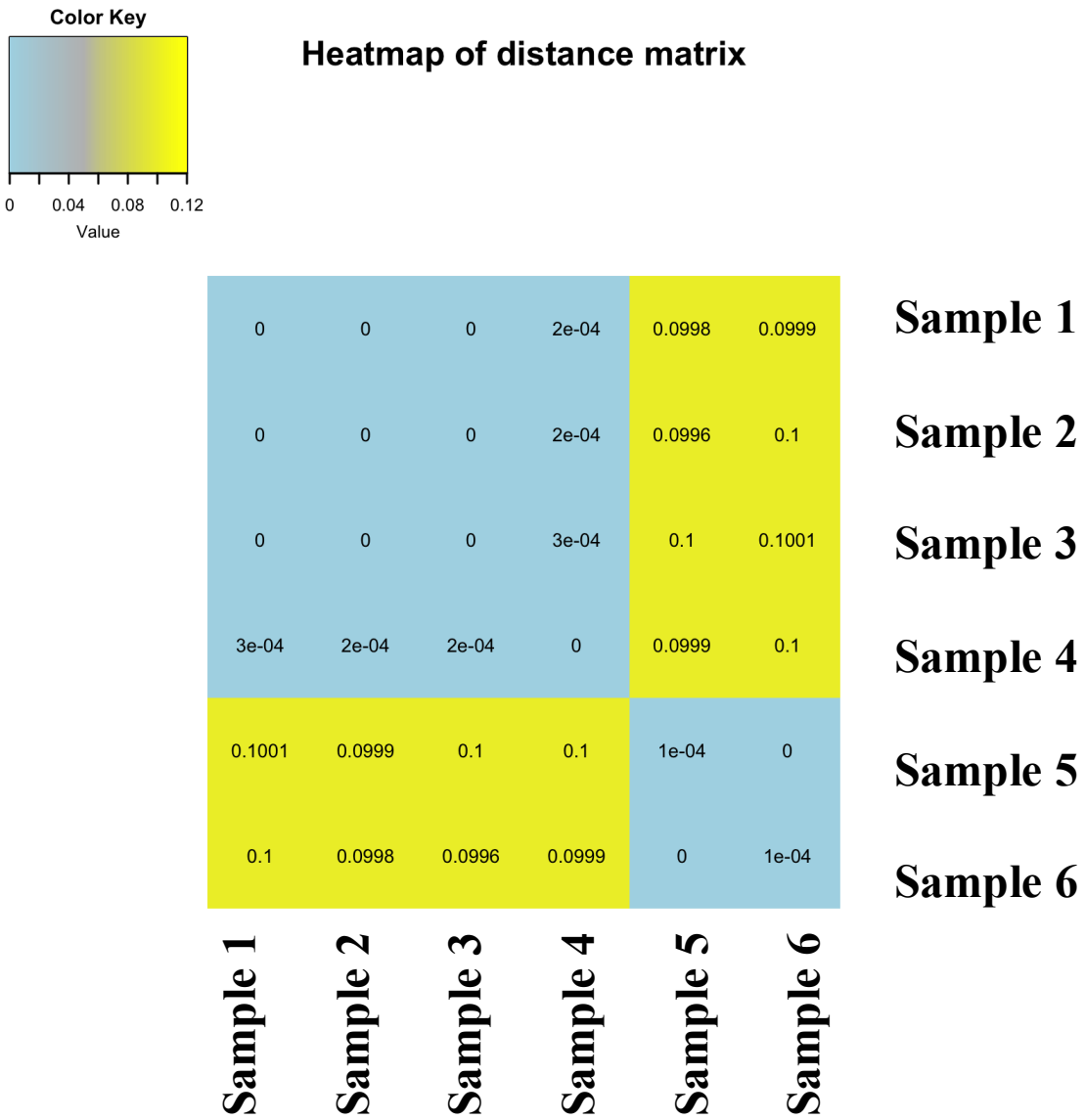
In R

```
>?dist
```

Distance matrix



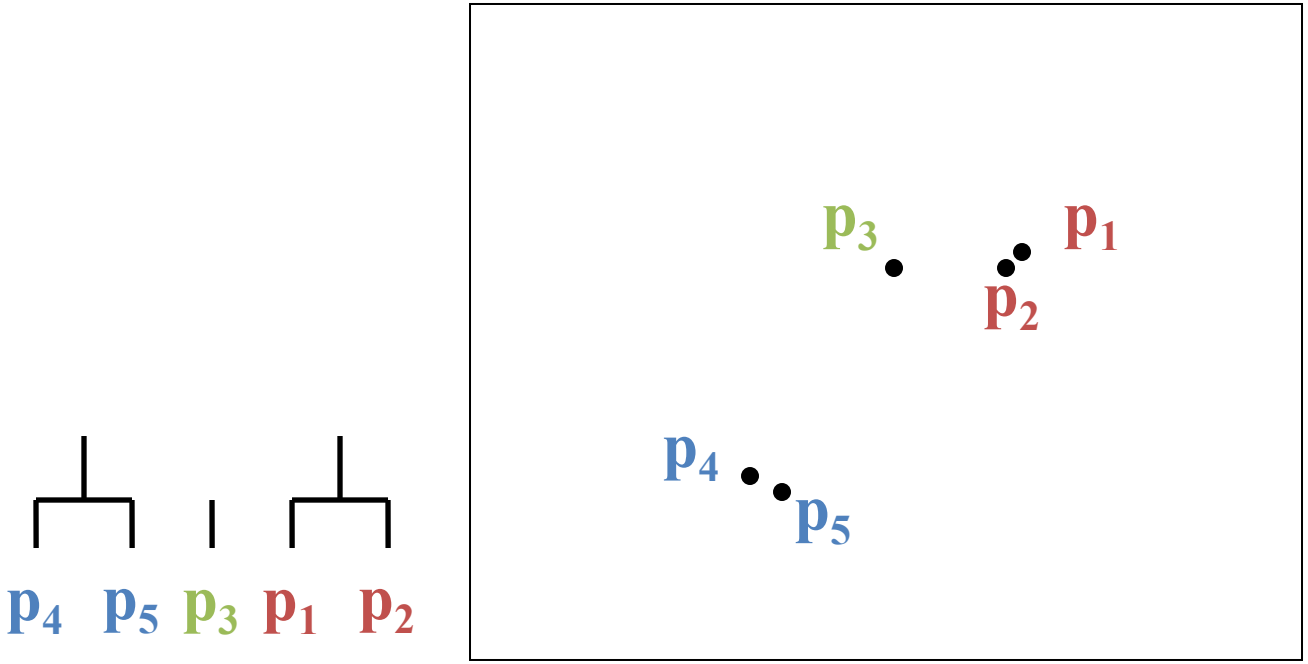
Distance matrix



In R

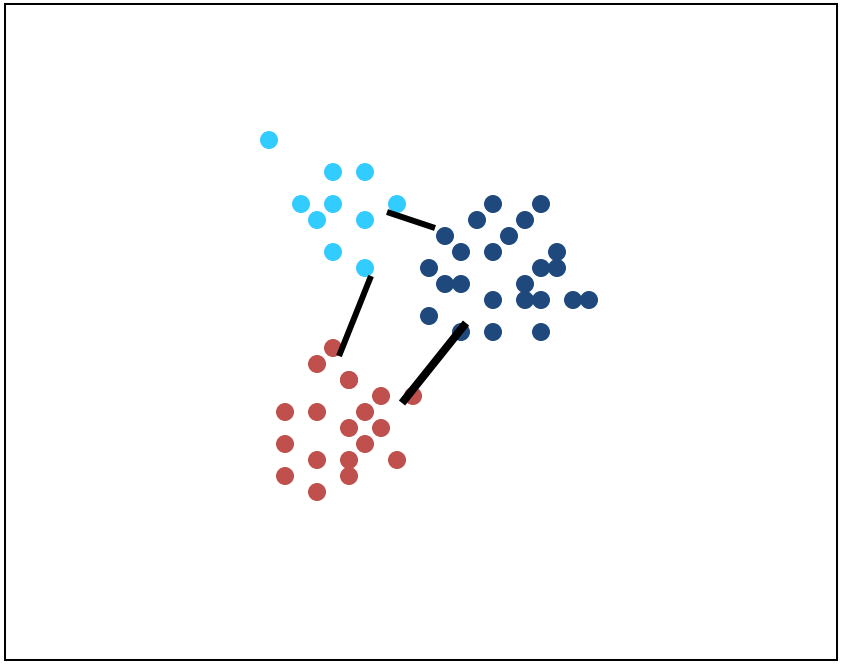
```
>?heatmap
>heatmap(distanceMatrix,Colv=NA, Rowv=NA,
scale="none")
```

How to aggregate clusters?


p3
p1
p2
p4
p5
p4 p5 p3 p1 p2

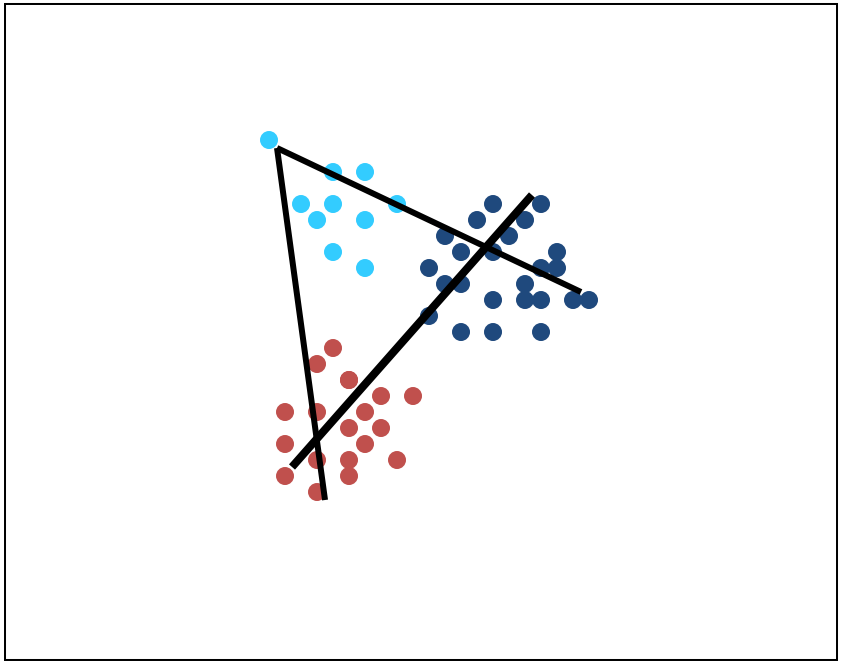
Which clusters to combine?

Single linkage



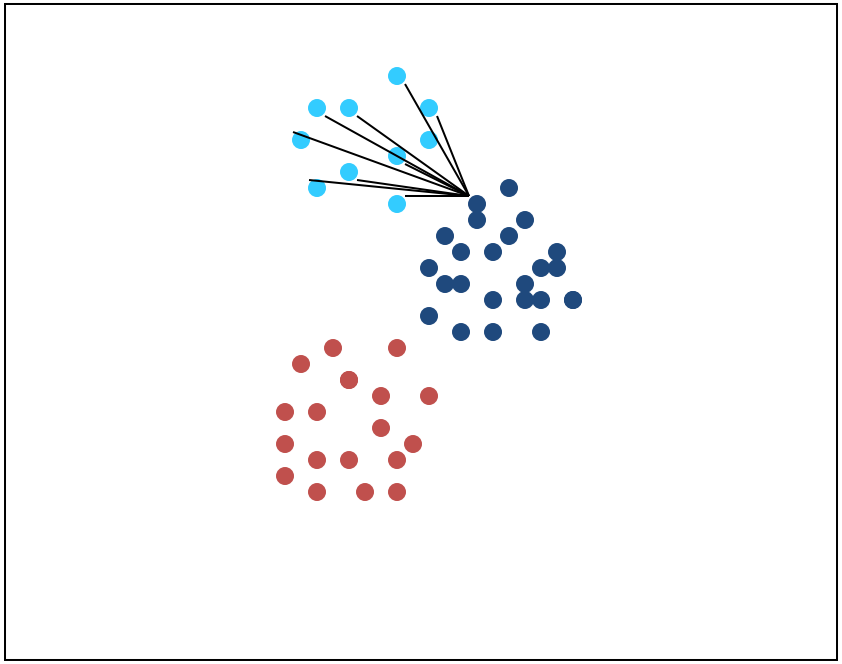
Distance between closest elements in clusters

Complete Linkage



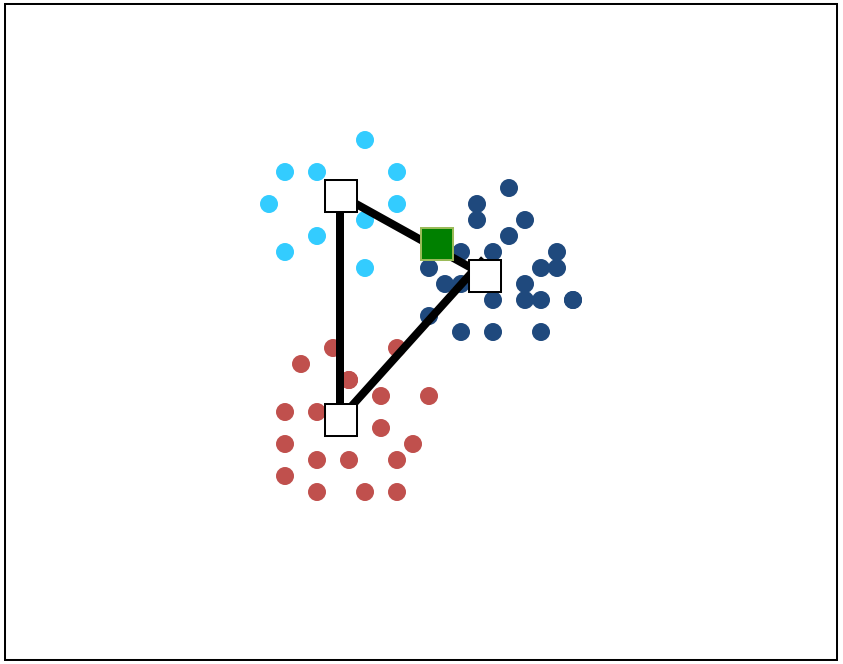
Distance between furthest elements in clusters

Average Linkage



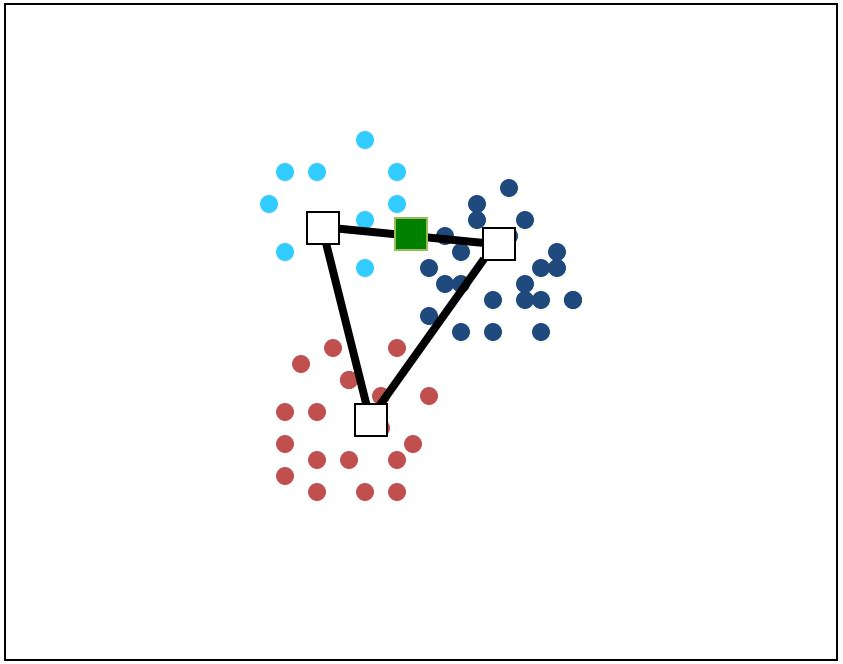
Average of all pairwise distances

Centroid Condensation (mean)



Distance between centroids (means) of two clusters

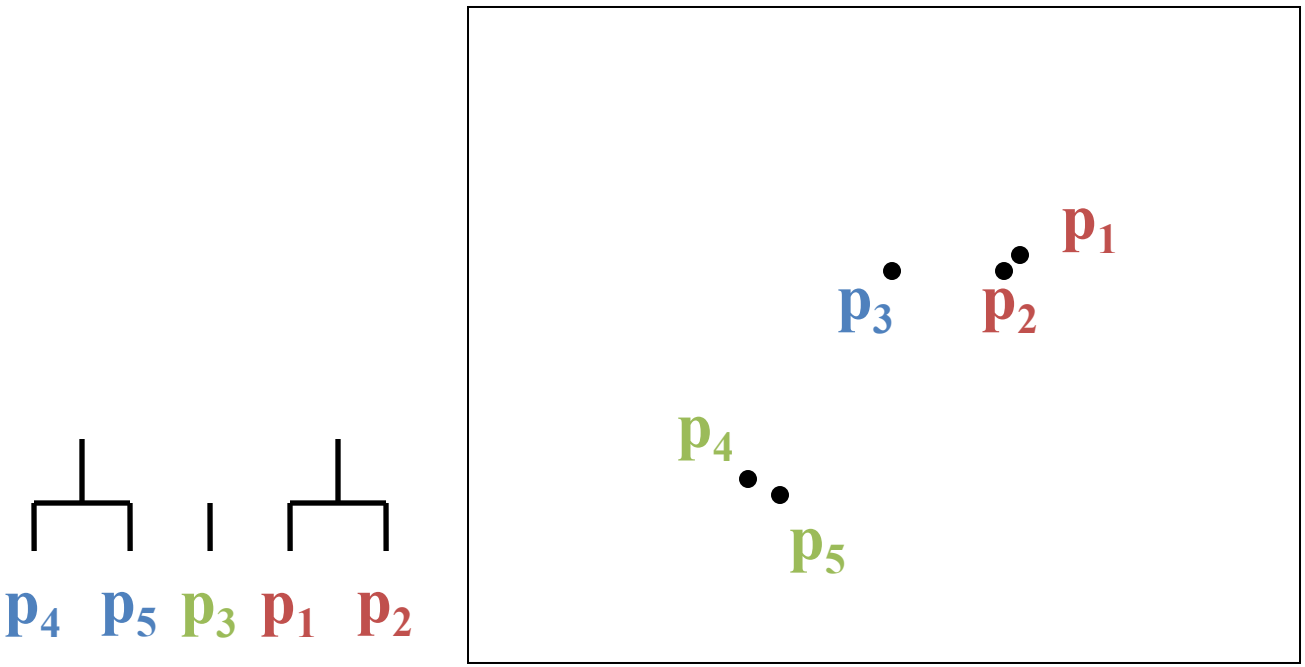
Median Condensation



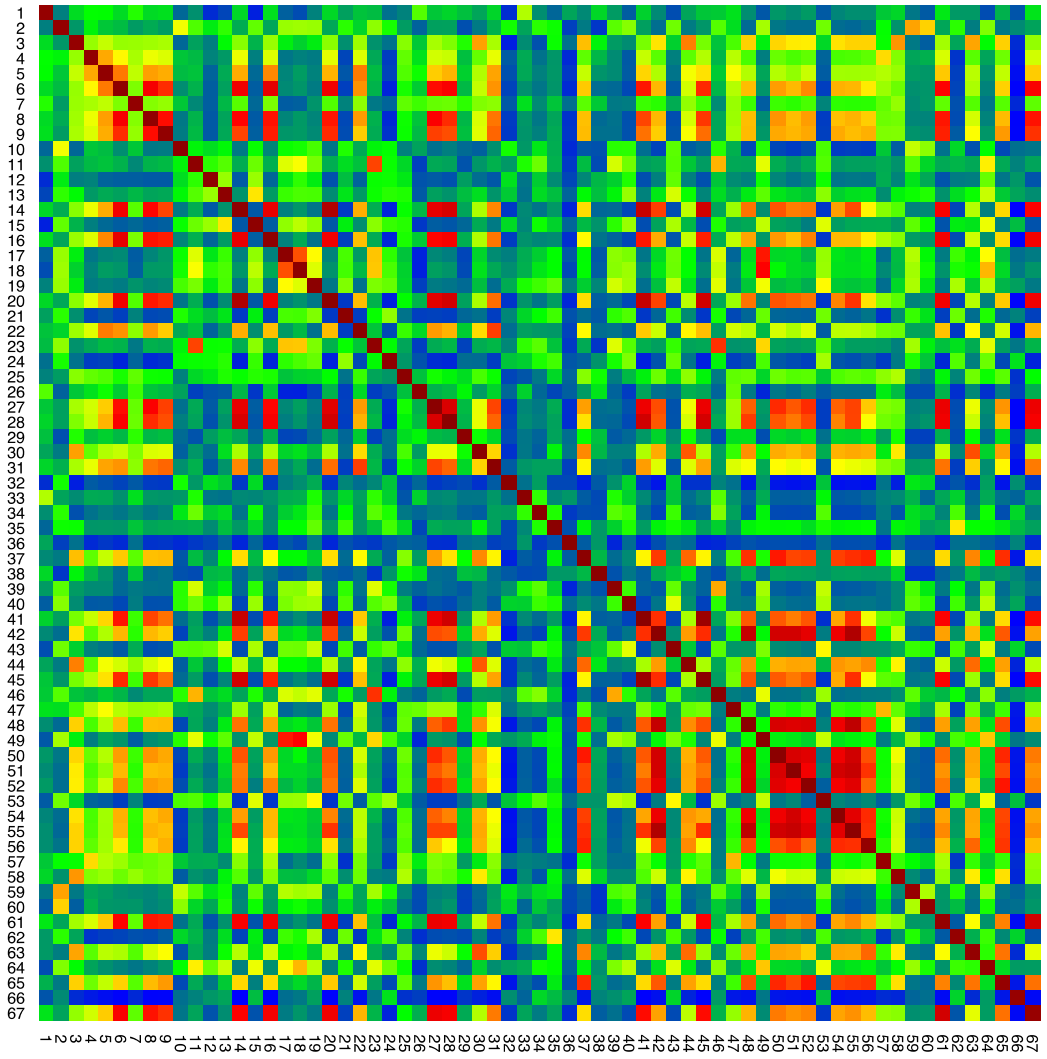
Distance between median distances of two clusters

Clustering methods

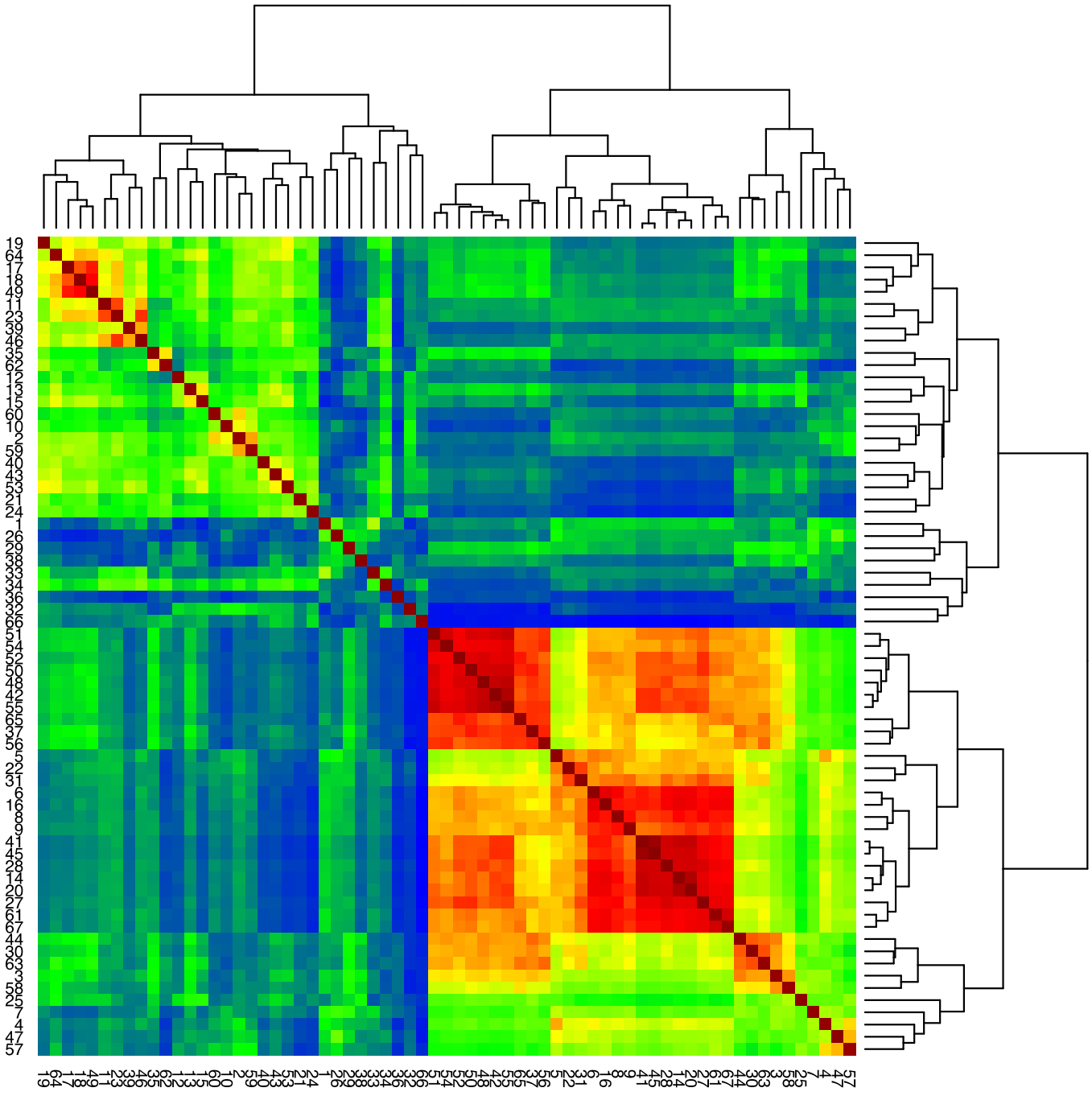
Hierarchical Clustering



At the beginning every point is a cluster in it self, then we agglomerate …

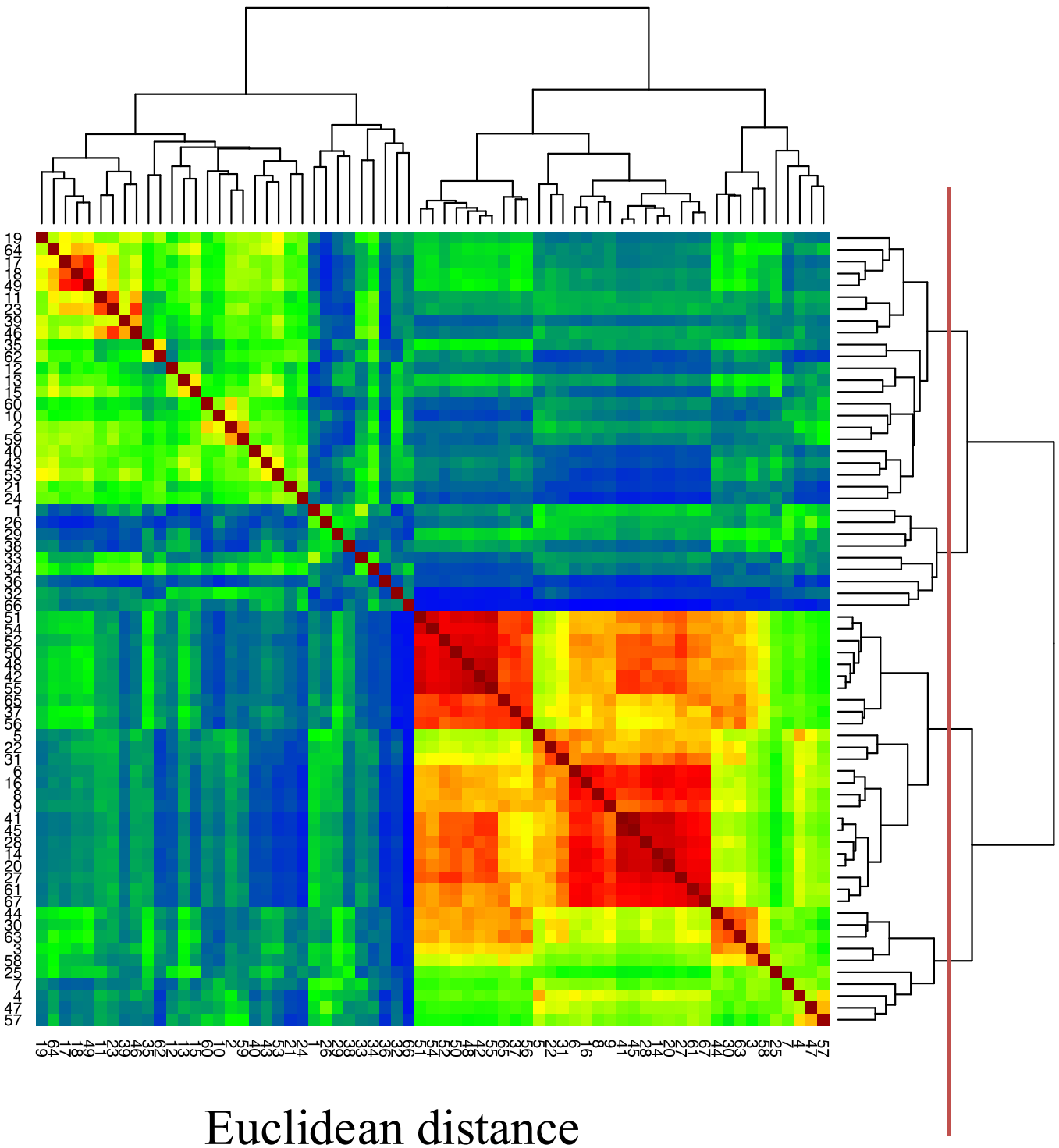


Euclidean distance

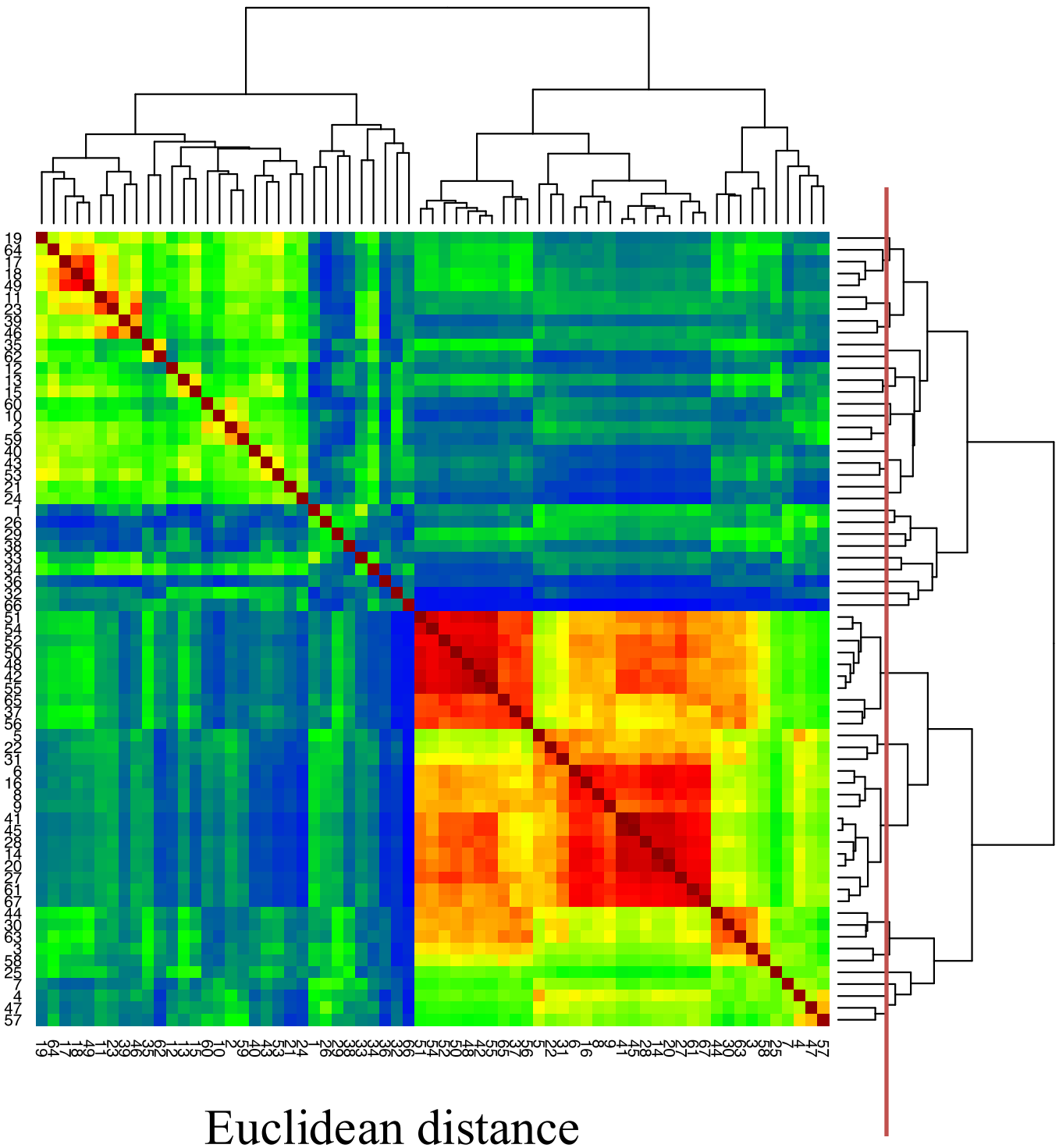


Euclidean distance
complete Linkage

Determine the Termination Condition (TC)



Euclidean distance
complete Linkage



Euclidean distance
complete Linkage

Let's practice In R

```
#create a random matrix
>mat <- matrix(data = rnorm(300, mean= 100,
sd=10), nrow = 150, ncol = 2)

#Euclidian distance
>mat.dist<-as.matrix(dist(mat))

#show heatmap
>heatmap(mat.dist,Colv=NA, Rowv=NA, scale="none")

#change heatmap's color
>colorScale <- colorRampPalette(c("blue",
"green","yellow","red","darkred"))(1000)
>heatmap(mat.dist,Colv=NA, Rowv=NA, scale="none",
col=colorScale)
```

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```
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"green","yellow","red","darkred"))(1000)
>heatmap(mat.dist,Colv=NA, Rowv=NA, scale="none",
col=colorScale)
```

How to do hierarchical clustering in R?

```
>?hclust

#Euclidian distance
>distE<-dist(mat)
>mat.distE<-as.matrix(dist(mat))
>heatmap(mat.distE, Colv=NA, Rowv=NA, scale="none”)
>hE<-hclust(distE,”complete")
>plot(hE)

#manhattan distance
>distC<-dist(mat,method=“manhattan”
>mat.distC<-as.matrix(dist(mat,method=“manhattan”)
>heatmap(mat.distC, Colv=NA, Rowv=NA, scale="none”)
>hC<-hclust(distC,”complete”)
>plot(hC)
```

How to do hierarchical clustering in R?

```
>?hclust

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>heatmap(mat.distE, Colv=NA, Rowv=NA, scale="none”)
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>heatmap(mat.distC, Colv=NA, Rowv=NA, scale="none”)
>hC<-hclust(distC,”complete”)
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```

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>?hclust

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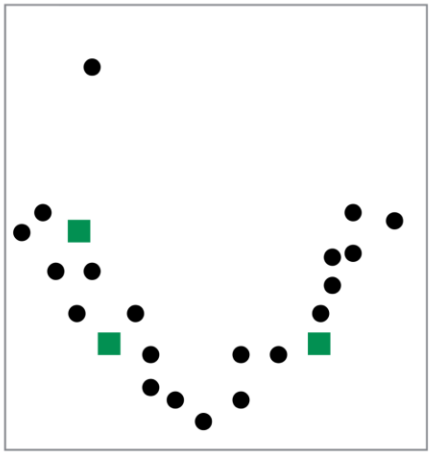
>mat.distC<-as.matrix(dist(mat,method=“manhattan”)
>heatmap(mat.distC, Colv=NA, Rowv=NA, scale="none”)
>hC<-hclust(distC,”complete”)
>plot(hC)
```

K-means Clustering

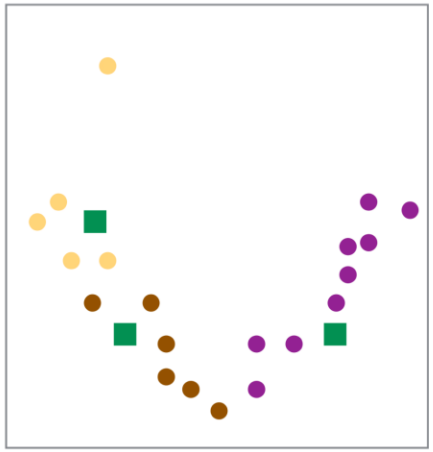
Number of clusters = 3

a



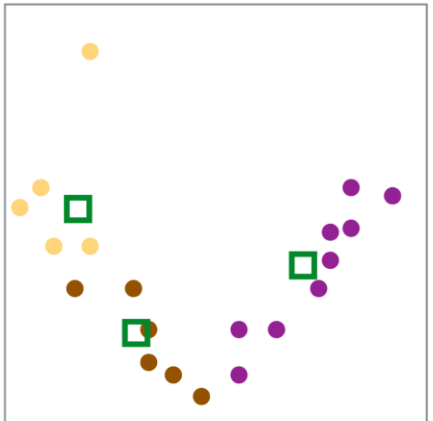
Start with 3 initial points

b



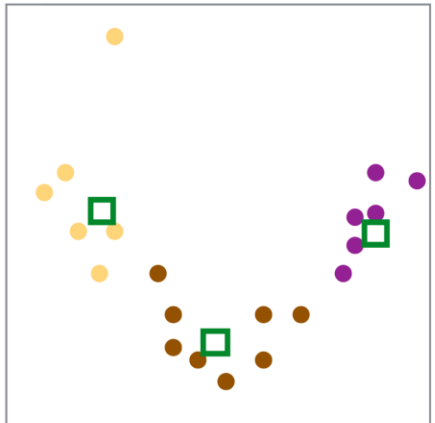
For each point determine to which initial point it is the closest

c



Move initial points to the centroids of the clusters

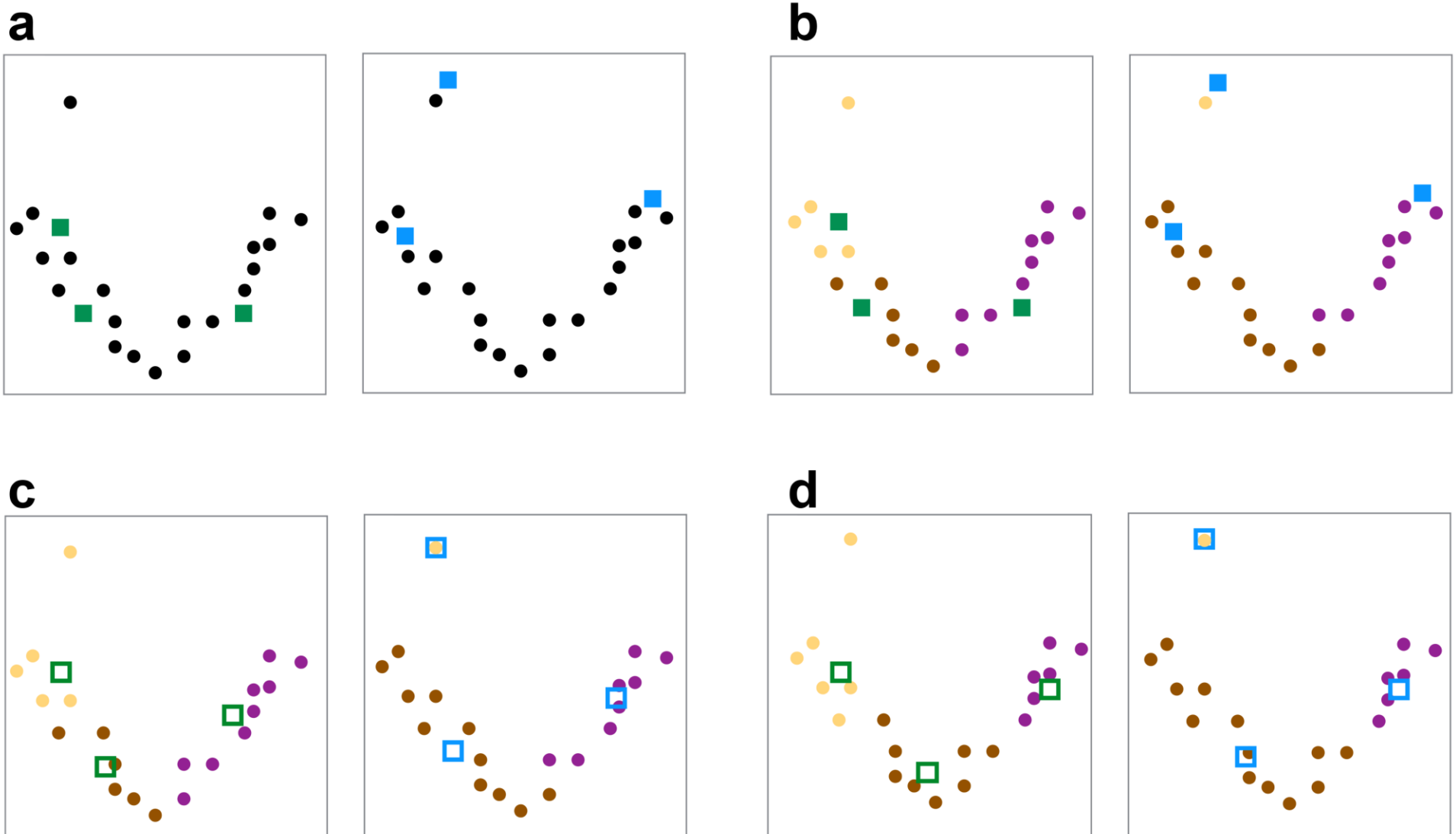
d



Color again each point! Repeat b and c until obtaining stabilisation

K-means Clustering

Number of clusters = 3



K-means & C-means

Drawbacks:

1. Specify number of clusters
2. Non probabilistic methods
3. Not stable

Kmeans in R

```
>mat <- matrix(data = rnorm(300, mean=
100, sd=10),
          nrow = 150,
          ncol = 2)
>df<-data.frame(x)

>kmeans(df,3)
```

Kmeans in R

```
>mat <- matrix(data = rnorm(300, mean= 100, sd=10),
               nrow = 150,
               ncol = 2)
>df<-data.frame(mat)

>kmeans(df,3)
```

```
>cl.1 <- kmeans(df, 3, iter.max = 1)
>plot(df, col = cl.1$cluster)
>points(cl.1$centers, col = 1:5, pch = 8)
```

Kmeans in R

```
>mat <- matrix(data = rnorm(300, mean= 100, sd=10),
               nrow = 150,
               ncol = 2)
>df<-data.frame(mat)

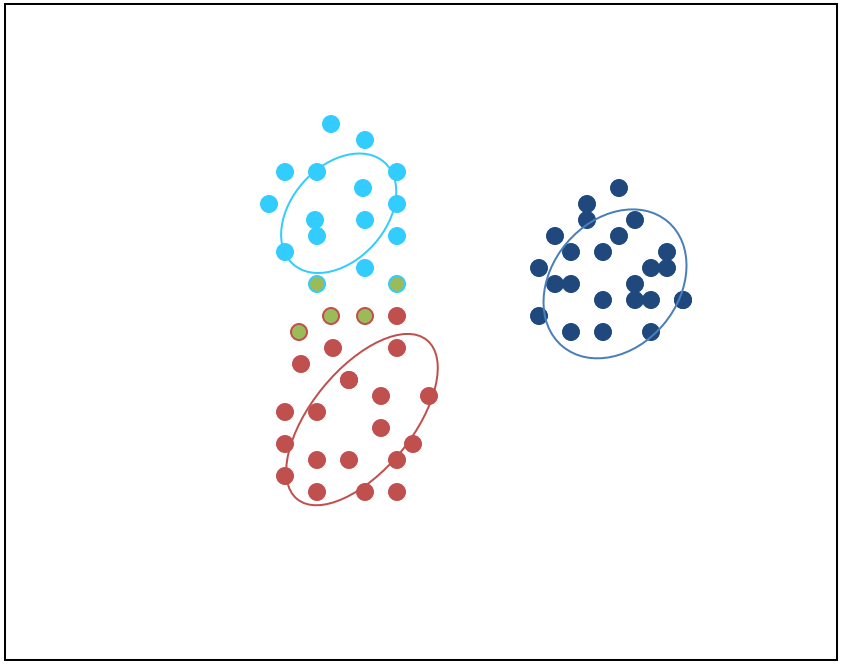
>kmeans(df,3)

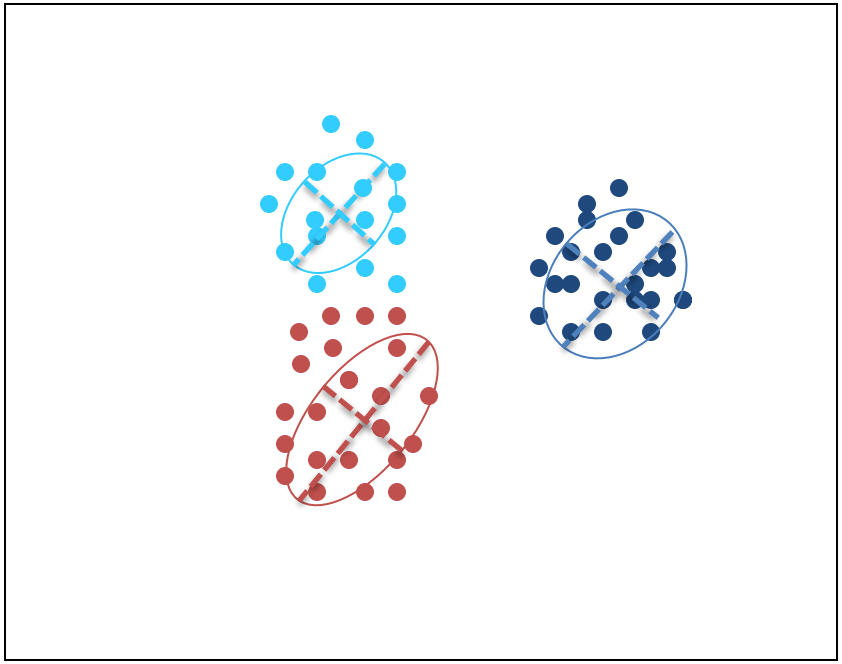
>cl.1 <- kmeans(df, 3, iter.max = 1)
>plot(df, col = cl.1$cluster)
>points(cl.1$centers, col = 1:5, pch = 8)

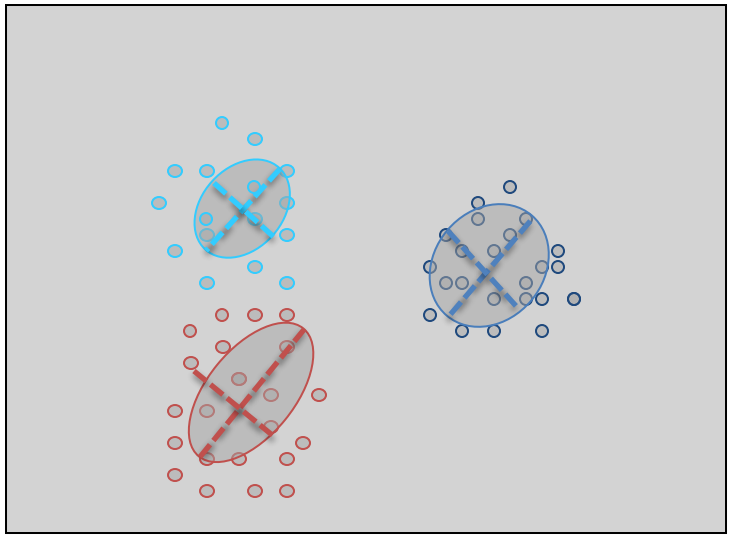
>cl.10 <- kmeans(df, 3, iter.max = 10)
>plot(df, col = cl.10$cluster)
>points(cl.10$centers, col = 1:5, pch = 8)

>cl.100 <- kmeans(df, 3, iter.max = 100)
>plot(df, col = cl.100$cluster)
>points(cl.100$centers, col = 1:5, pch = 8)
```

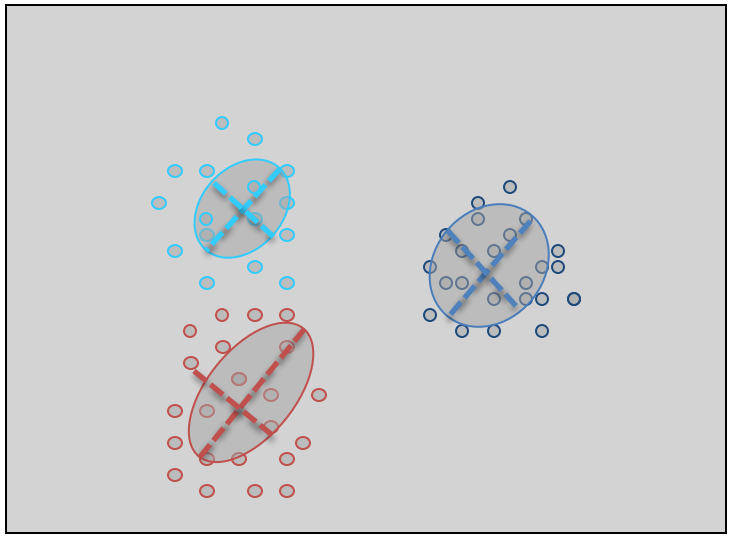
Model-based Clustering



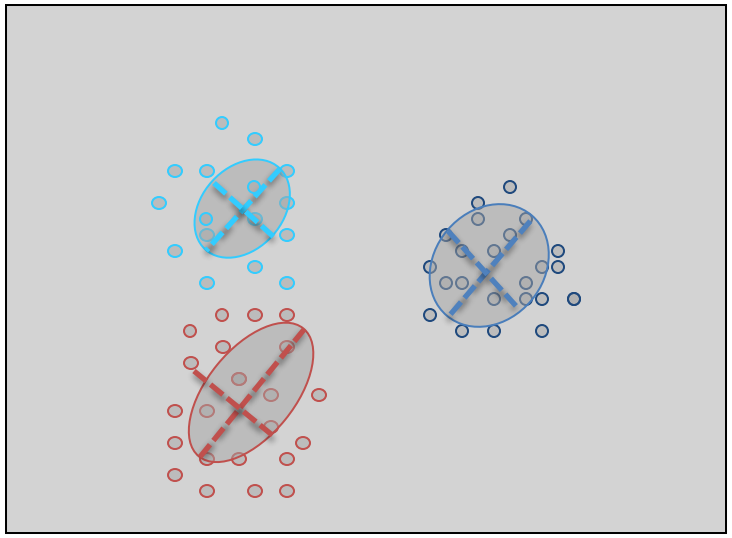




distribution
(univariate, spherical, diagonal, elipsoidal)

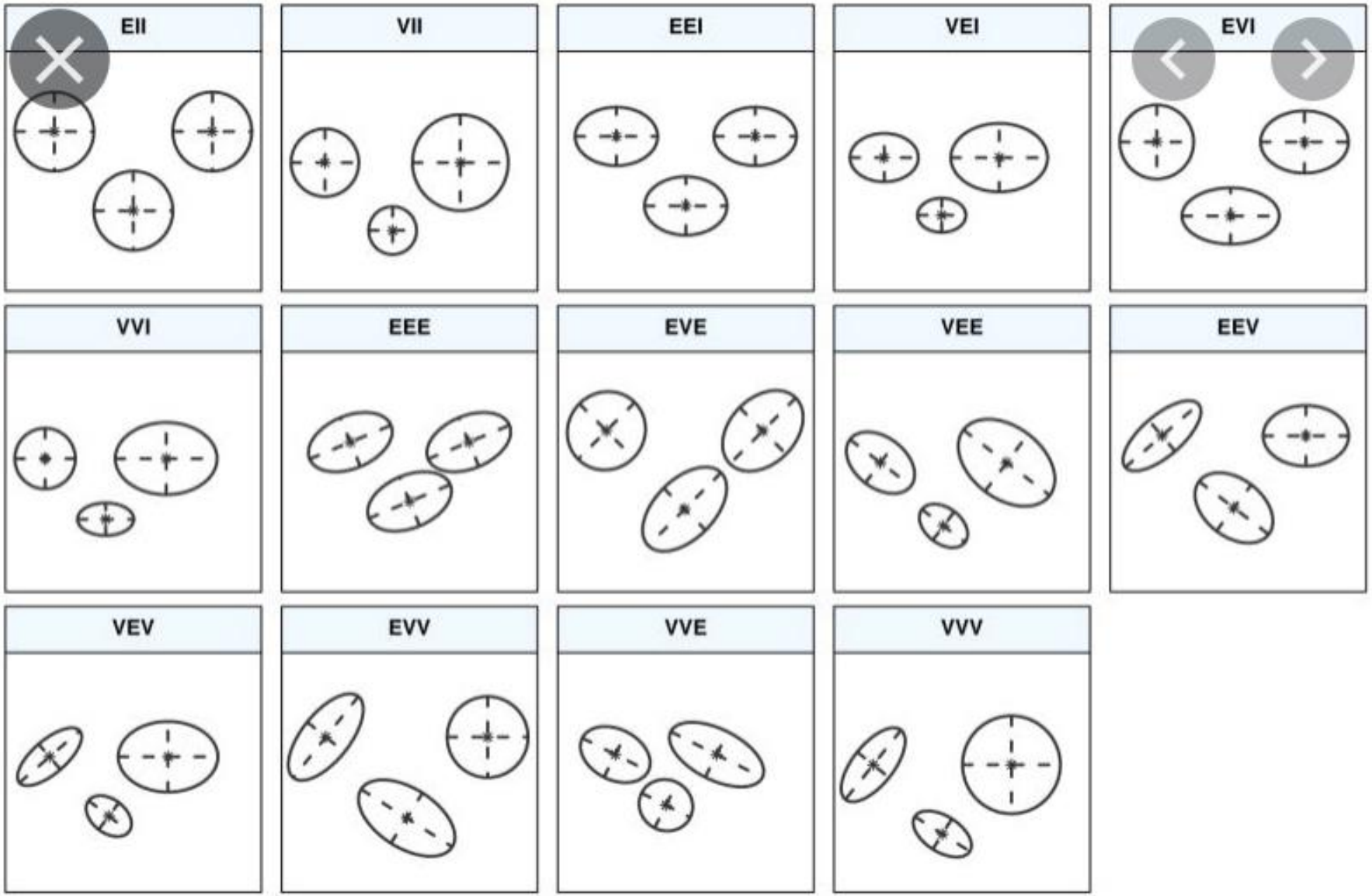


data volume
(equal, variable)



shape
(equal, variable)

Volume, Shape, Orientation



Model selection

identifier	Model	HC	EM	Distribution	Volume	Shape	Orientation
E		•	•	(univariate)	equal		
V		•	•	(univariate)	variable		
EII	λI	•	•	Spherical	equal	equal	NA
VII	$\lambda_k I$	•	•	Spherical	variable	equal	NA
EEI	λA		•	Diagonal	equal	equal	coordinate axes
VEI	$\lambda_k A$		•	Diagonal	variable	equal	coordinate axes
EVI	λA_k		•	Diagonal	equal	variable	coordinate axes
VVI	$\lambda_k A_k$		•	Diagonal	variable	variable	coordinate axes
EEE	$\lambda D A D^T$	•	•	Ellipsoidal	equal	equal	equal
EEV	$\lambda D_k A D_k^T$		•	Ellipsoidal	equal	equal	variable
VEV	$\lambda_k D_k A D_k^T$		•	Ellipsoidal	variable	equal	variable
VVV	$\lambda_k D_k A_k D_k^T$	•	•	Ellipsoidal	variable	variable	variable

BIC= Baysian information criterion

BIC

- the model with **the highest BIC** is preferred
- BIC is a function of the number of parameters of the model
- The goodness of the fit of the model
- The sample size

Number of parameters

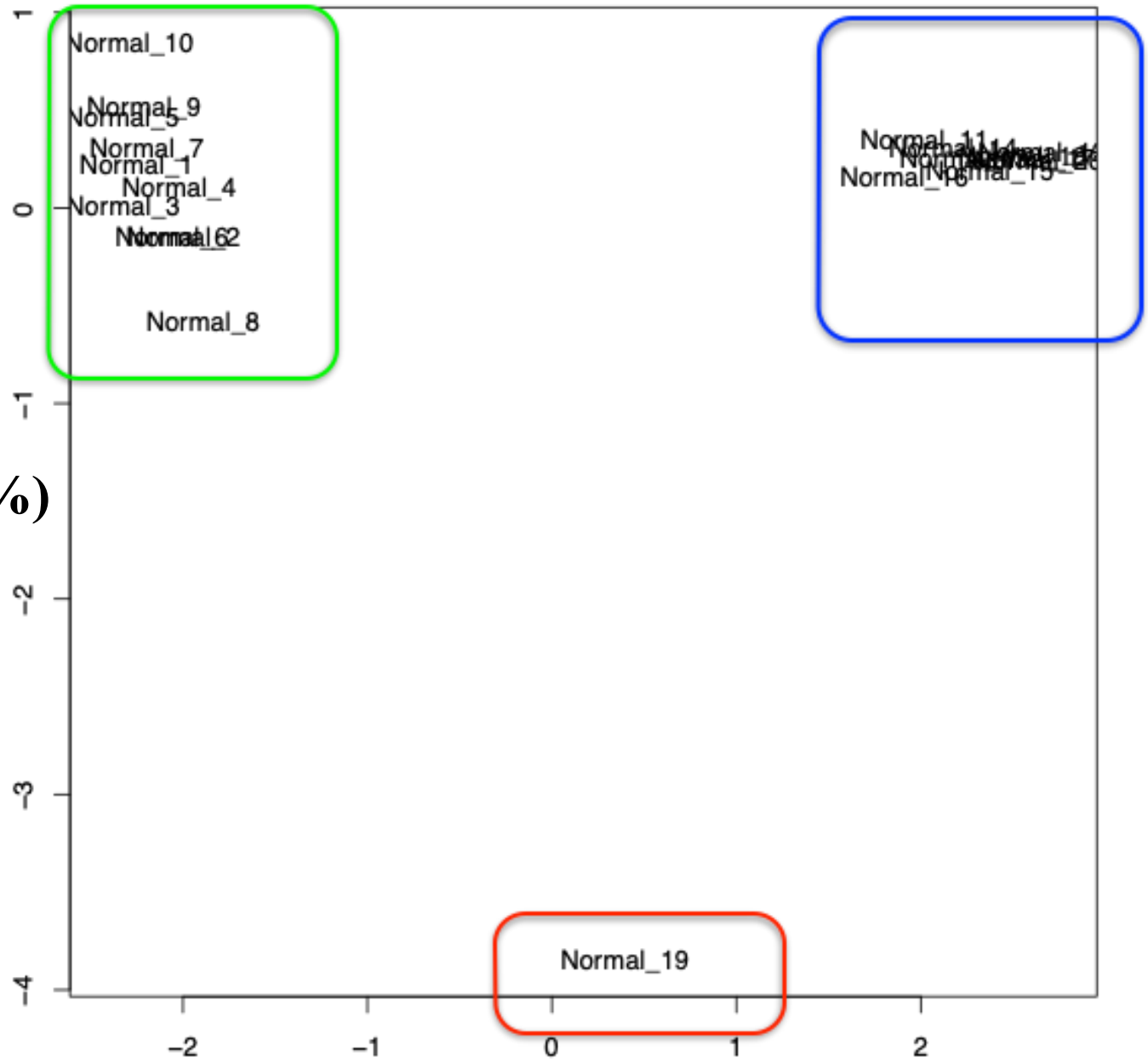
Best likelihood

Mclust In R

```
>?mclustBIC
>?Mclust


>BIC <- mclustBIC(df)
>plot(BIC)
>summary(BIC)
>mod1 <- Mclust(df, x = BIC)
>summary(mod1, parameters = TRUE)
>plot(mod1, what = "classification")
```

Once you have the clusters,
what do you do with them ?



PC2 (12.8%)

PC1 (20.5%)

Challenge

Points in plates

1. Import the data from dataClustering.csv
2. What is the dimension of this dataset?
3. How many data point do we have?
4. Evaluate Euclidean distance of points in a plates
5. Classify points to find clusters using hierarchical clustering and the average agglomeration method

Challenge

Points in plates-continuous

6. We expect to have 3 clusters. When you apply k-means algorithm using 1 iteration, does it differ from applying it using 10 or 100 iterations?

7. What is the outcome of the C-means clustering?

install.packages("e1071")

library(e1071)

?cmeans

Challenge

Points in plates-continuous

Library(mclust)

8. What are the top 3 models *mclustBIC* function suggests based on the BIC criterion?

9. How many clusters did it find using the top model?

10. Plot the outcome

Thank you for your attention