

La cellule Le chromosome l'ADN, Le gène la protéine la maladie

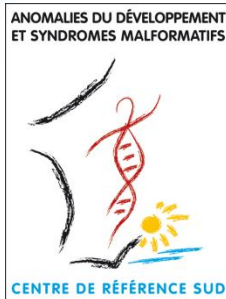
Docteur David Geneviève, MCU-PH
Génétique Médicale

Centre de Référence Anomalies du Développement

06/12/2011



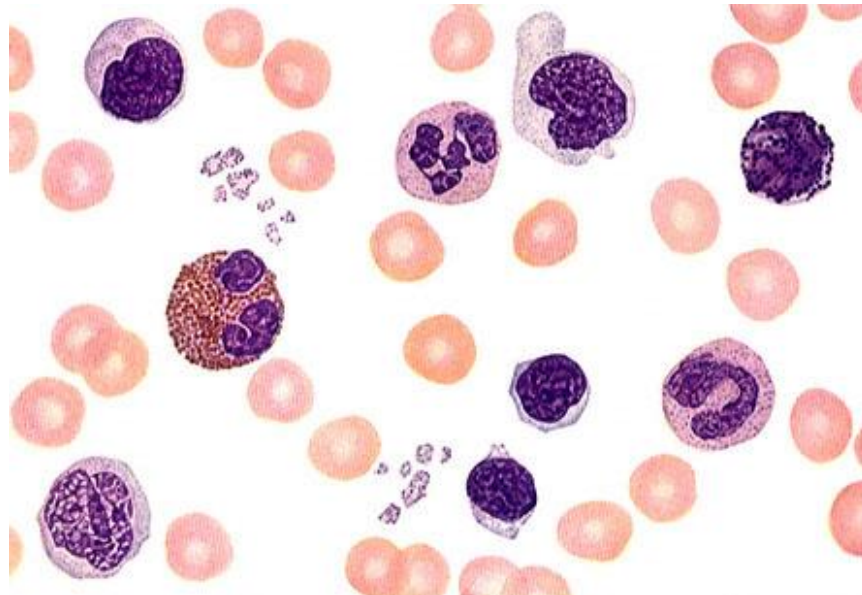
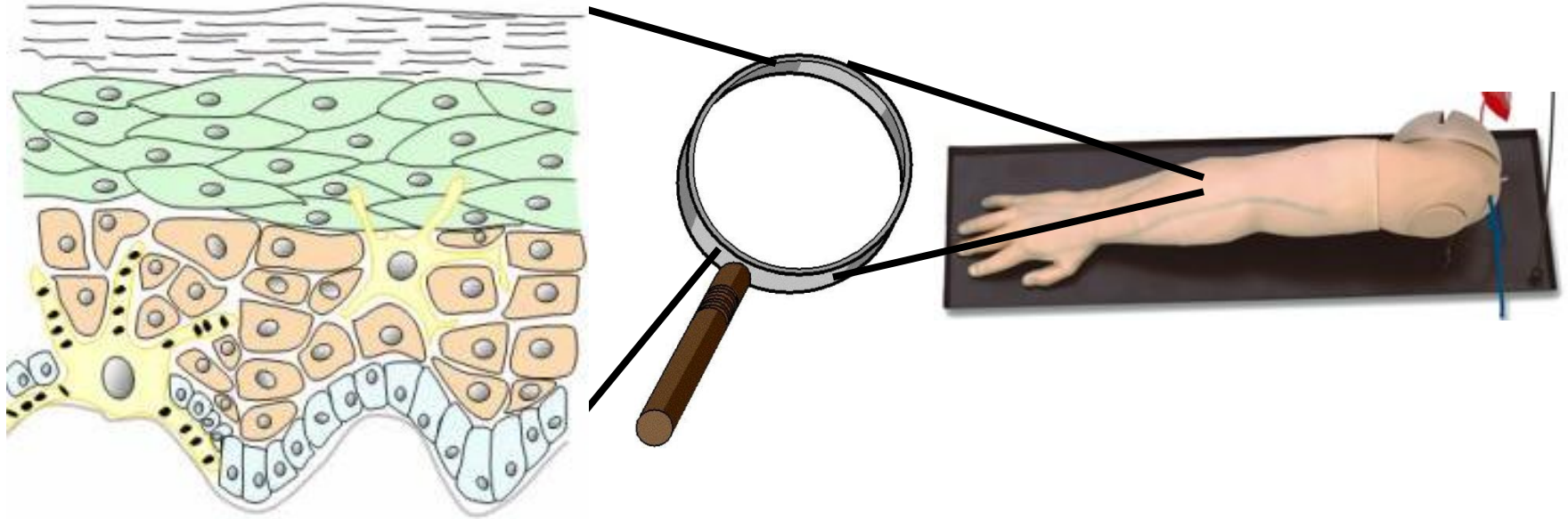
CENTRE DE REFERENCE
Anomalies du développement
et syndromes malformatifs



Réseau
vivre
avec une Anomalie
du Développement
en Languedoc Roussillon



La cellule

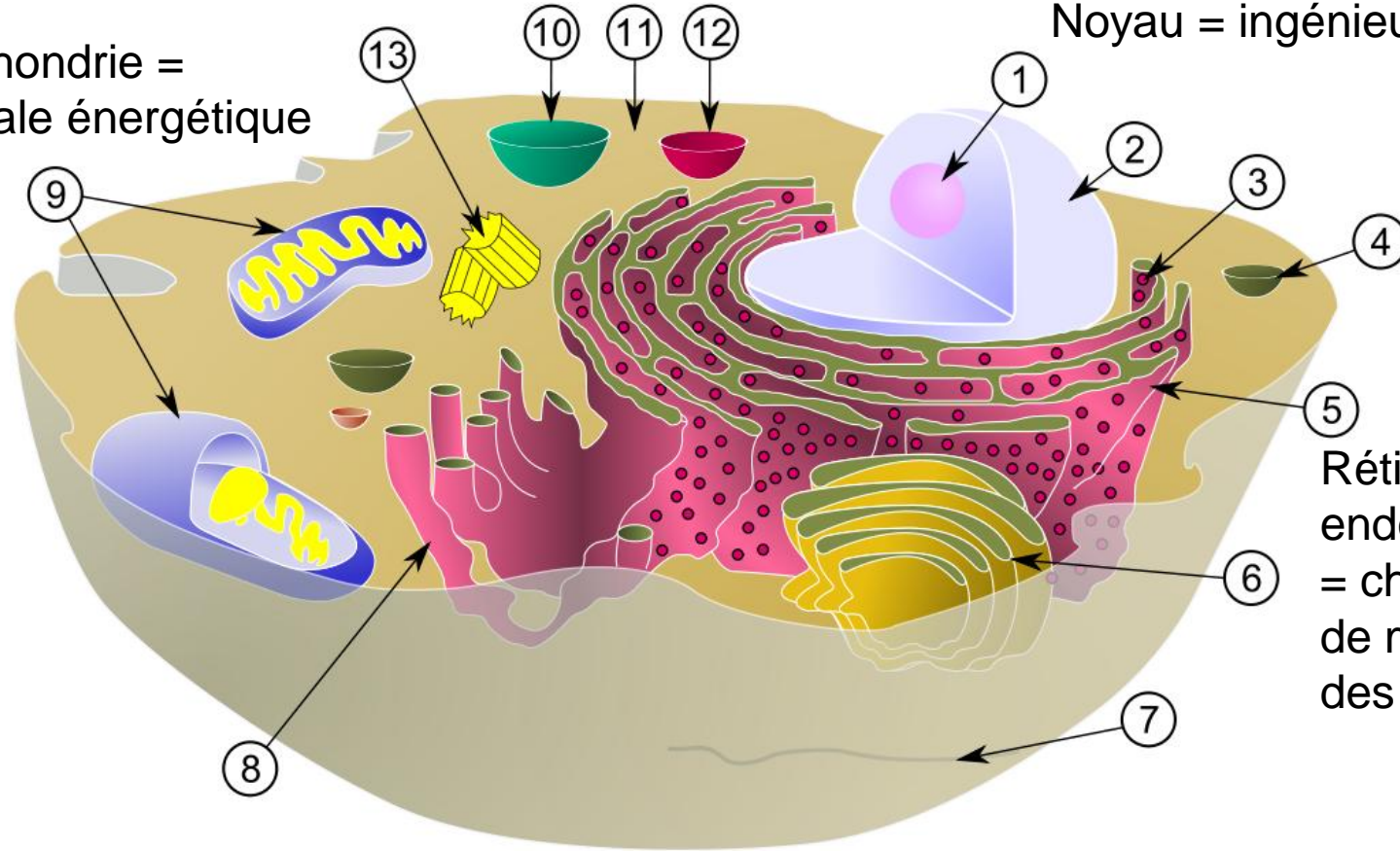


La cellule

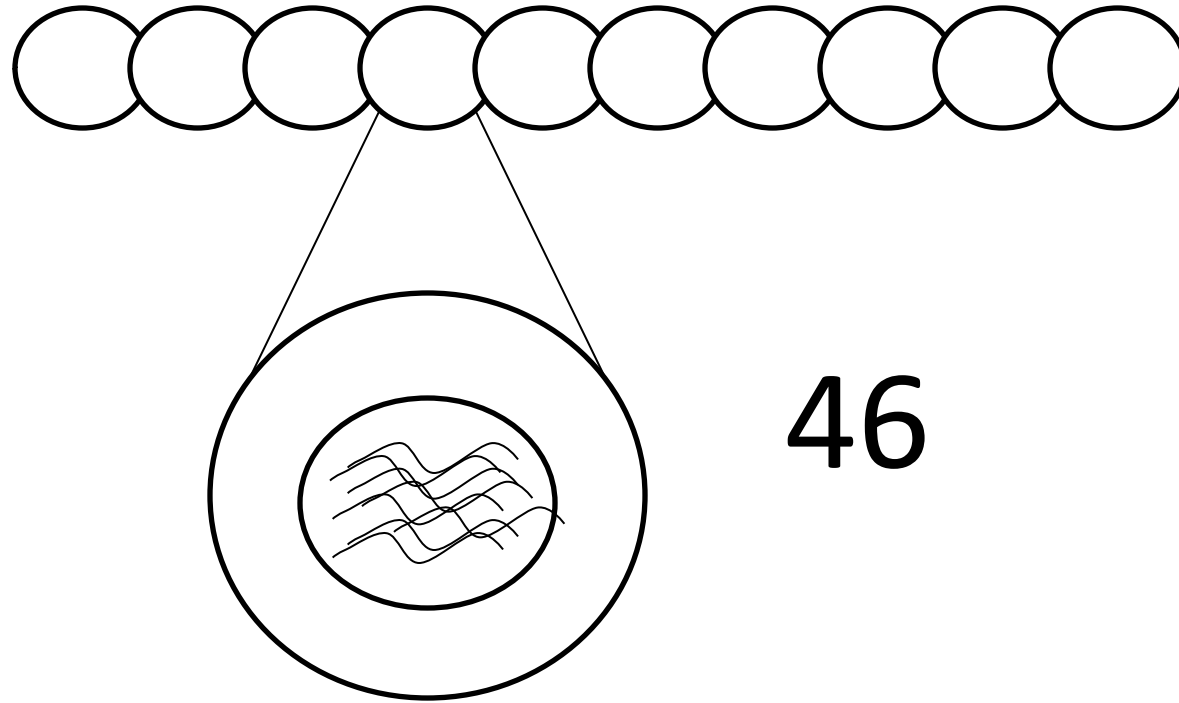
Lysosome =
déchetterie

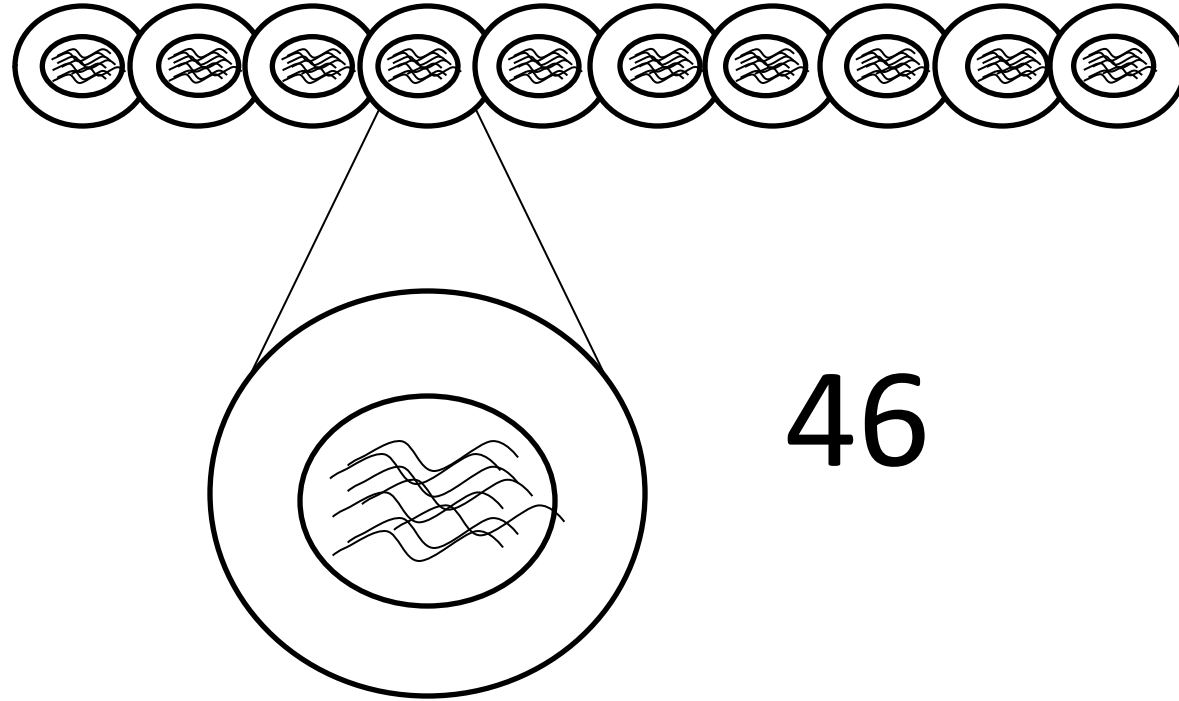
Noyau = ingénieurs

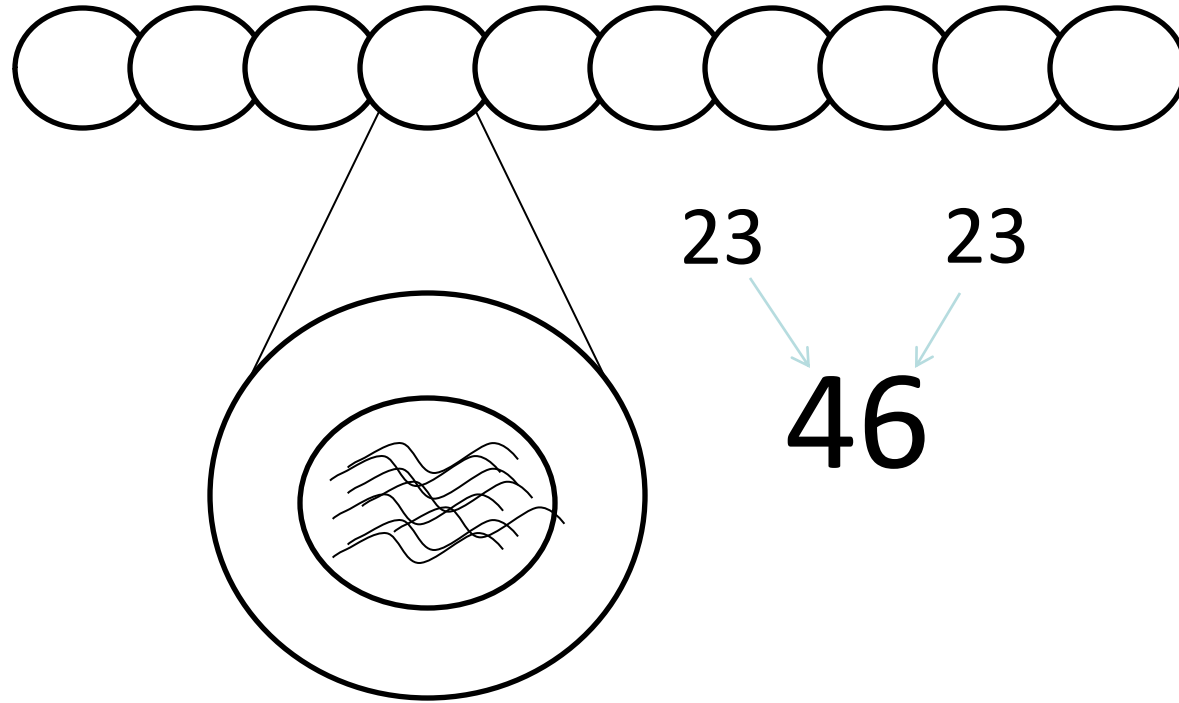
Mitochondrie =
Centrale énergétique



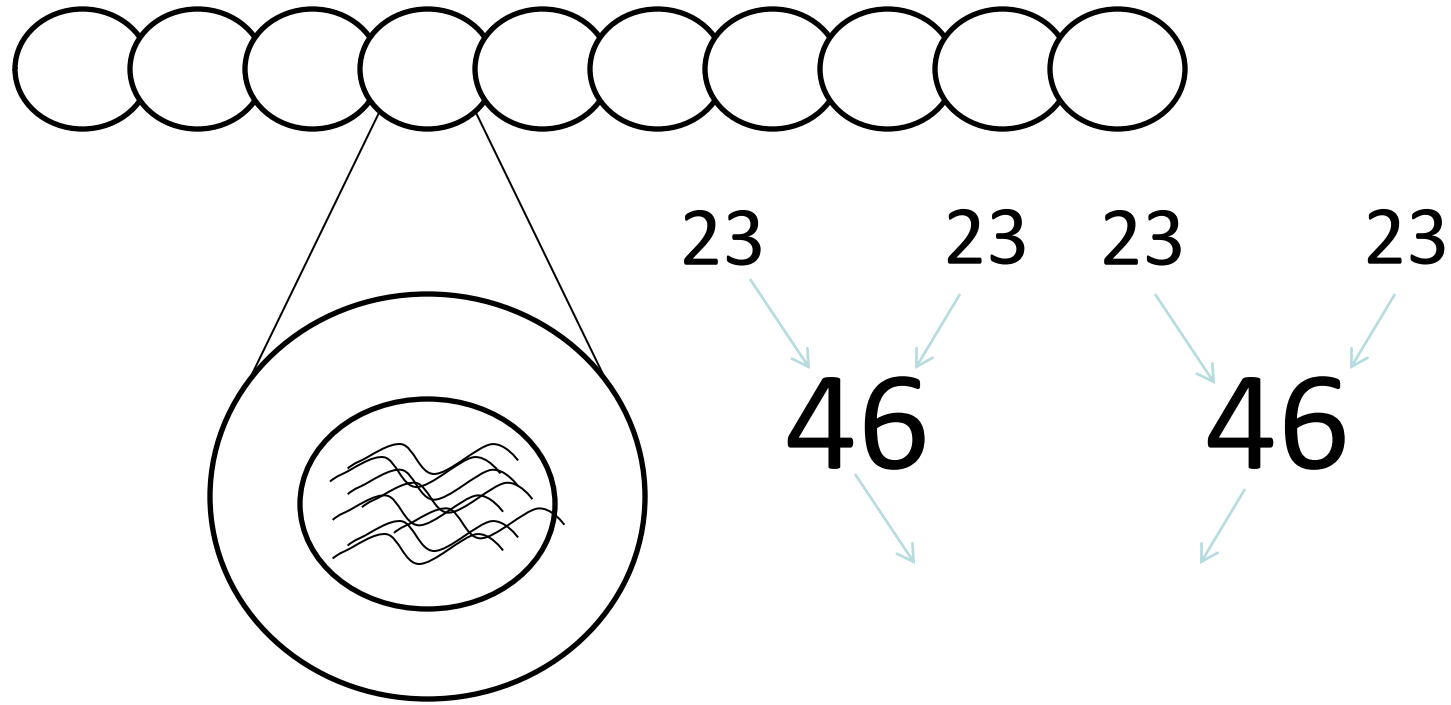
Réticulum
endoplasmique
= chaîne
de montage
des protéines



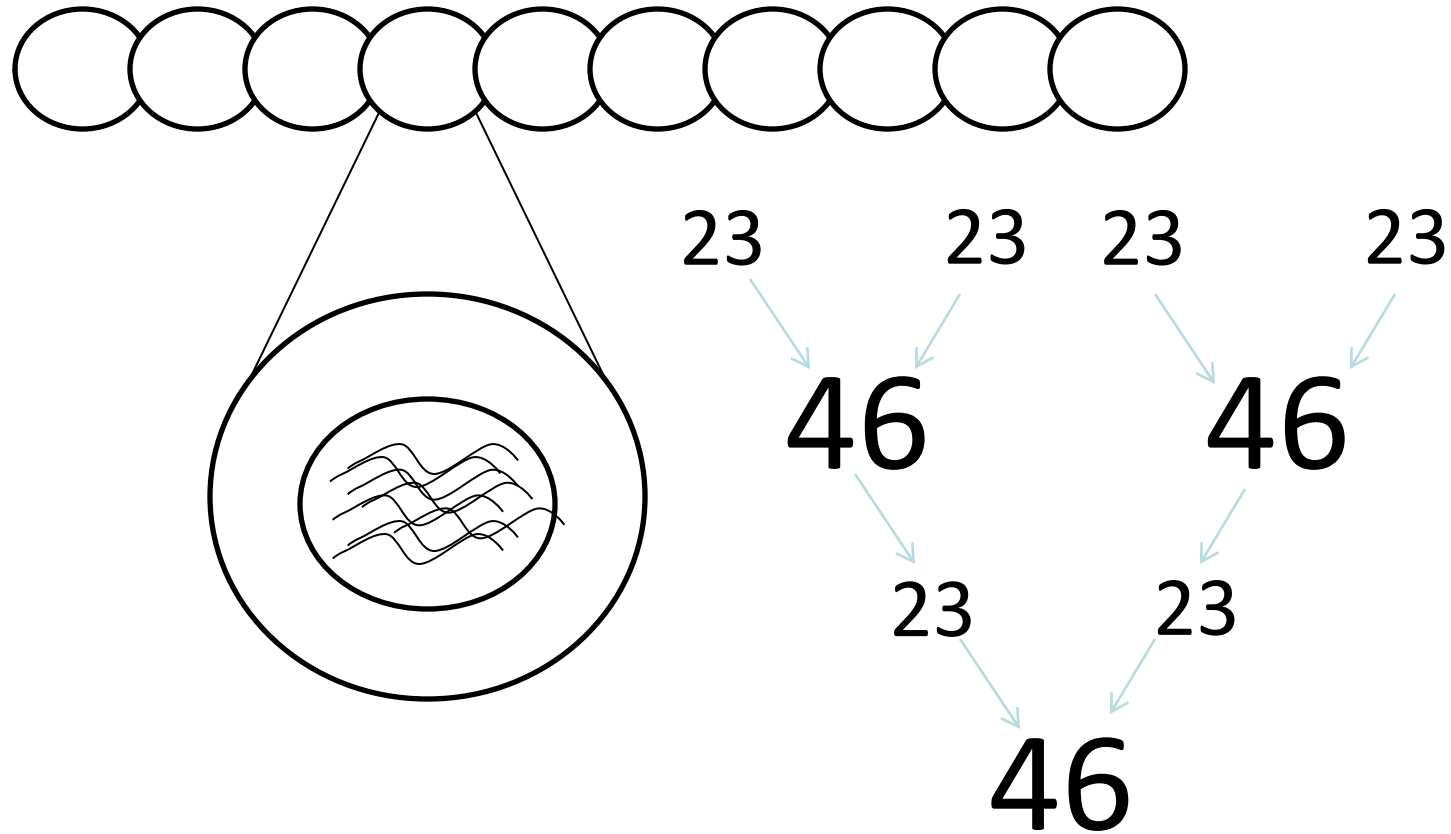




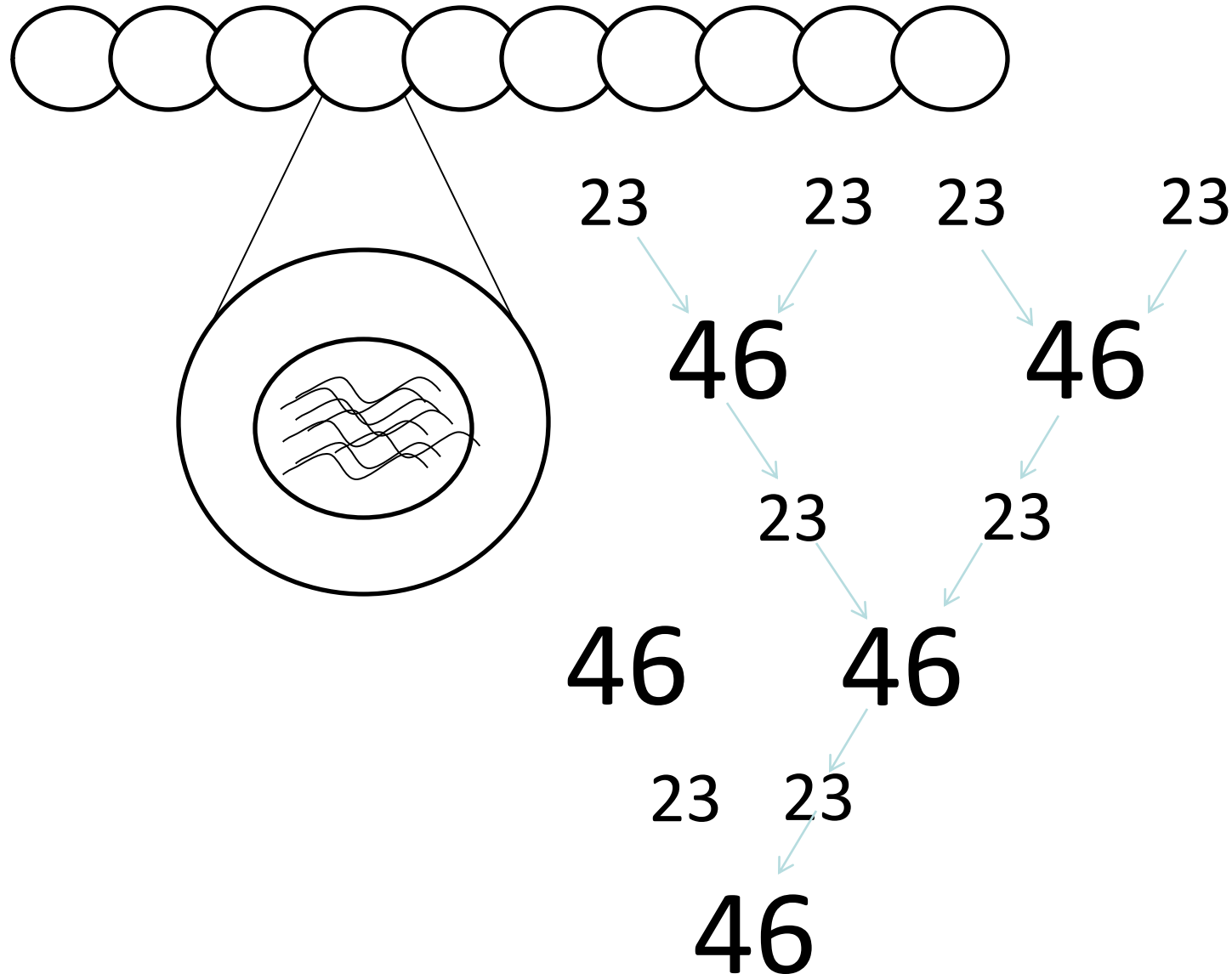
L'ADN



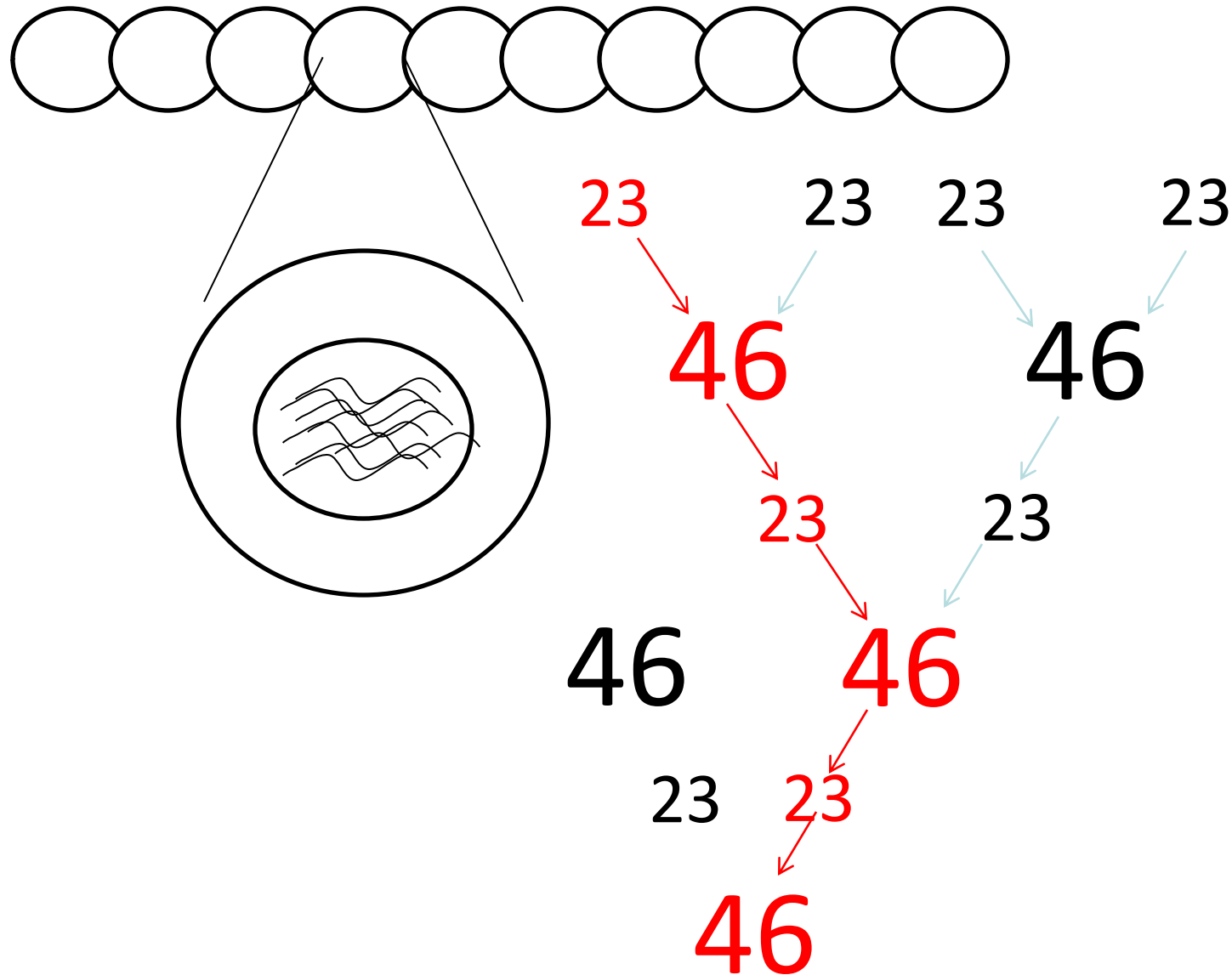
L'ADN



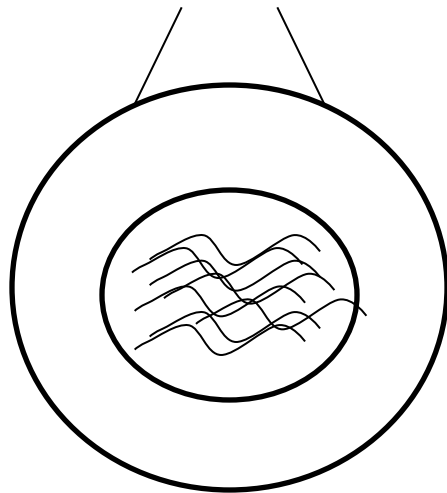
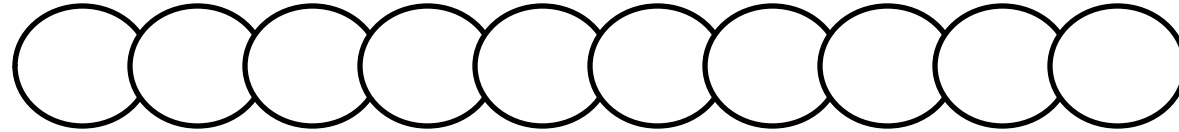
L'ADN



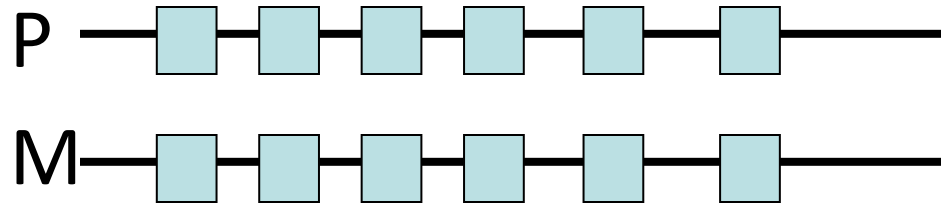
L'ADN



L'ADN et les gènes



46

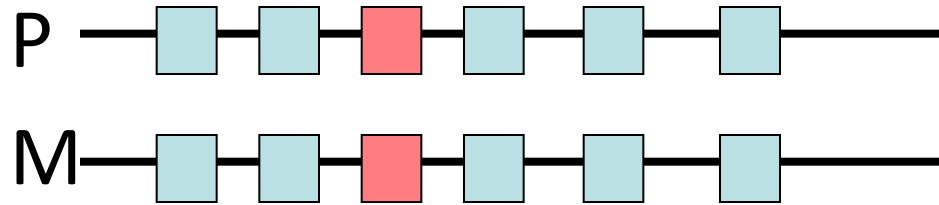
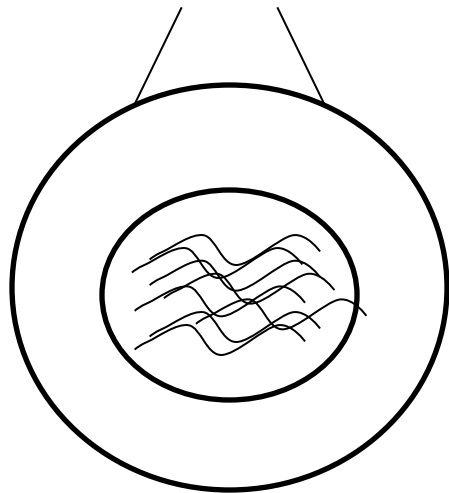
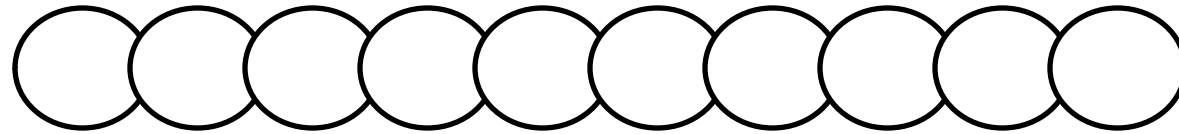


2x20 000 gènes

100 000 fonctions

Chaque gène code pour
une ou plusieurs fonctions

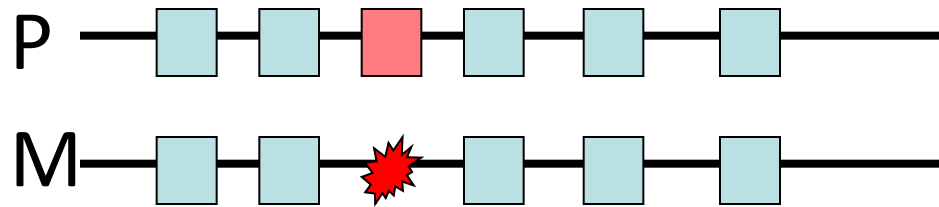
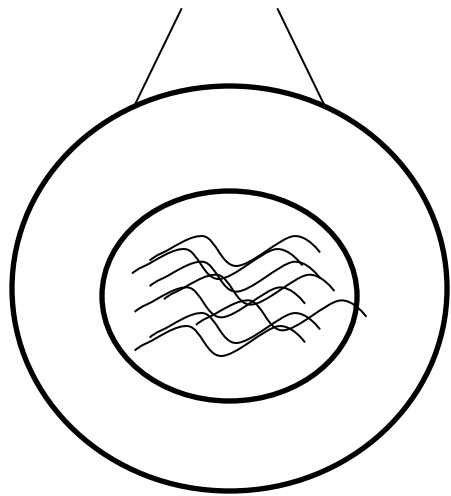
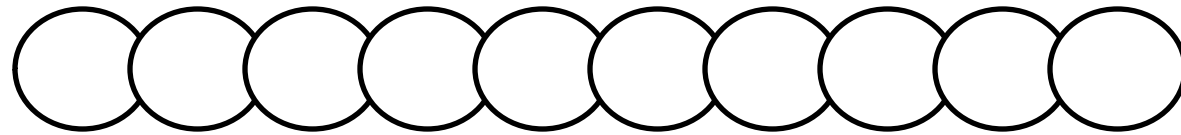
L'ADN et les gènes



100 %

46

L'ADN et les gènes



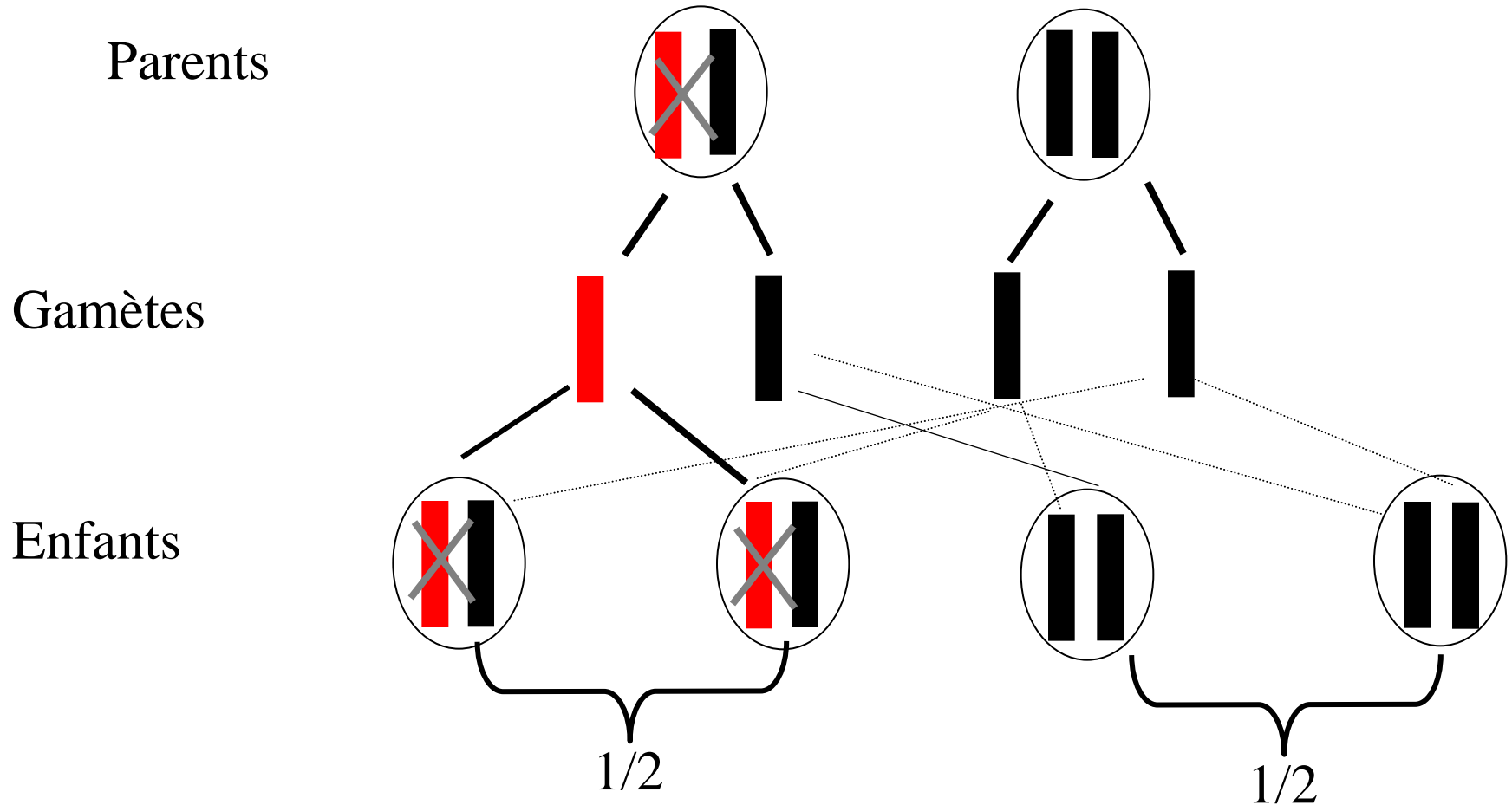
~~100 %~~

50 %

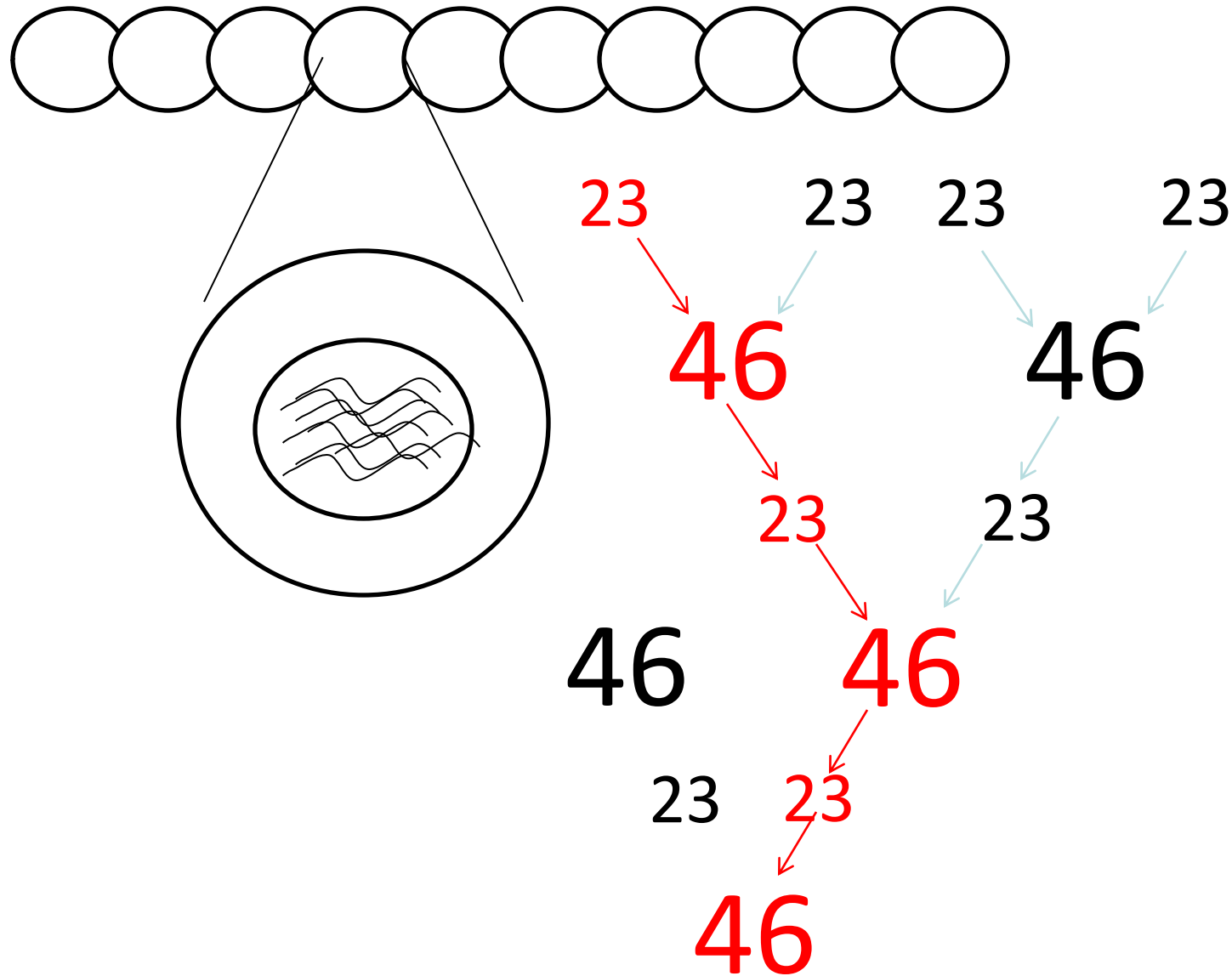
46

Maladie génétique dominante

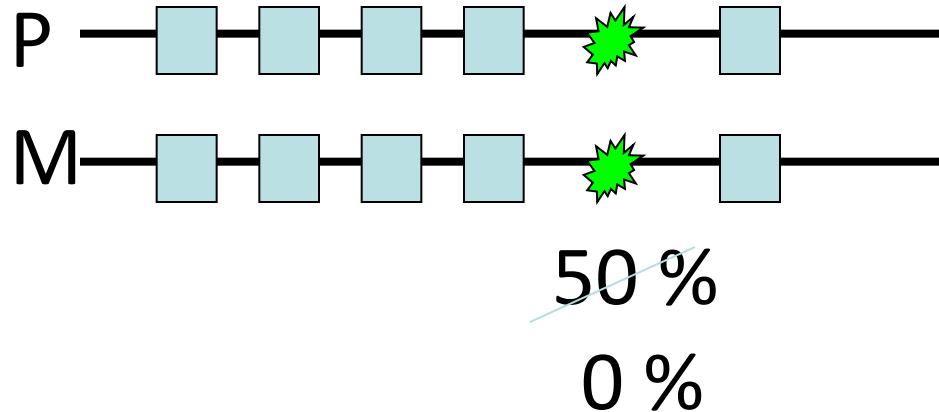
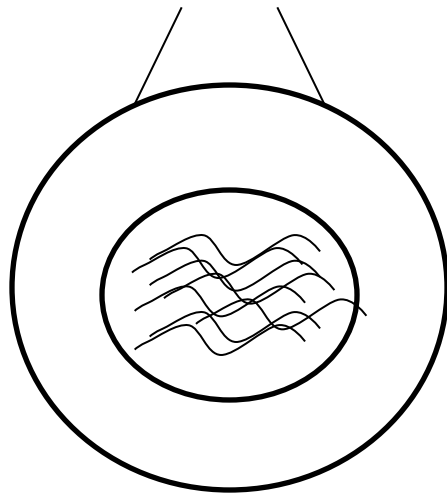
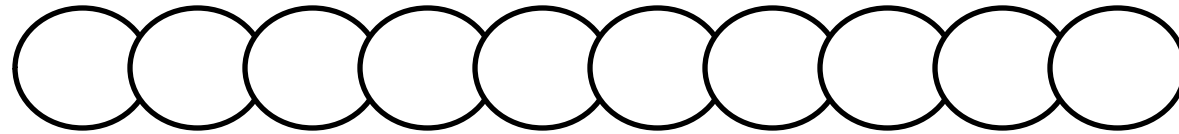
Pathologie dominante



L'ADN

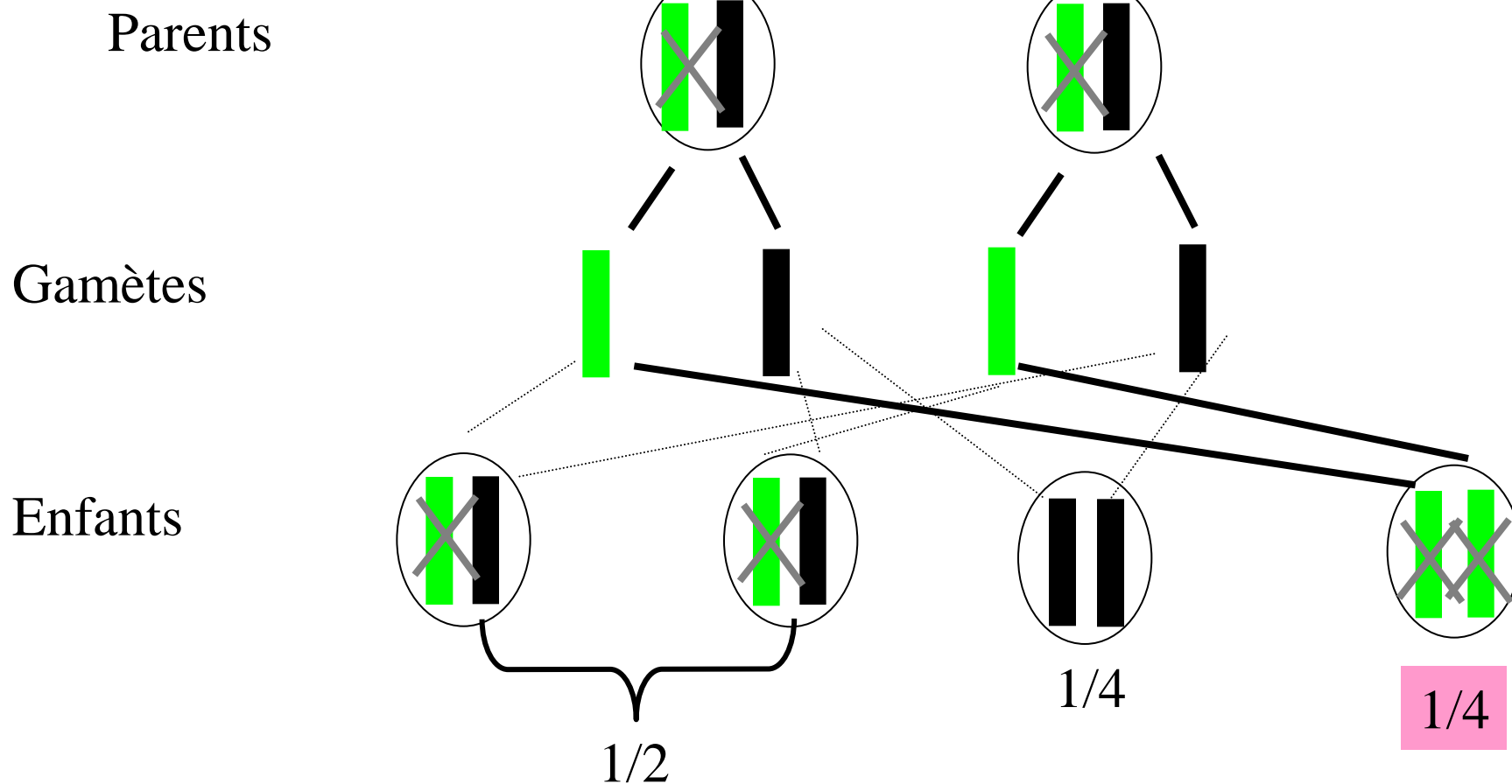


L'ADN et les gènes

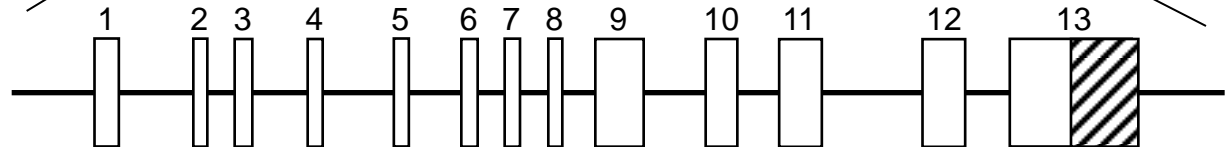
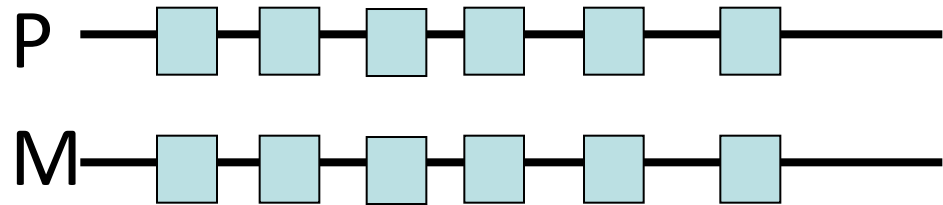
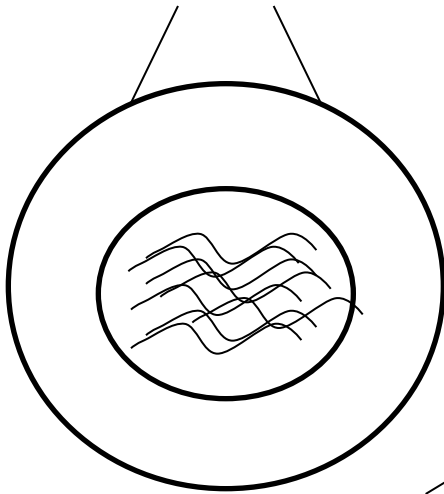
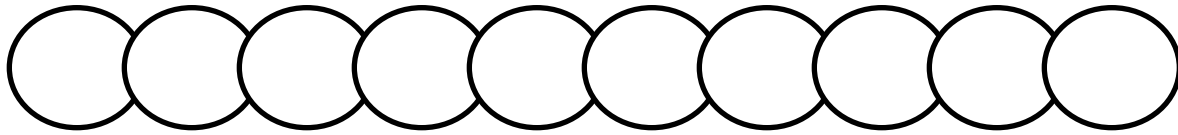


Maladie génétique Récessive

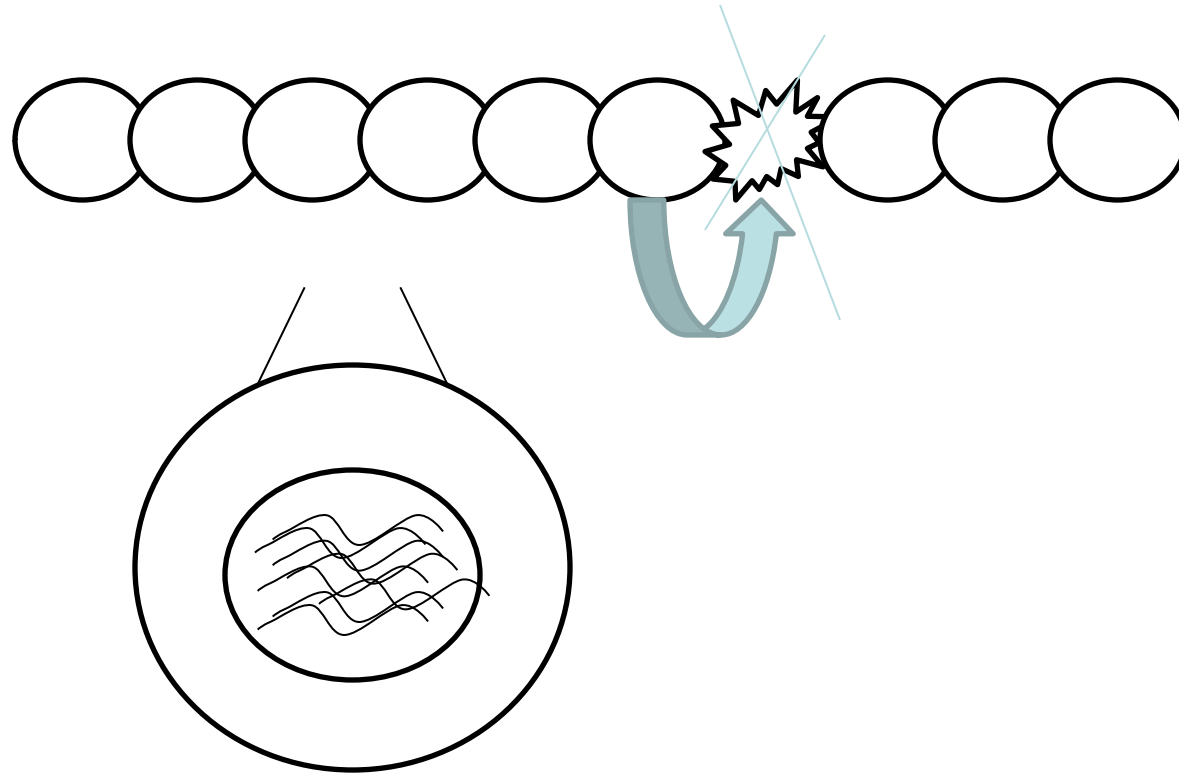
Pathologie récessive



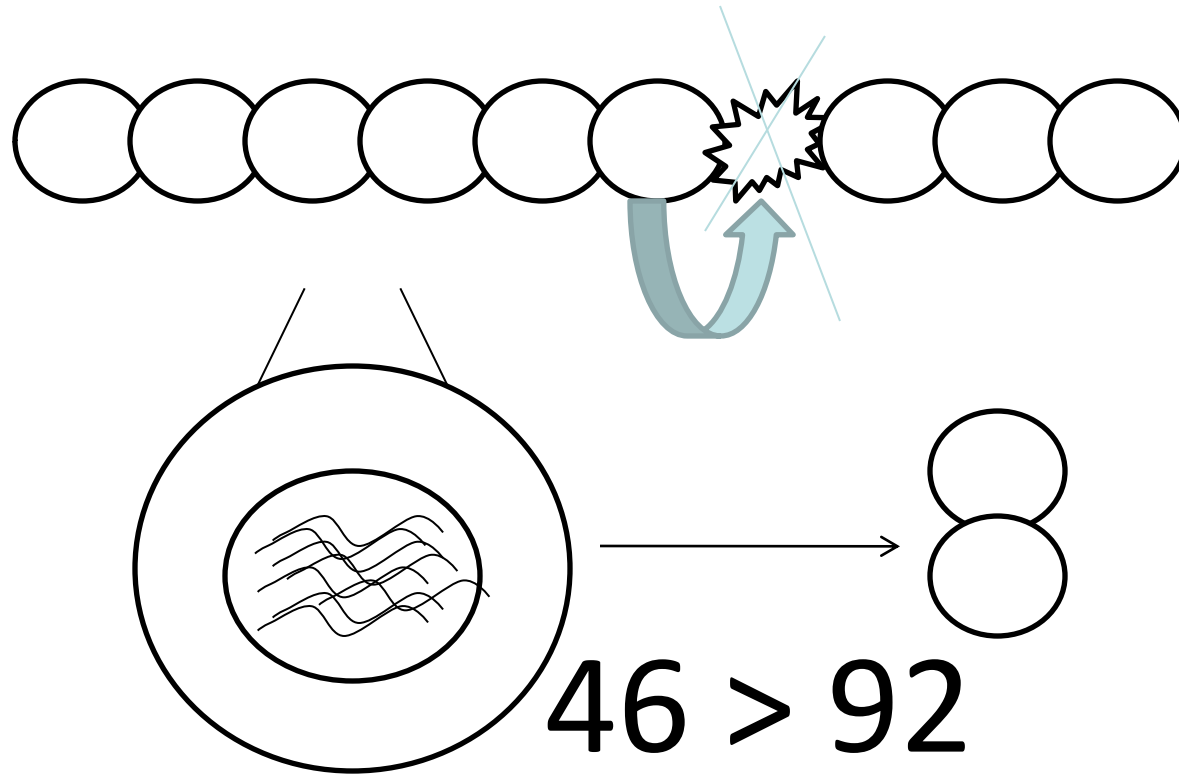
Anatomie d'un gène



L'ADN et les chromosomes



L'ADN et les chromosomes



Anatomie d'un chromosome



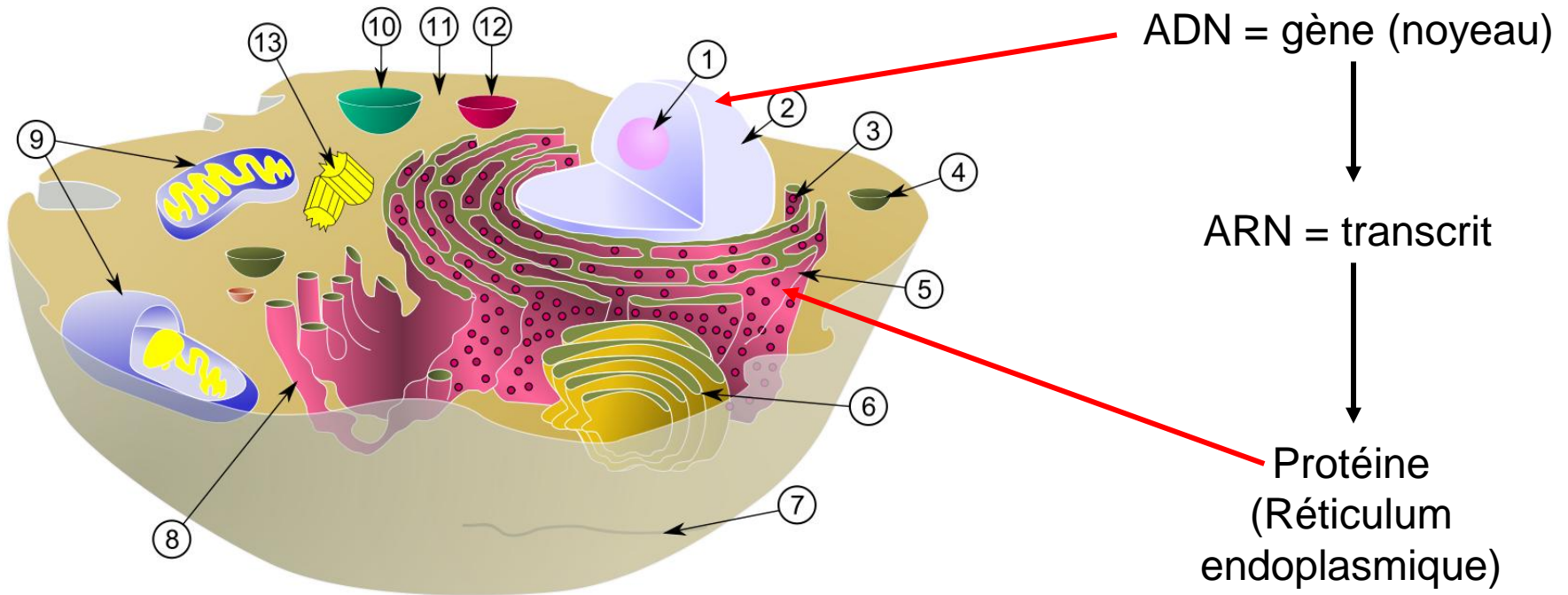
Gonosomes =
Chromosomes « sexuels »

Anatomie d'un chromosome

- Caryotype = analyse des chromosome uniquement
- Etude d'un gène = analyse spécifique

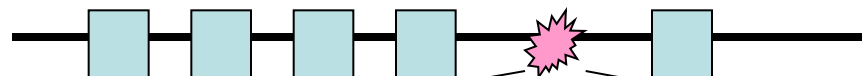


L'ADN – l'ARN- la protéine



Conséquence d'une mutation dans un gène

Mutation dans un gène



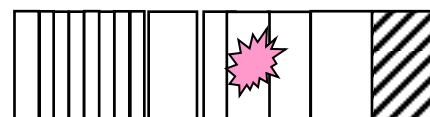
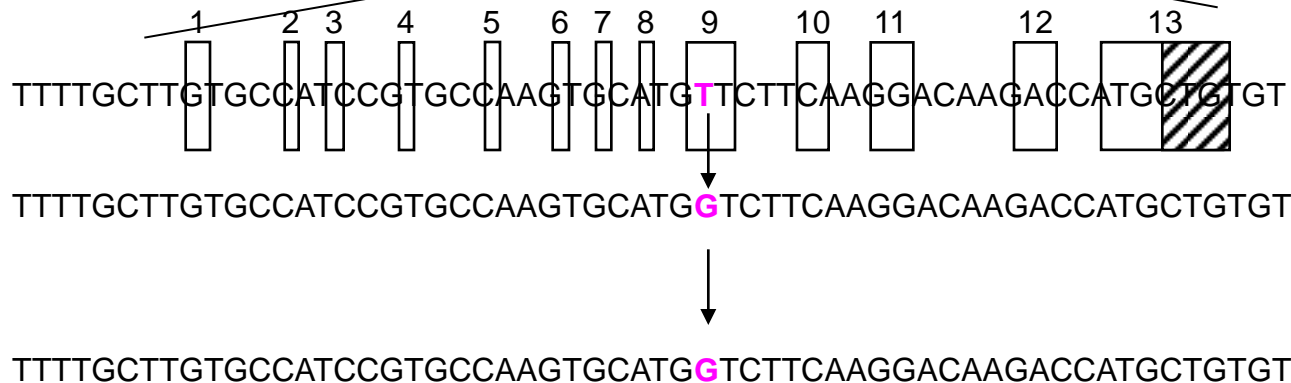
ADN = gène (noyau)



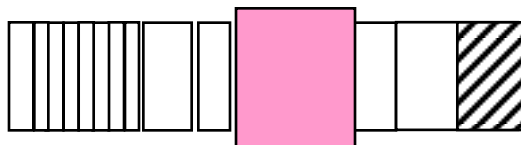
ARN = transcrit



Protéine
(Réticulum endoplasmique)



Gain de fonction



= expression de la maladie génétique

Perte de fonction

