



Hémochromatose: L'important et le nouveau en 2021



DES Hématologie
21 mai 2021



Hémochromatose

Définition et physiopathologie

Diagnostic

Traitement

Prévention

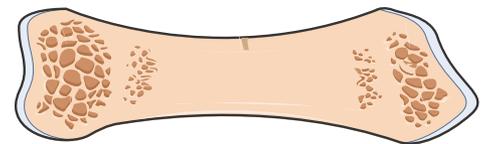
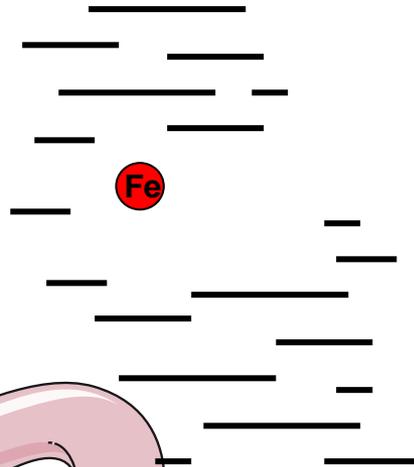
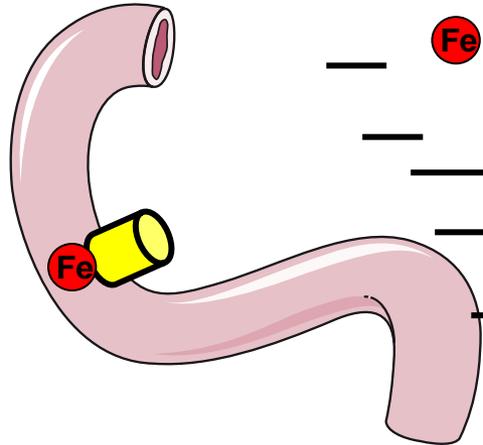
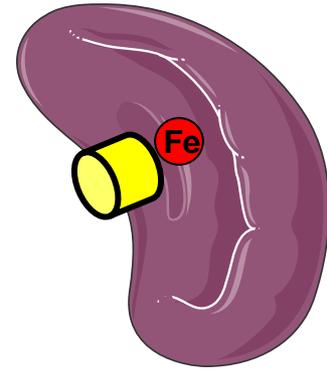
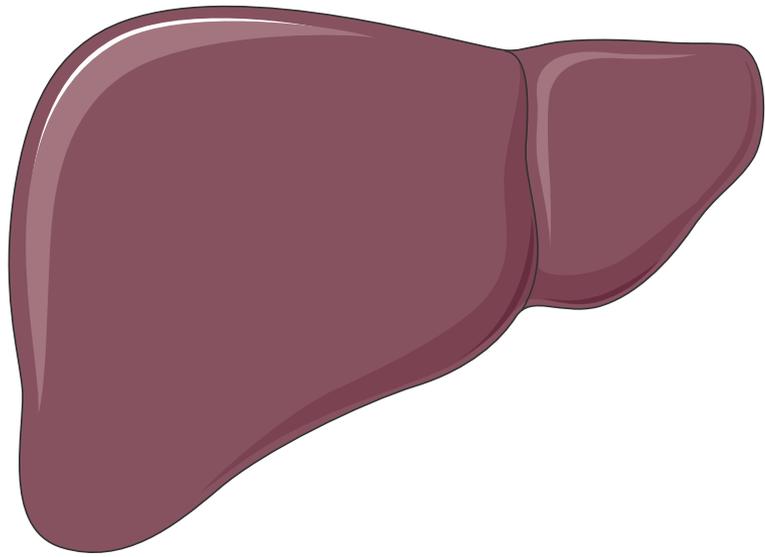
Hémochromatose

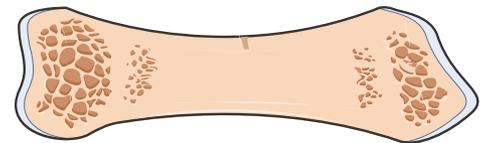
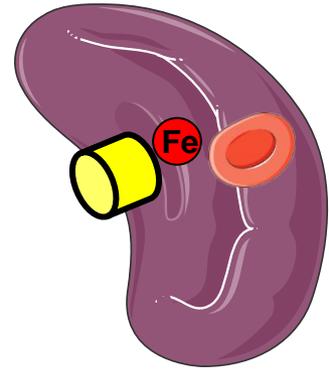
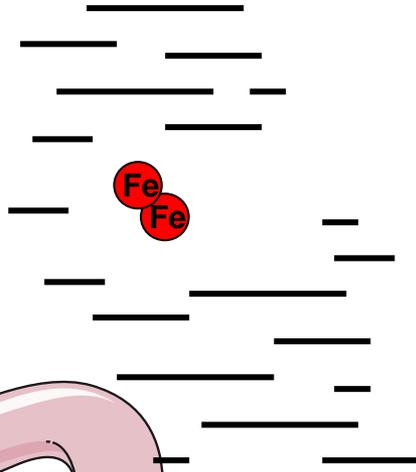
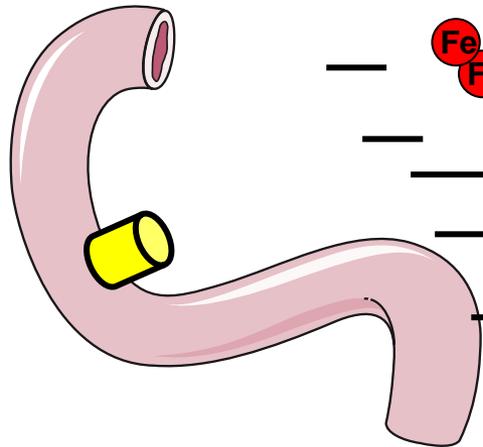
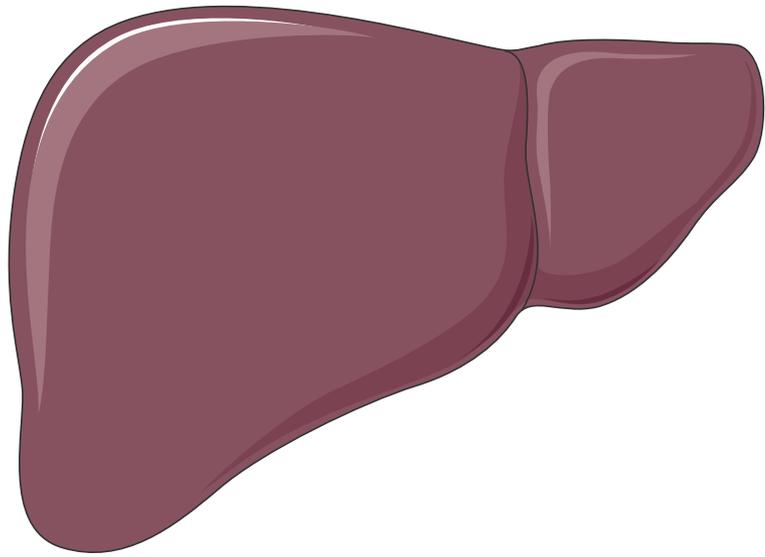
Surcharge en fer

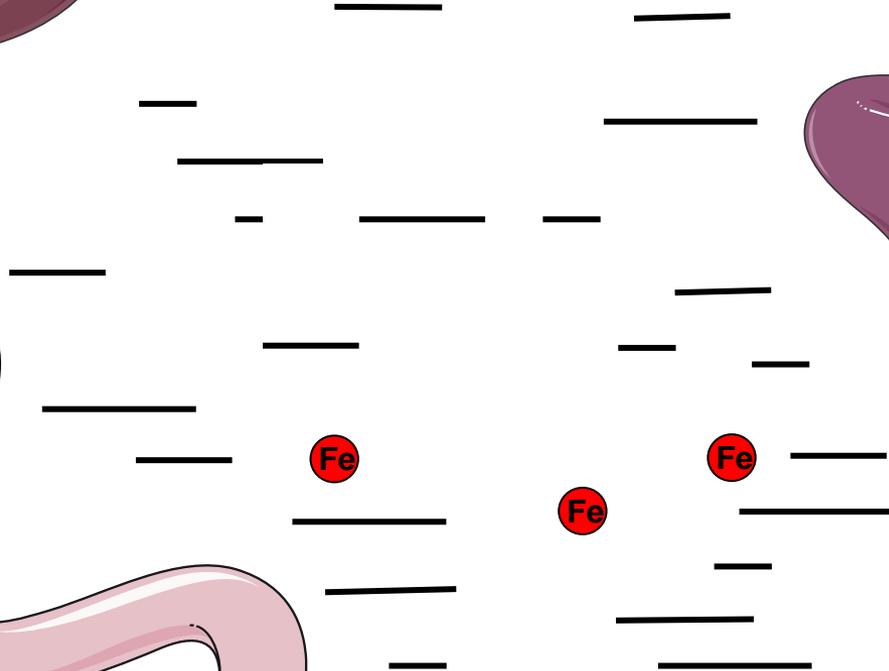
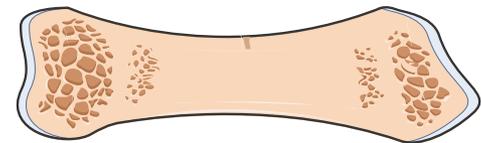
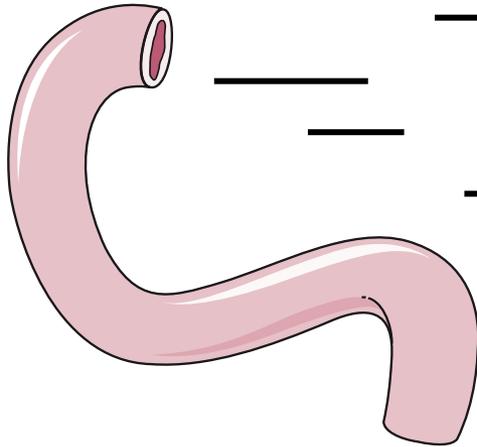
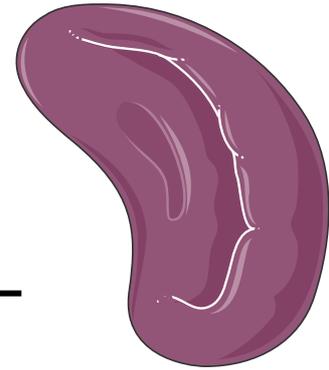
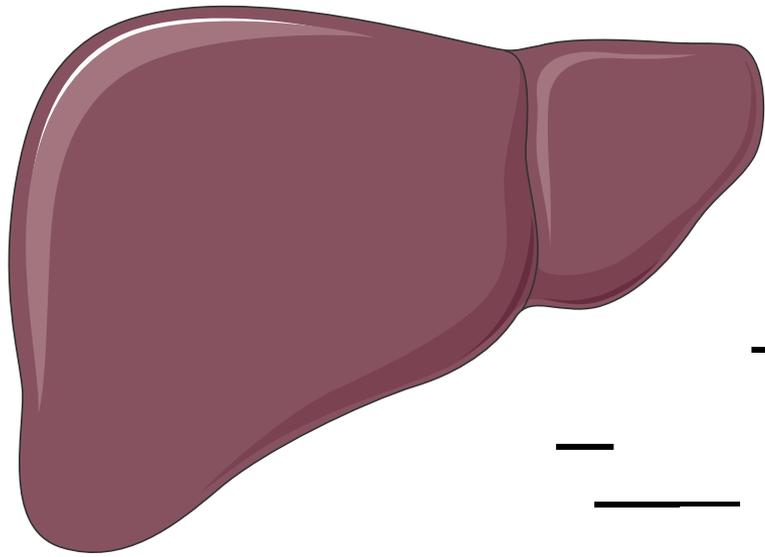
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graph TD; A[Surcharge en fer] --> B[Génétique]; A --> C[Déficiencia en hepcidine];
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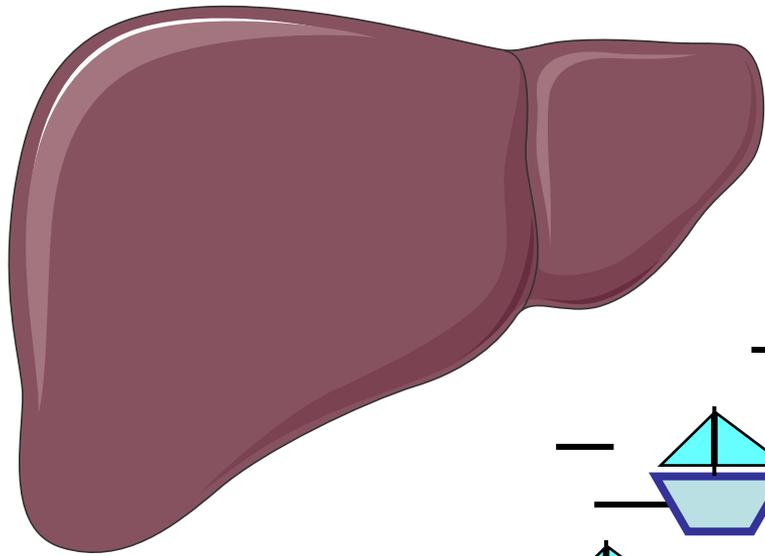
Génétique

**Déficiencia en
hepcidine**

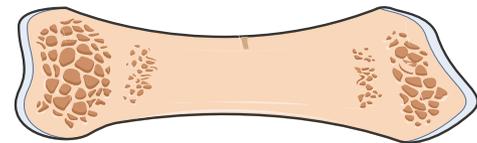
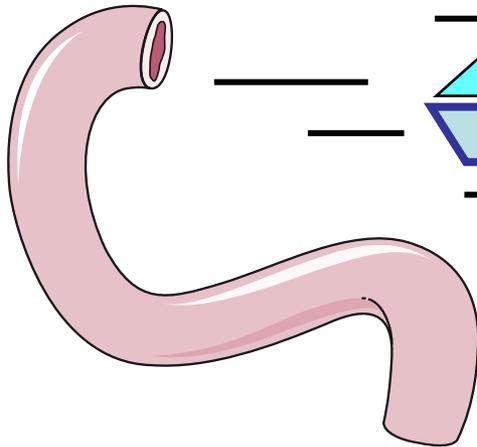
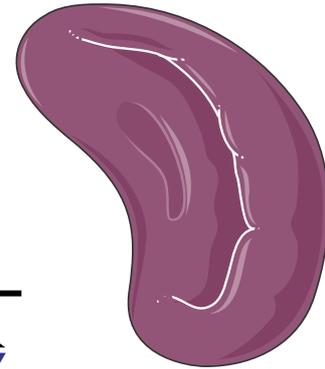
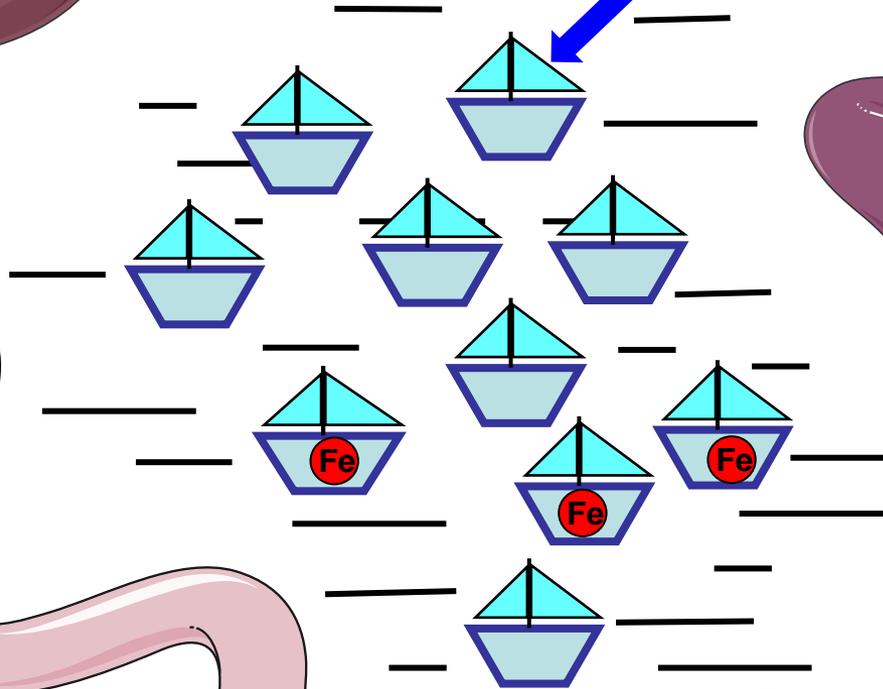
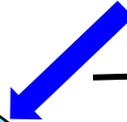


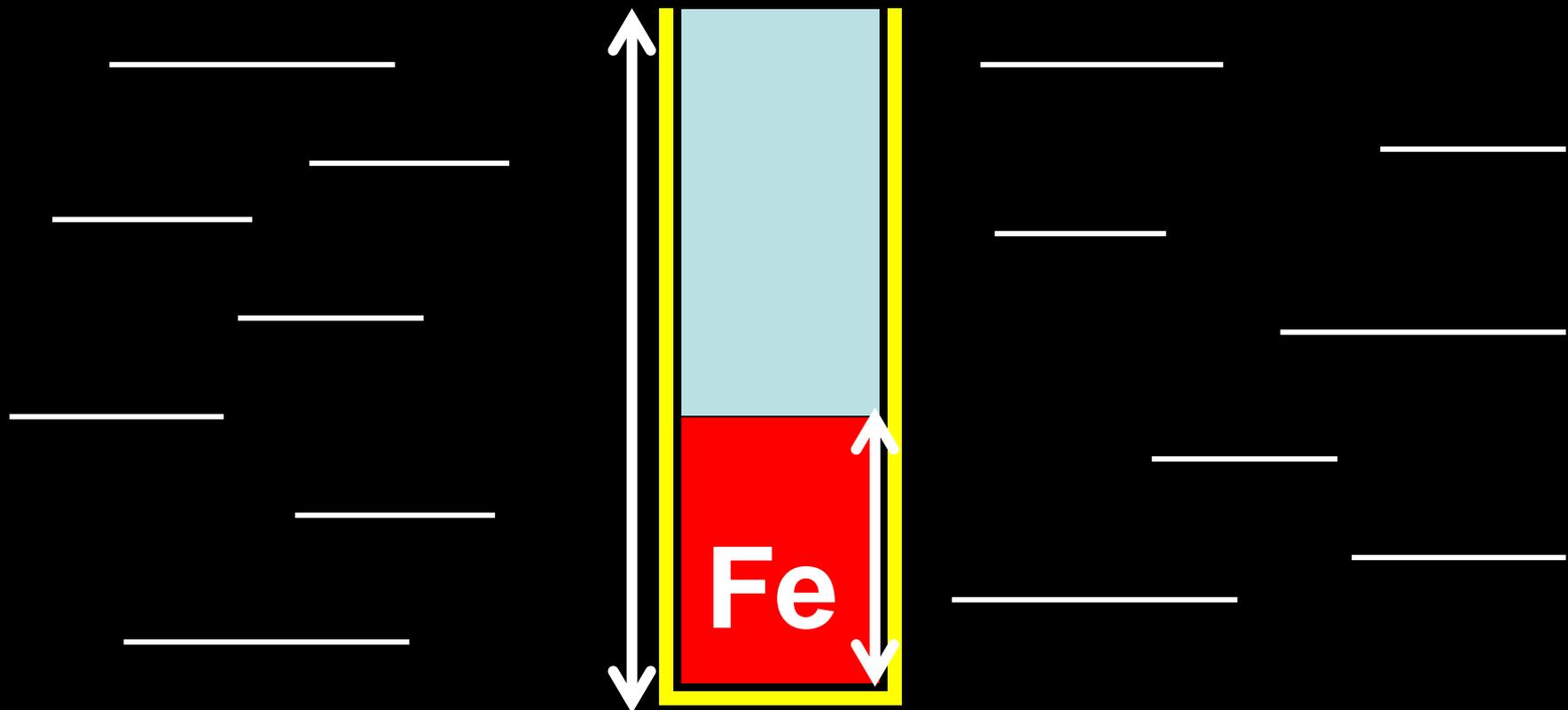




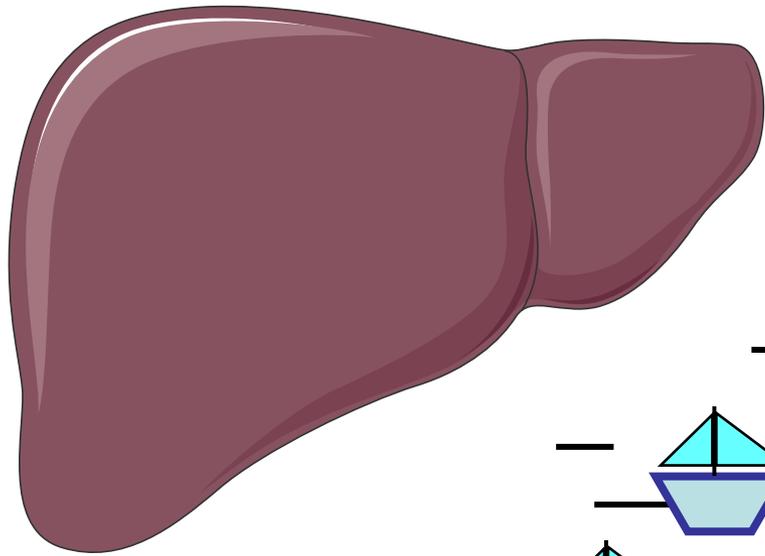


Transferrine

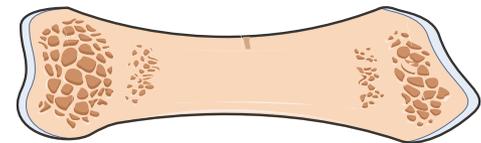
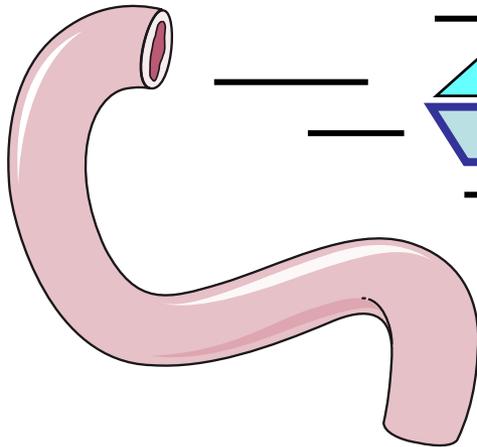
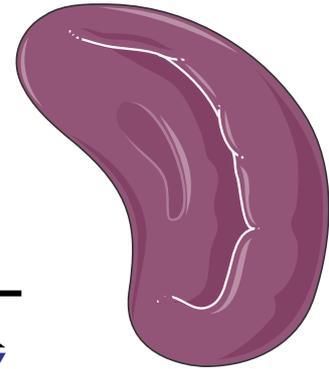
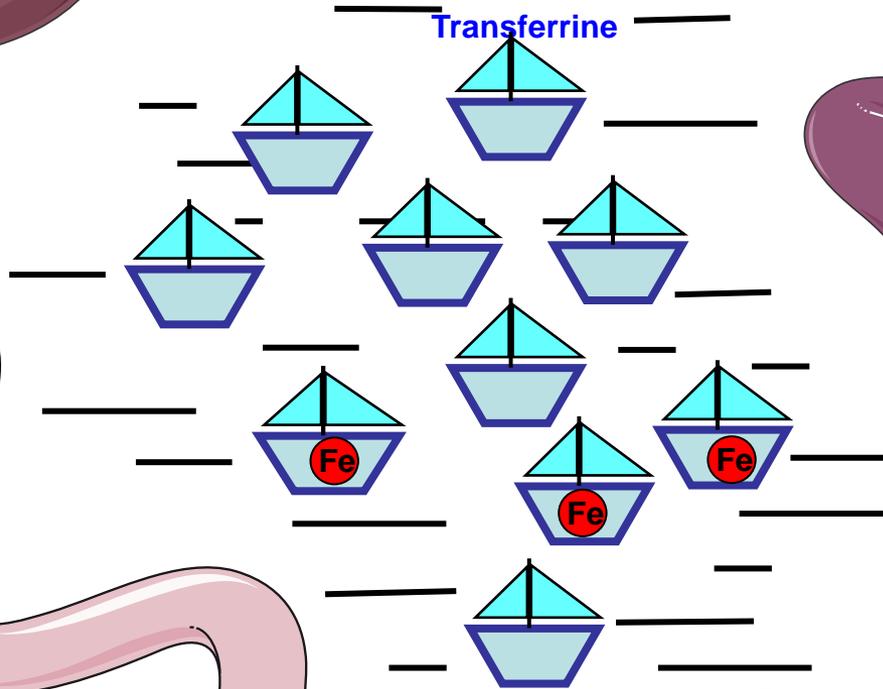


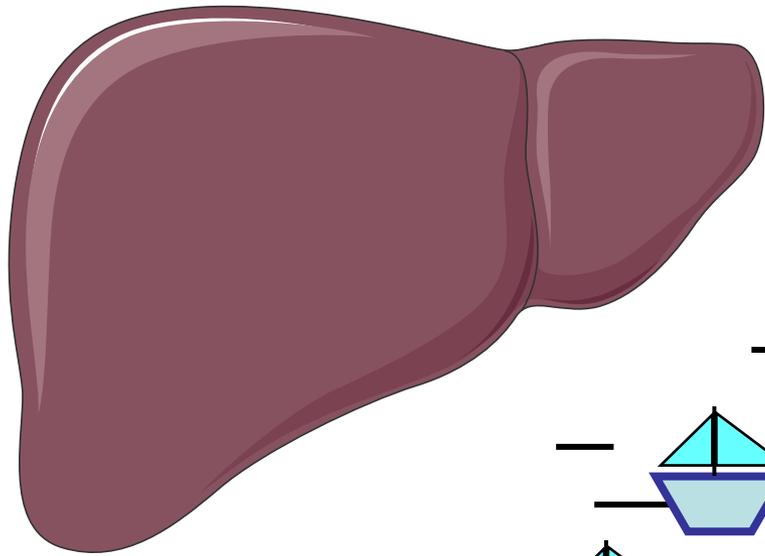


Saturation transferrine = $Fe/Tf < 45\%$

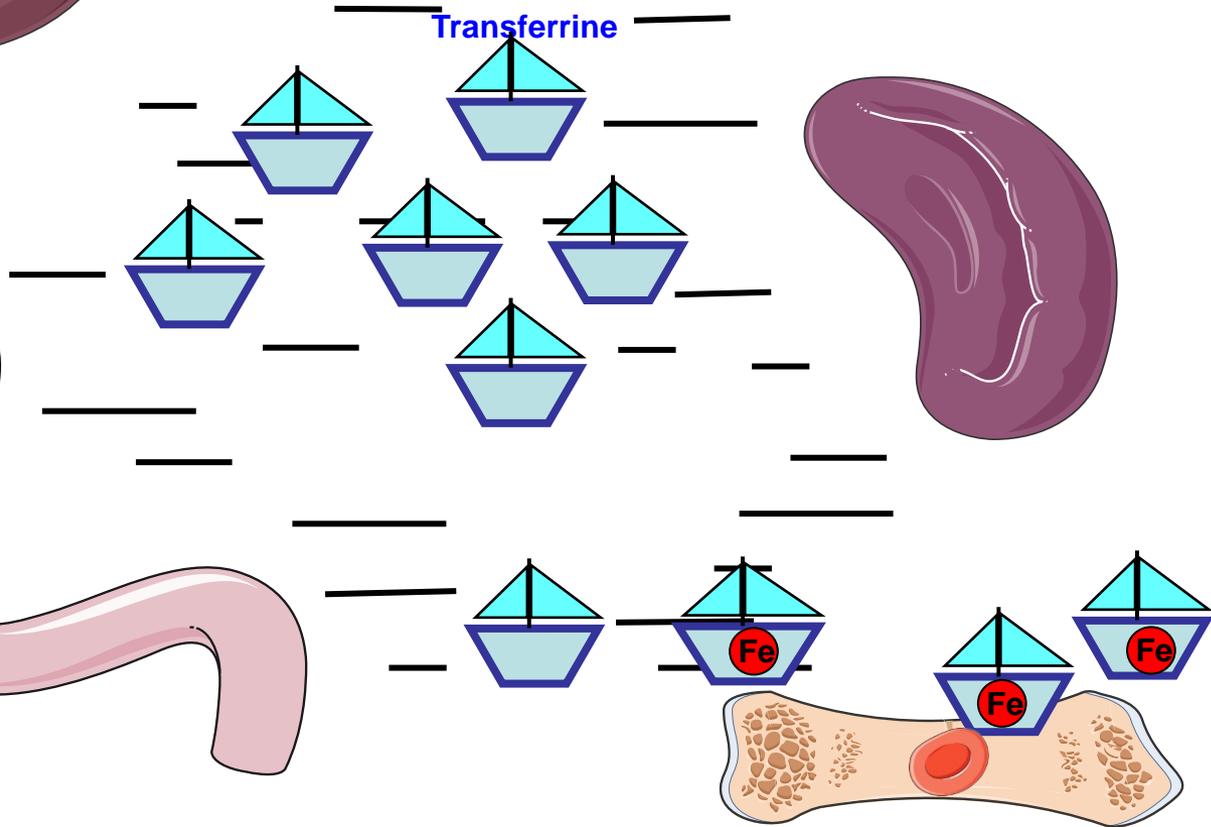
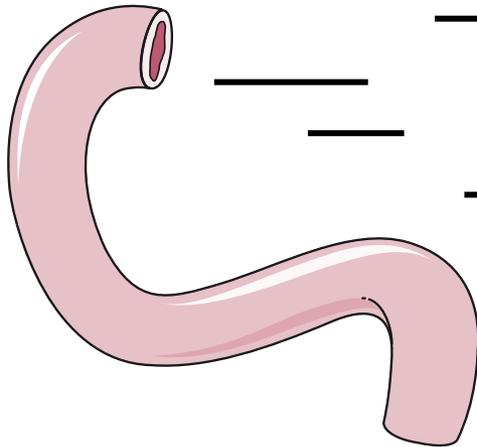
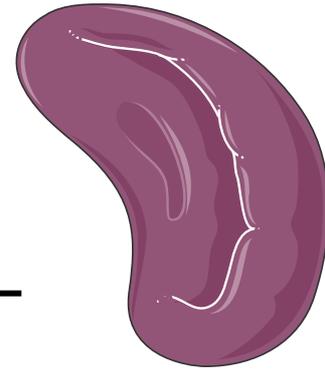


Transferrine





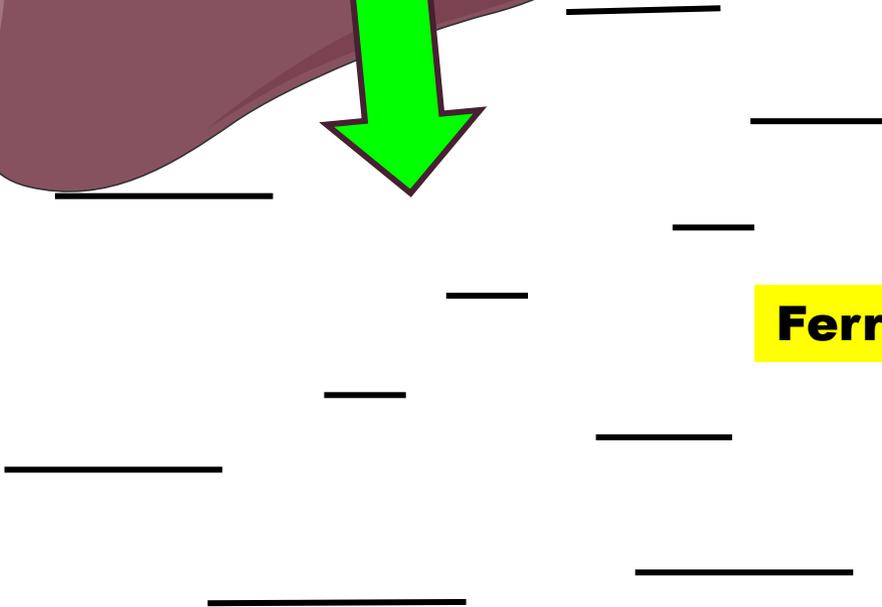
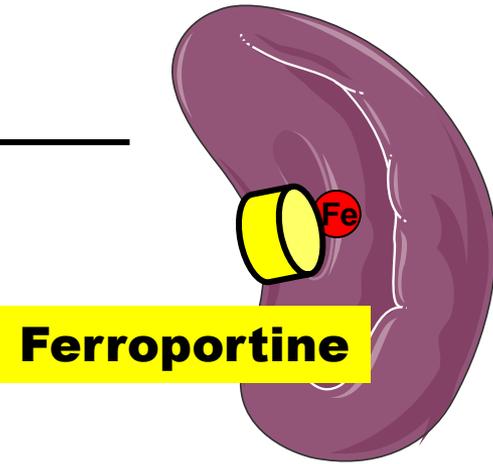
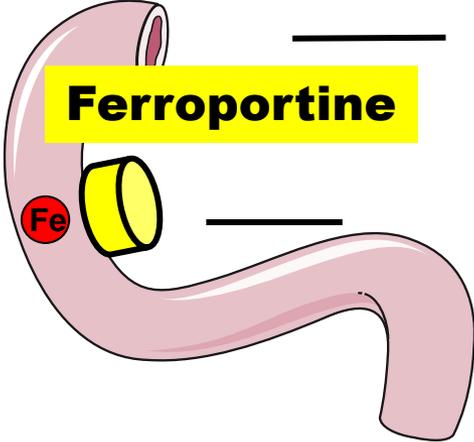
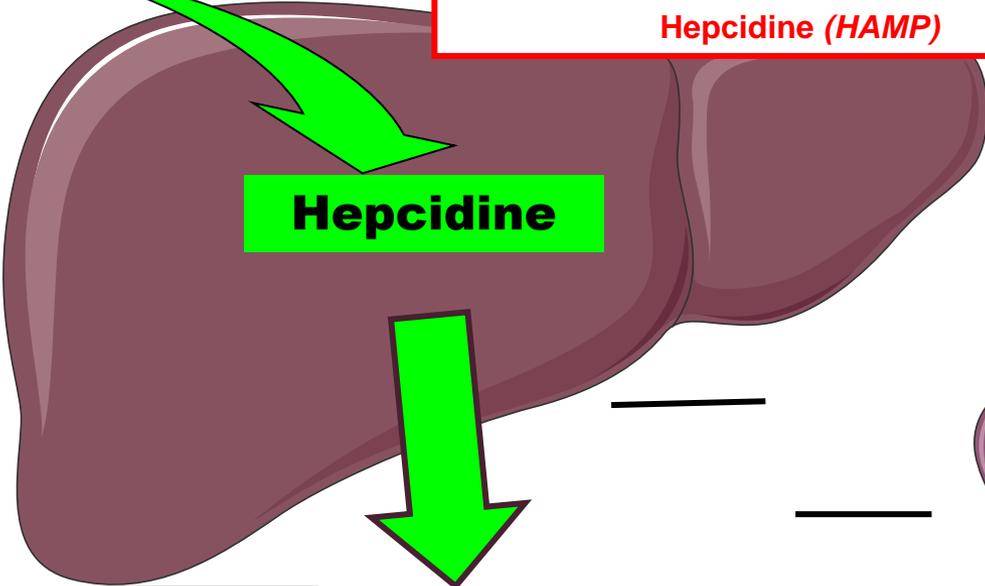
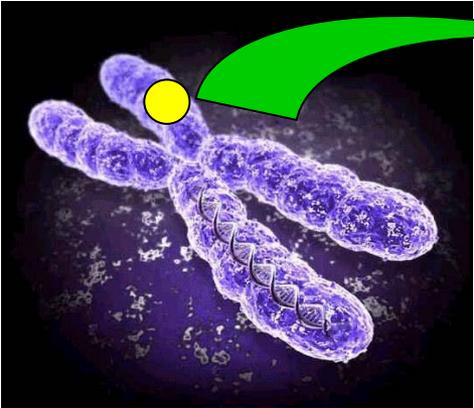
Transferrine



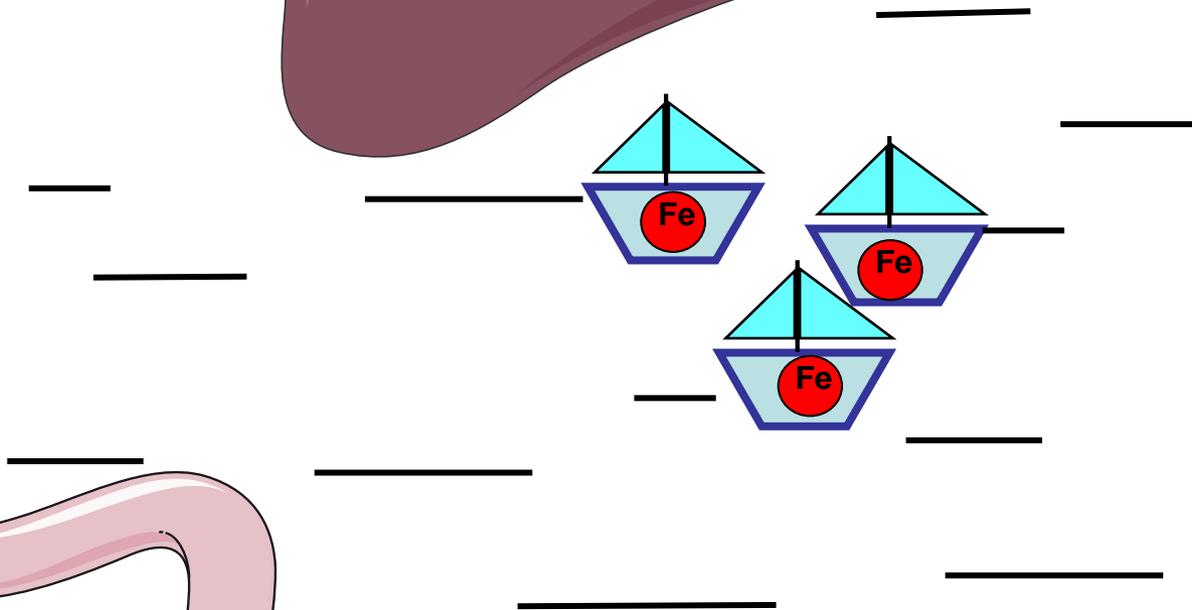
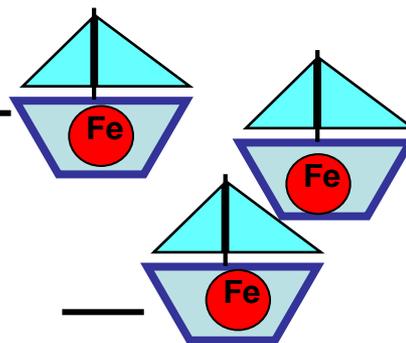
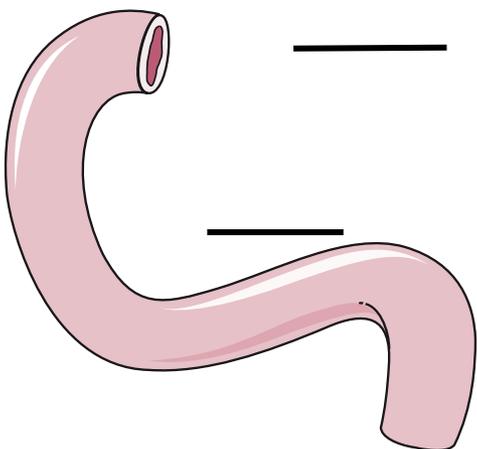
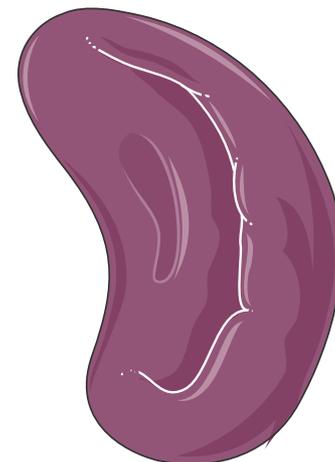
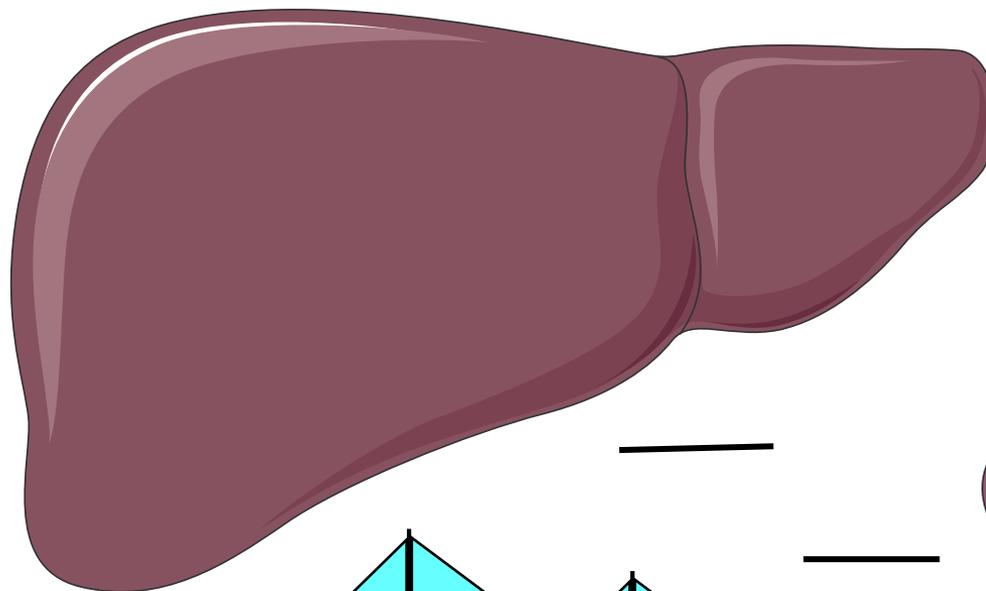
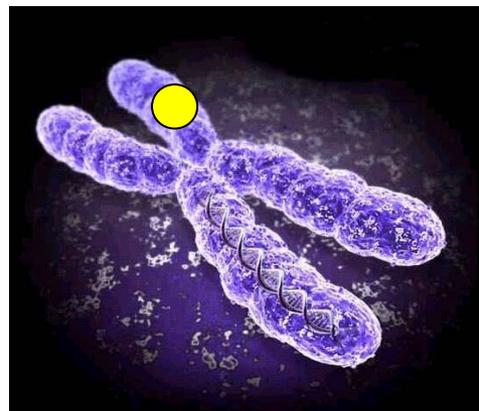
Hémochromatose

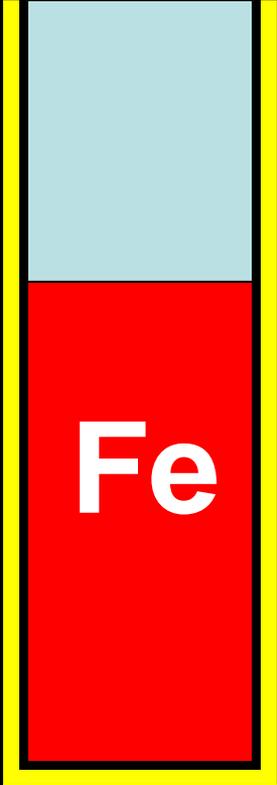
HFE:
C282Y/C282Y

Non HFE:
Hémojuvéline (HJV)
Récepteur Transferrine 2 (TFR2)
Hepcidine (HAMP)



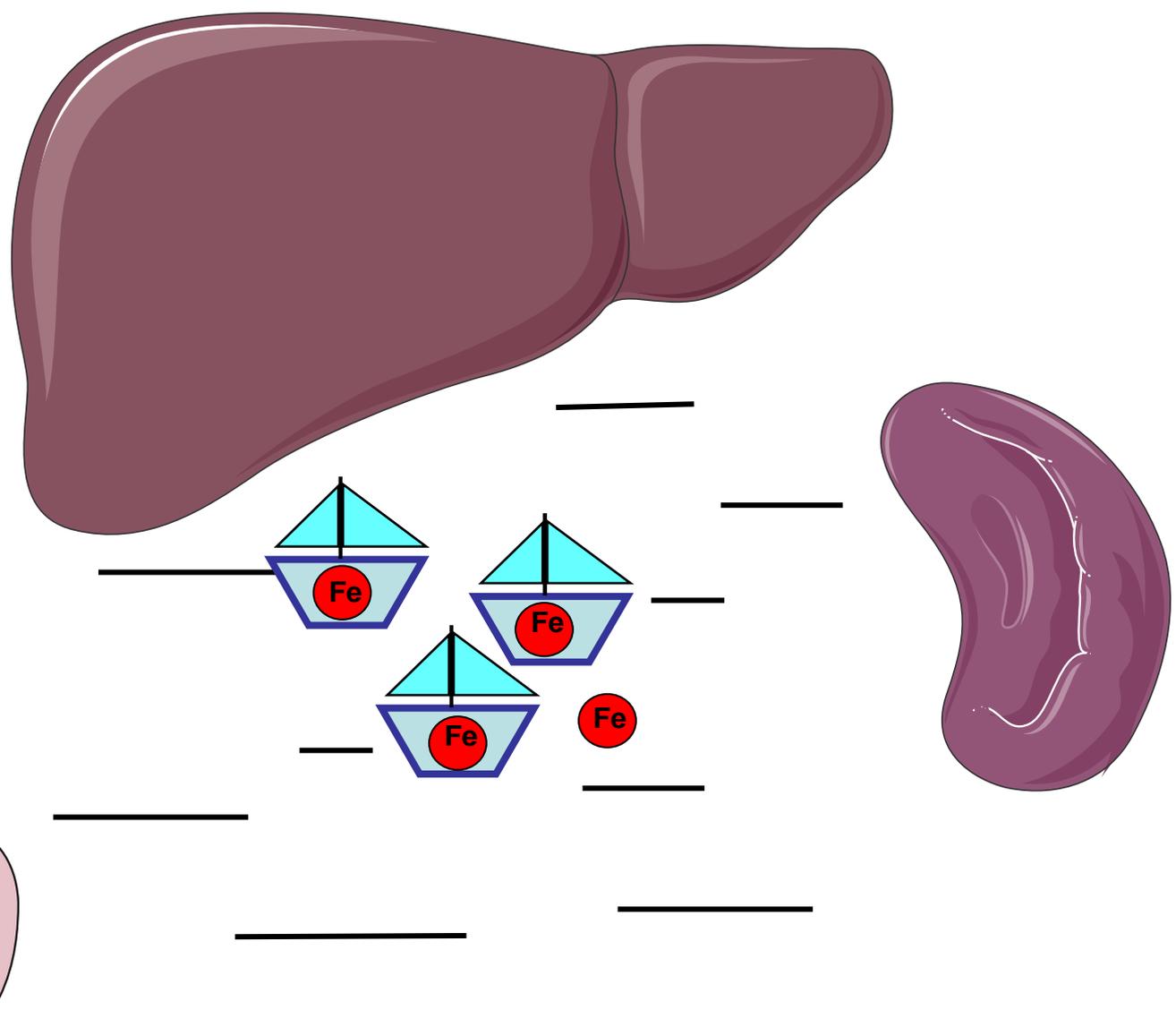
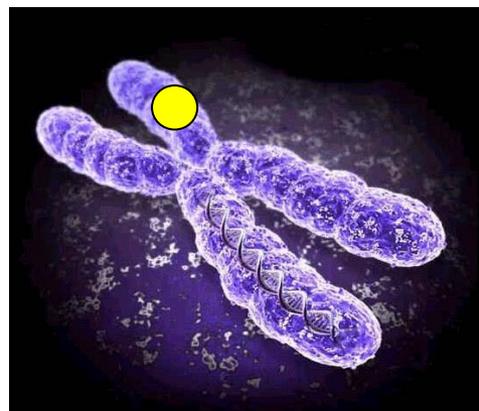
Hémochromatose



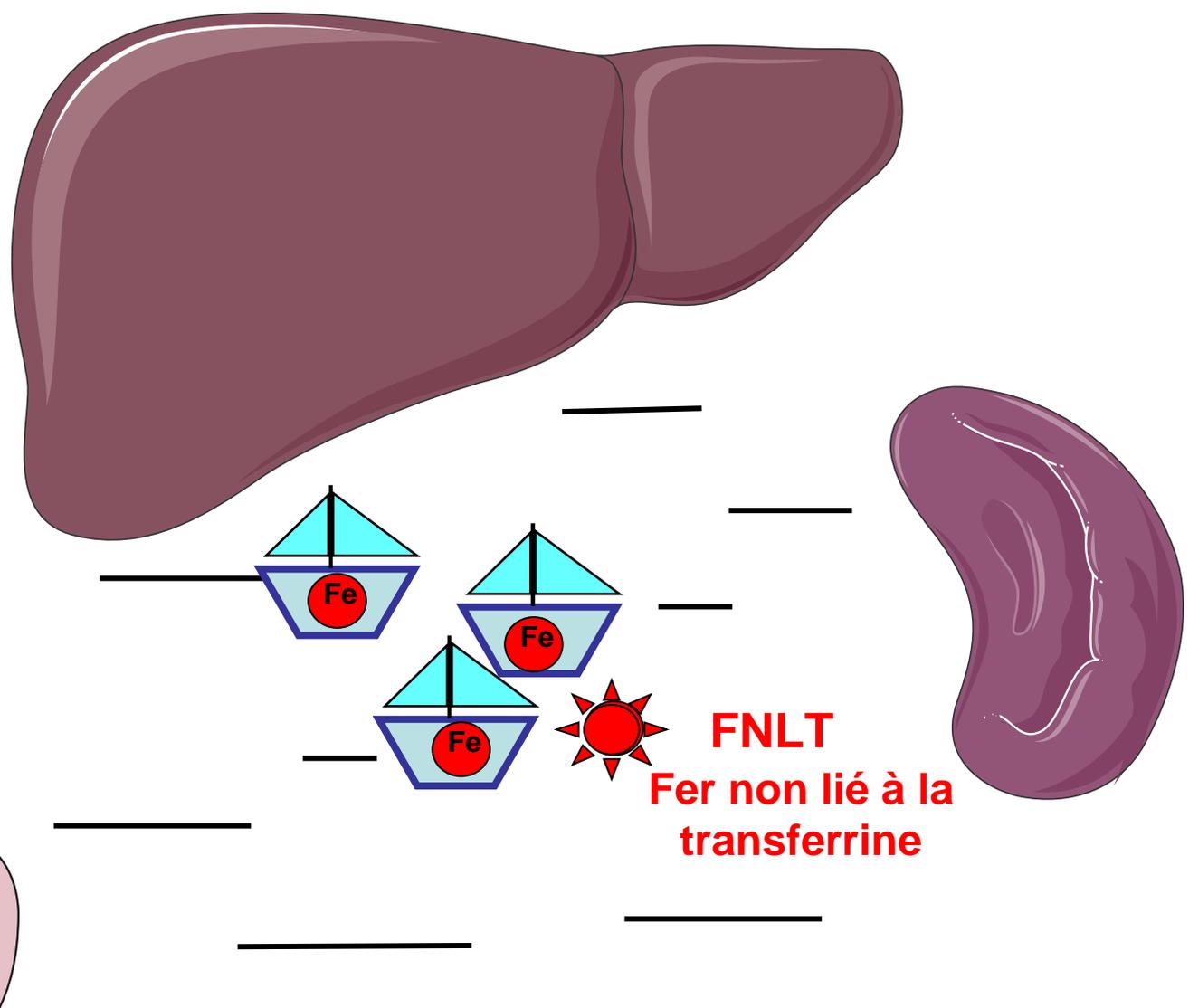
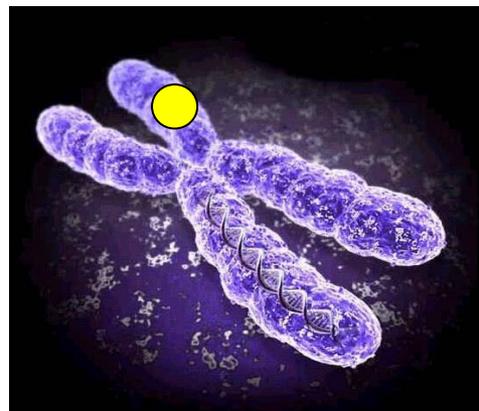


Saturation transferrine > 45%

Hémochromatose

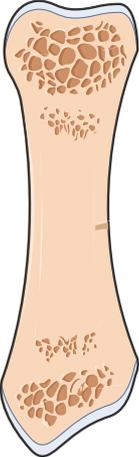
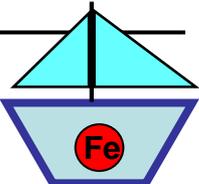


Hémochromatose

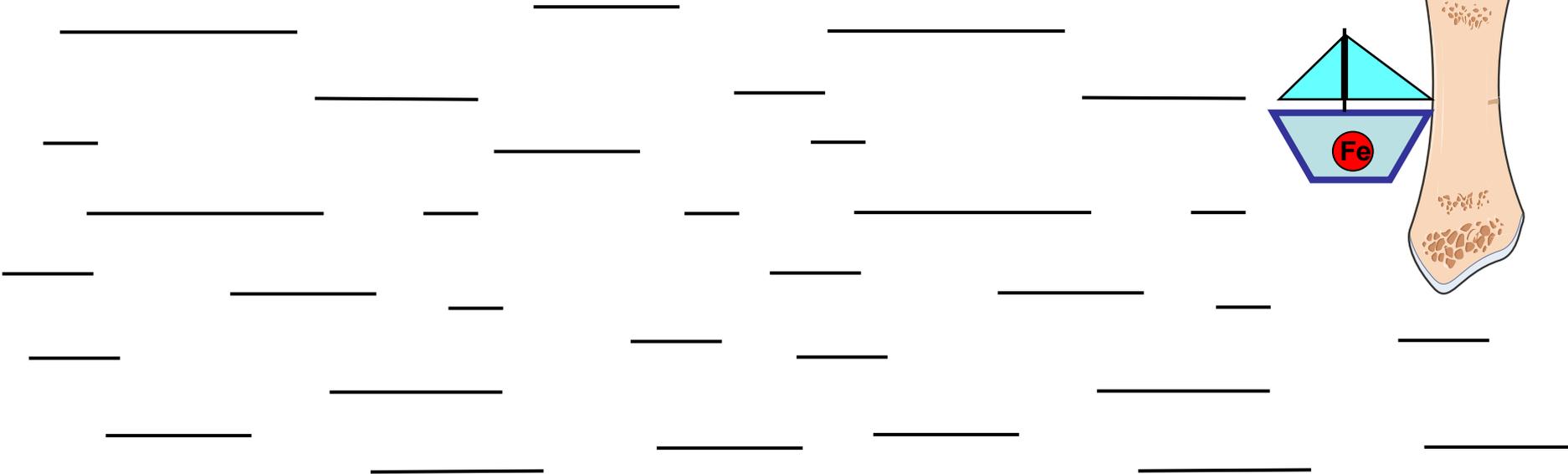


FNLT
Fer non lié à la transferrine

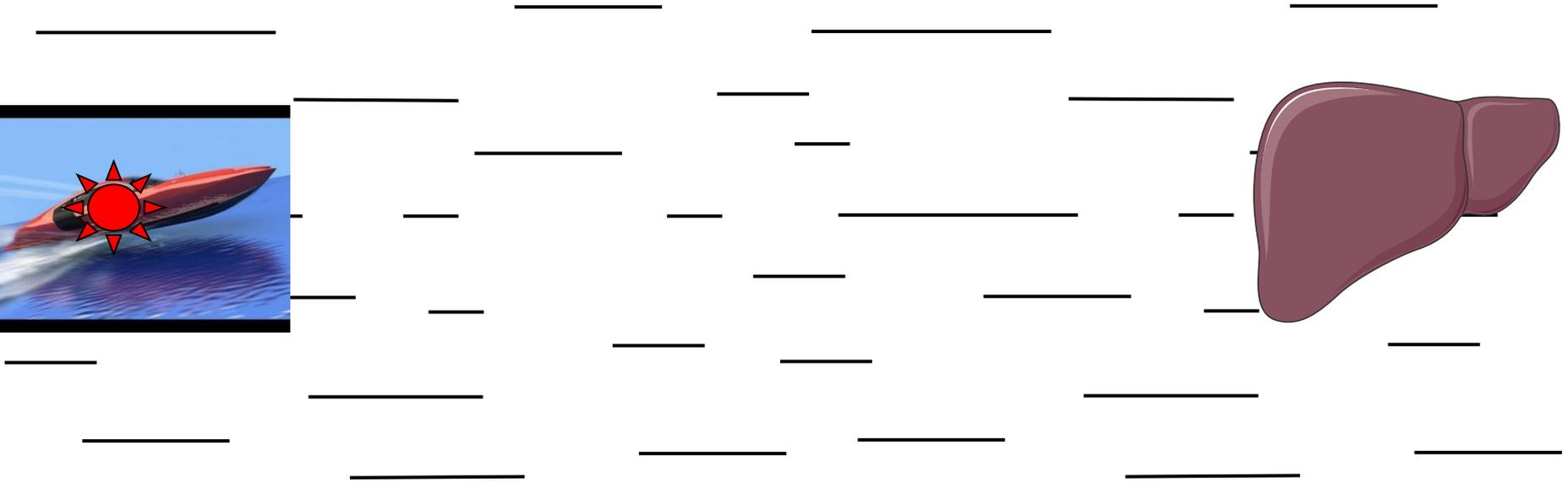
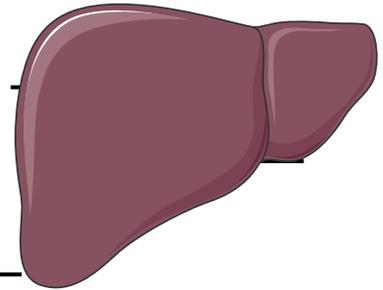
Fer lié à la transferrine



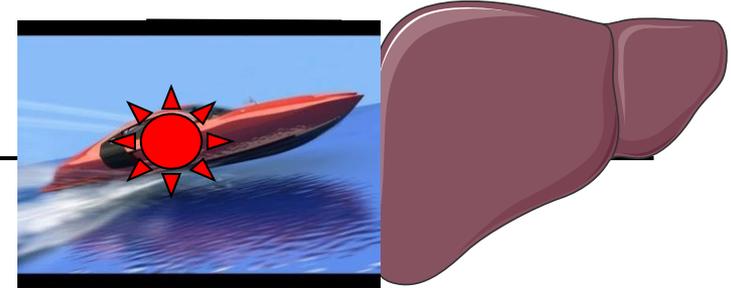
Fer lié à la transferrine



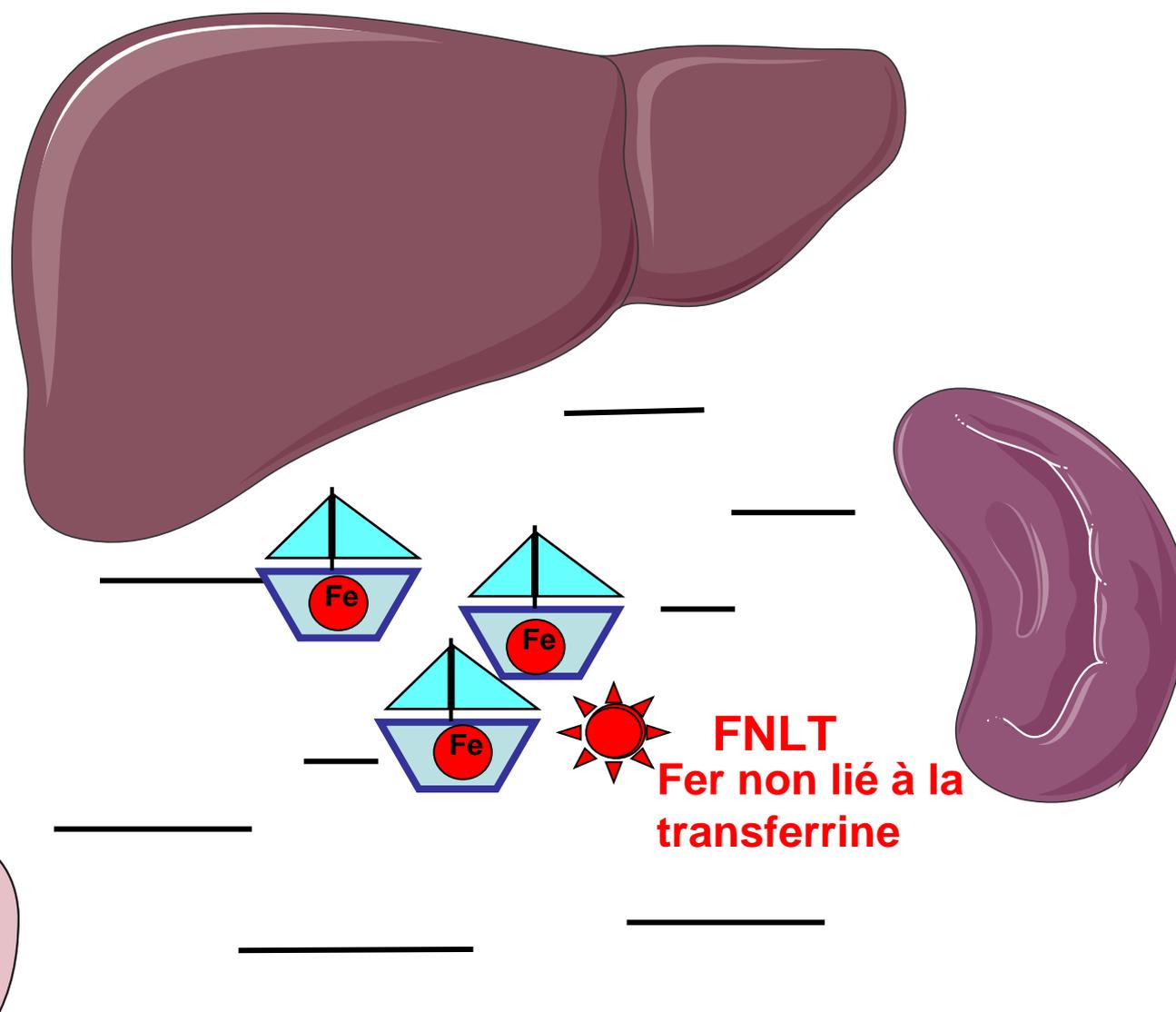
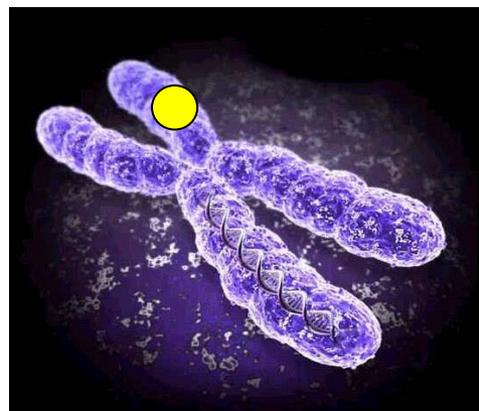
Fer non lié à la transferrine (FNL)



Fer non lié à la transferrine (FNLТ)

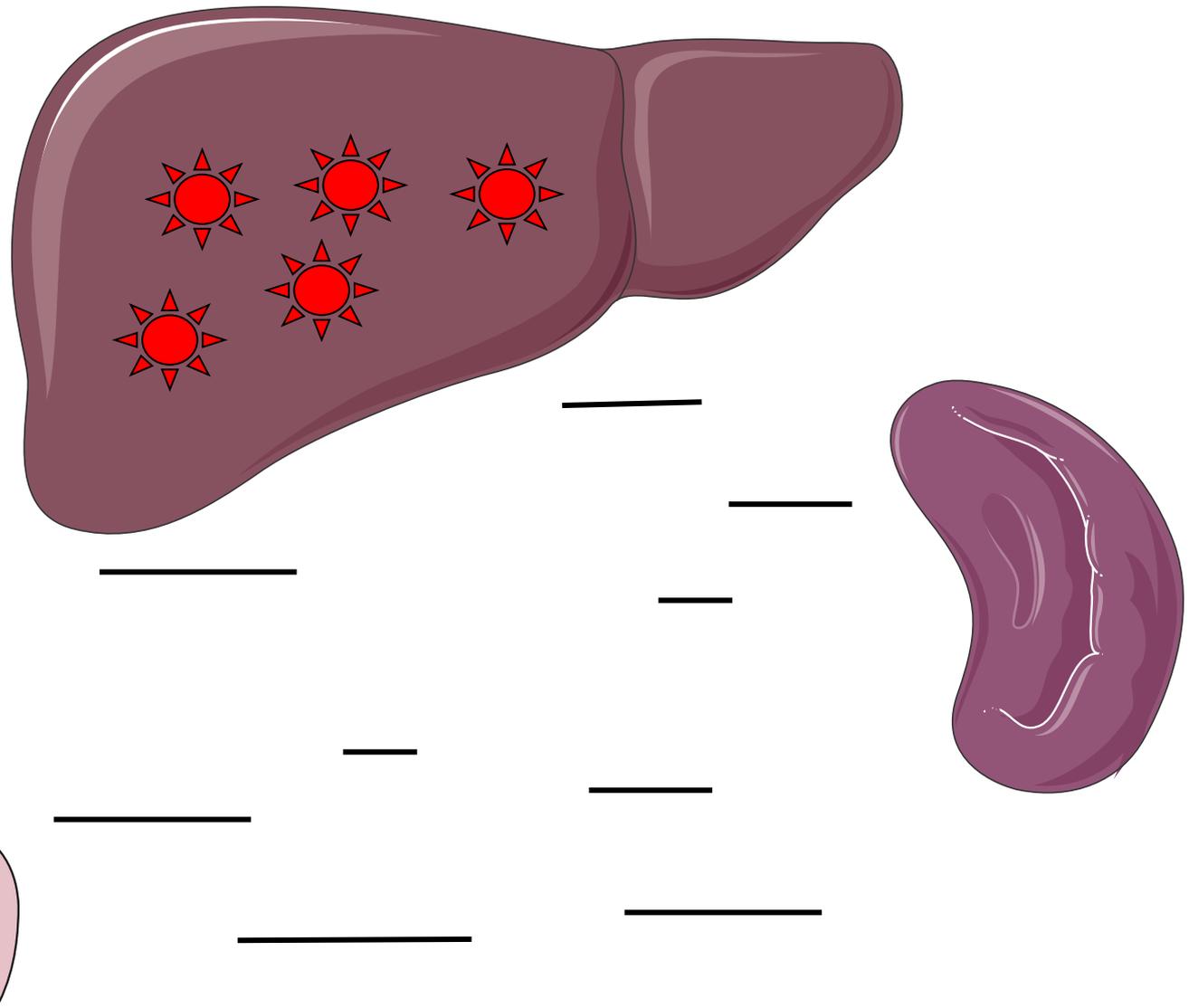
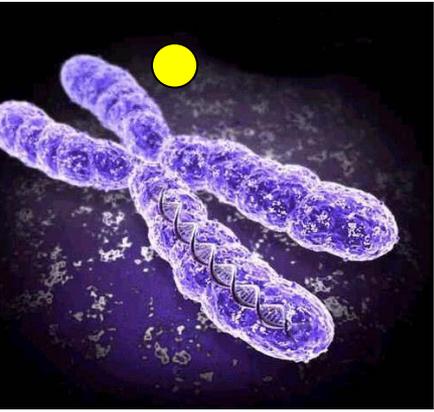


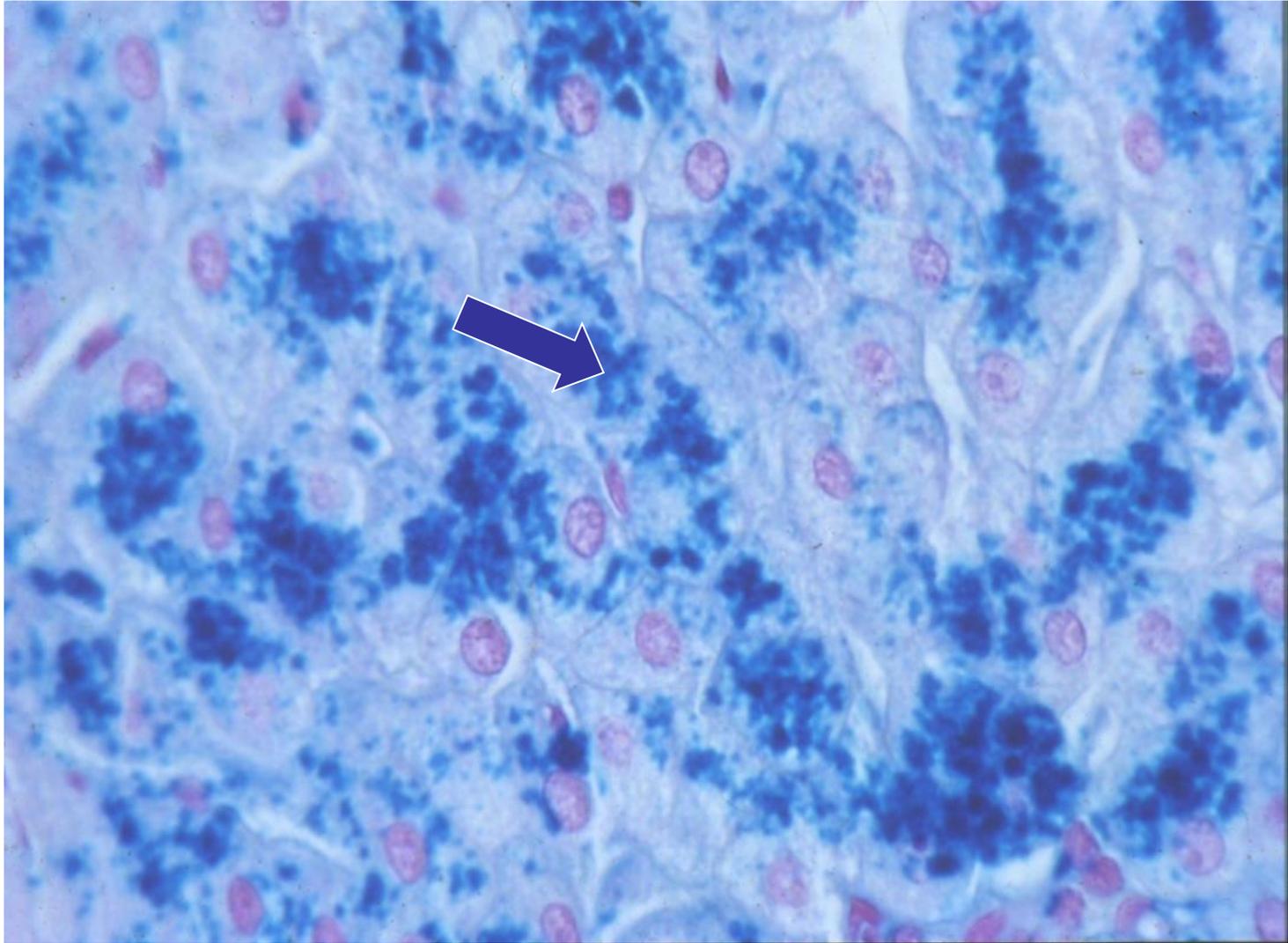
Hémochromatose



FNLT
Fer non lié à la
transferrine

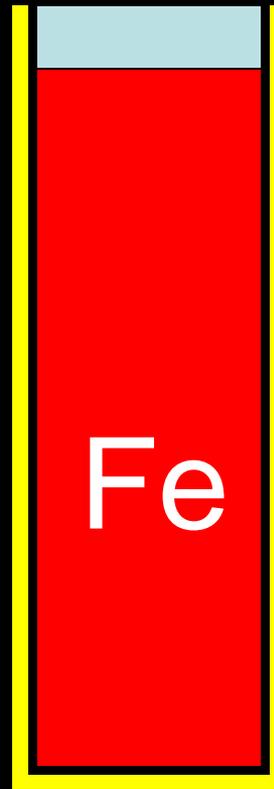
Hémochromatose



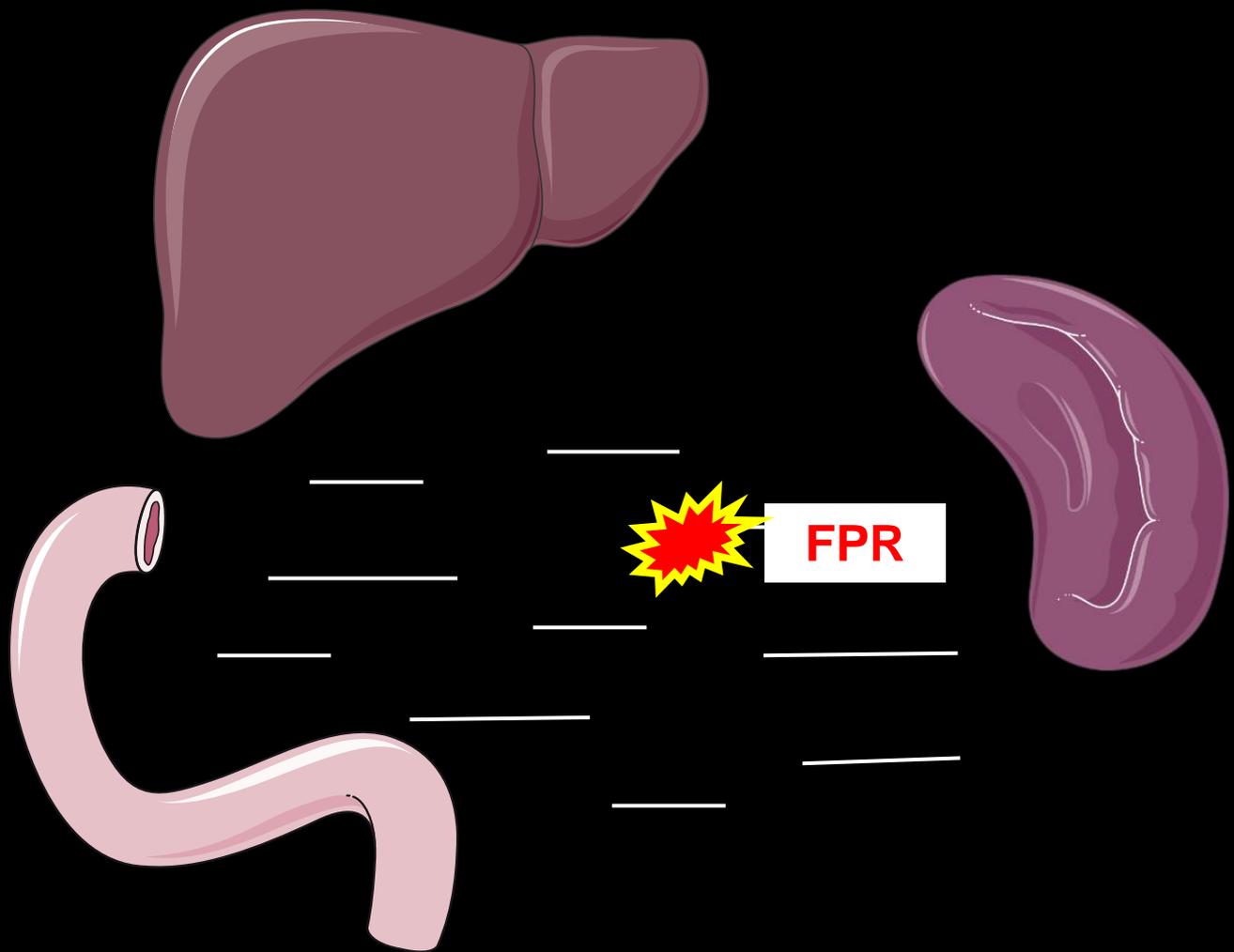


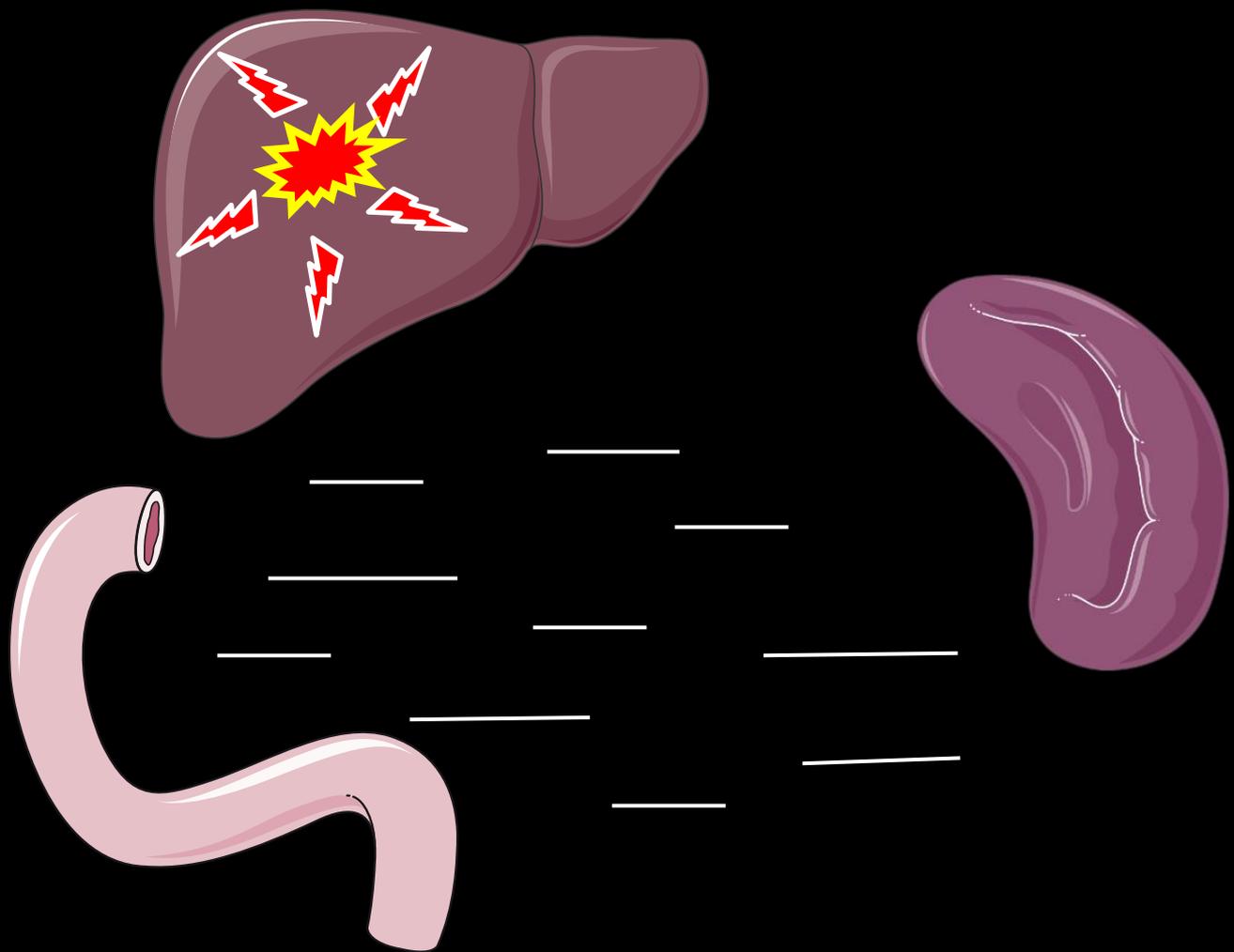
Hépatocytes

FPR (Fer Plasmatique Réactif)



Saturation de la transferrine > 75%

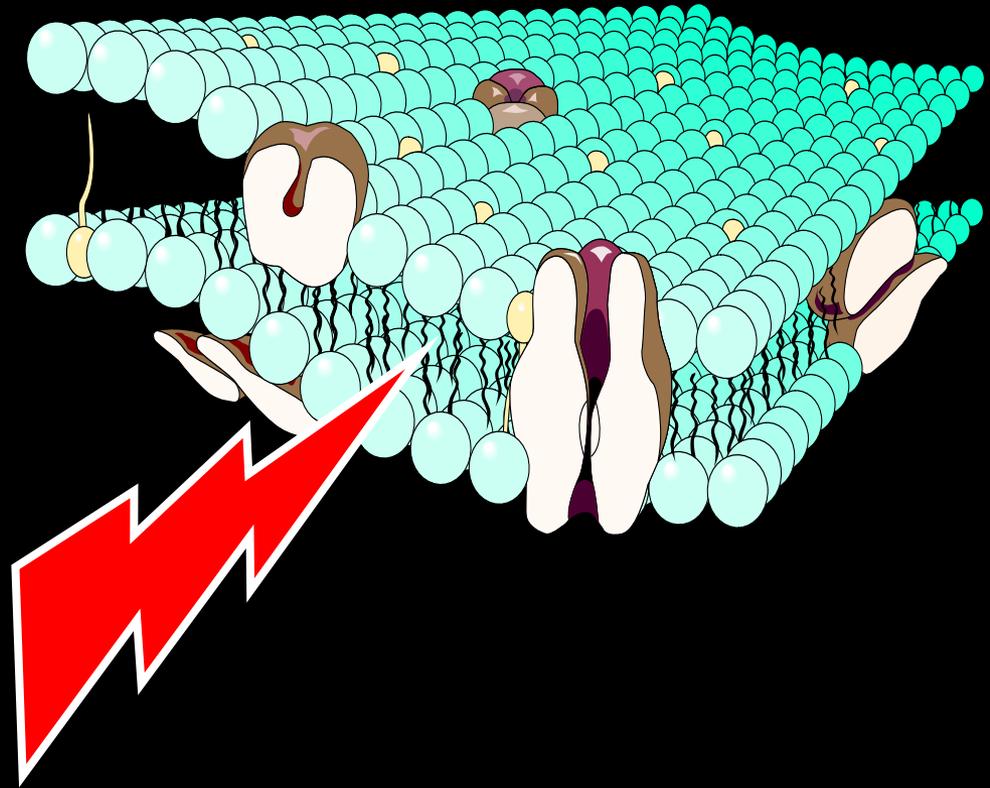


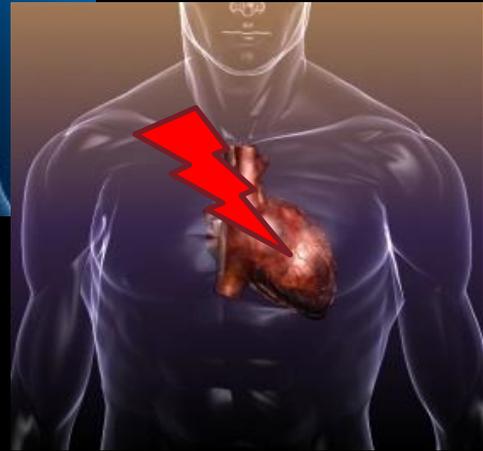
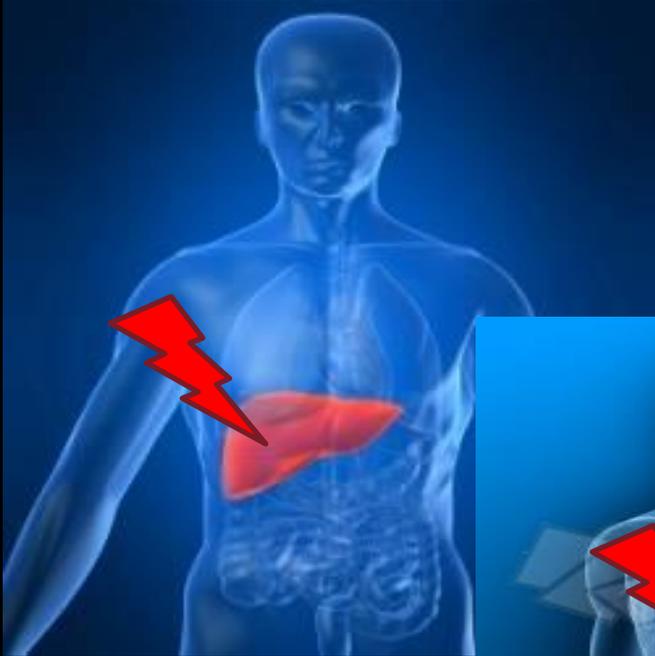


FPR



**Espèces
Radicales
Oxygénées**





Hémochromatose

Surcharge en fer

Génétique

Déficiencia en hepcidina

HFE (C282Y/C282Y)

Non-HFE (rare)

- Hémojuvéline (*HJV*)
- Récepteur Transferrine 2 (*TFR2*)
- Hepcidine (*HAMP*)

↑ Saturation transferrine

Pas de surcharge
splénique

Surcharge
hépatocyttaire

Ce qui n'est pas une hémochromatose

Ce qui n'est pas une hémochromatose

Surcharge en fer

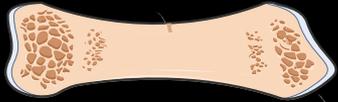
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graph TD; A[Surcharge en fer] --> B[Génétique]
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~~Génétique~~

Surcharge en fer acquise

- Transfusions
- Supplémentation i.v. en fer
- Dysérythropoïèse

Surcharge en fer



Erythroferrone

Déficiéne en hepcidine

Surcharge en fer acquise

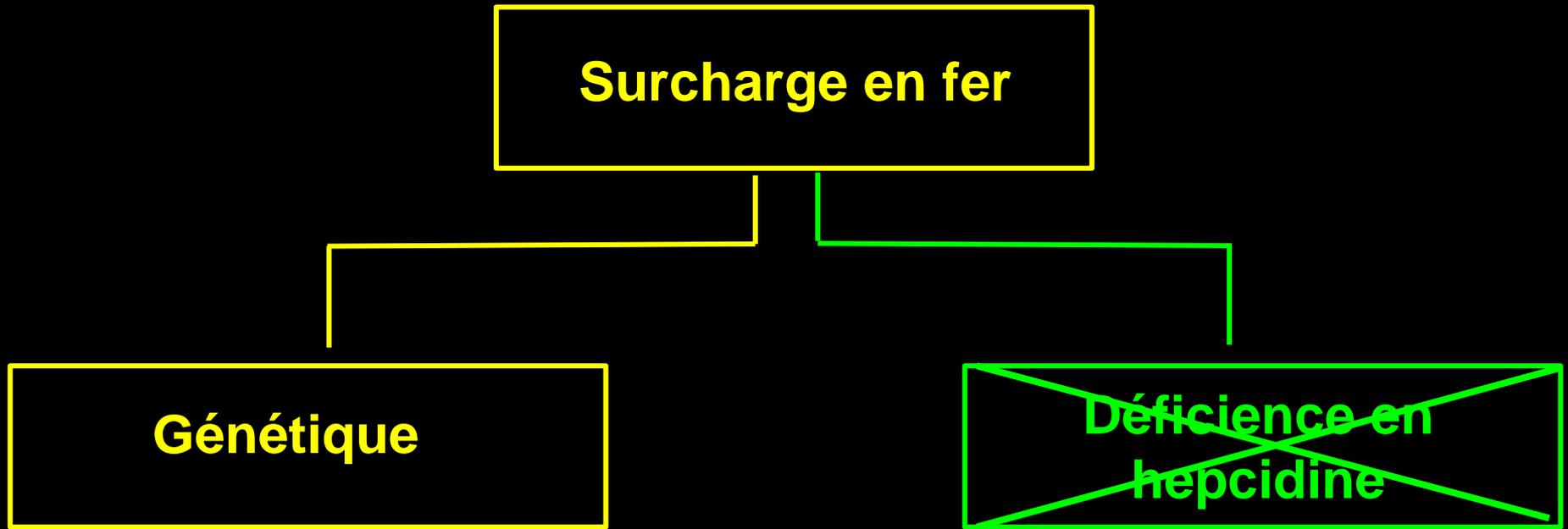
- Transfusions
- Supplémentation i.v. en fer
- Dysérythropoïèse

↑ Sat. Transferrine

Pas de surcharge splénique

Surcharge hépatocytaire

Ce qui n'est pas une hémochromatose



Maladie de la ferroportine

Acéruloplasminémie héréditaire

Hémochromatose

Définition et physiopathologie

Diagnostic

Hémochromatose liée à HFE

Hémochromatose non liée à HFE

Autres surcharges génétiques

Traitement

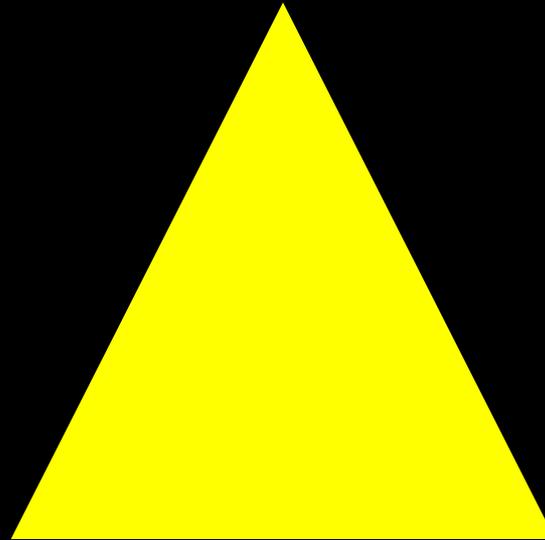
Prévention

Hémochromatose liée à *HFE*

Diagnostic non invasif



Expression clinique



Imagerie

Biologie



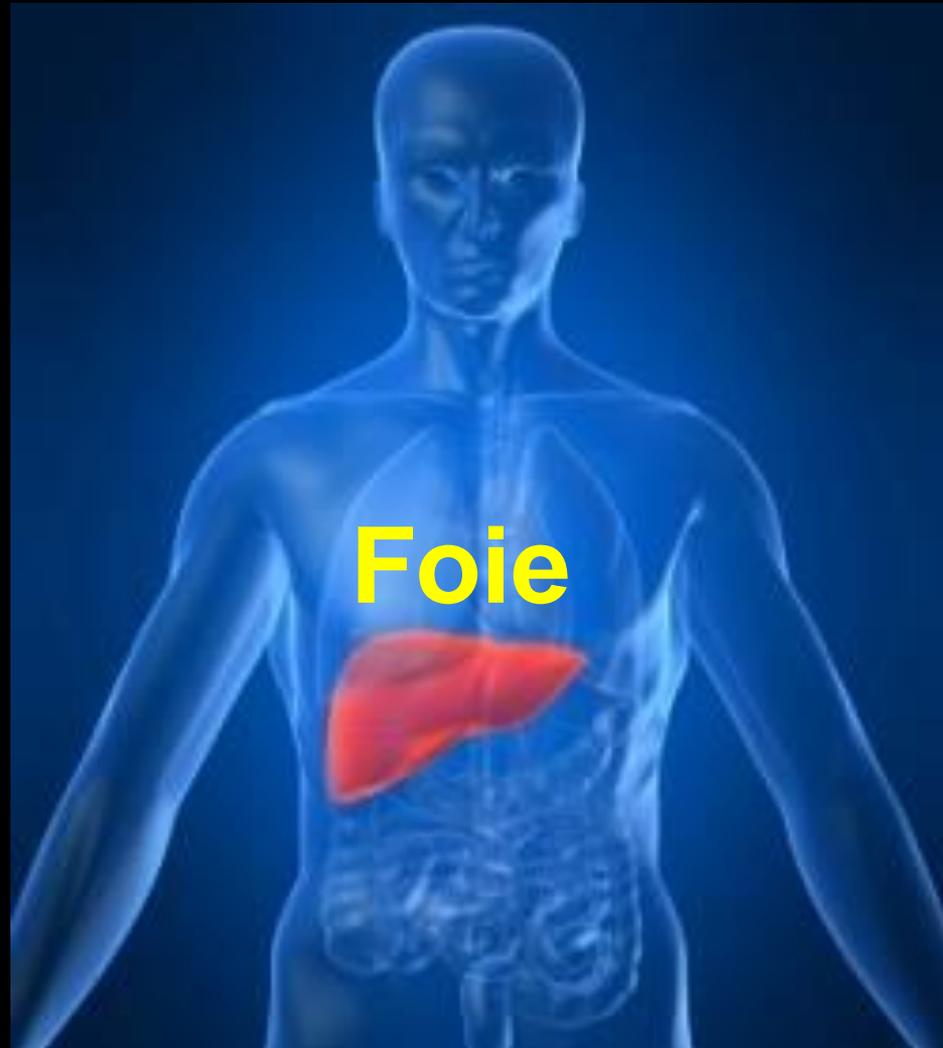






Ostéoporose





cirrhose





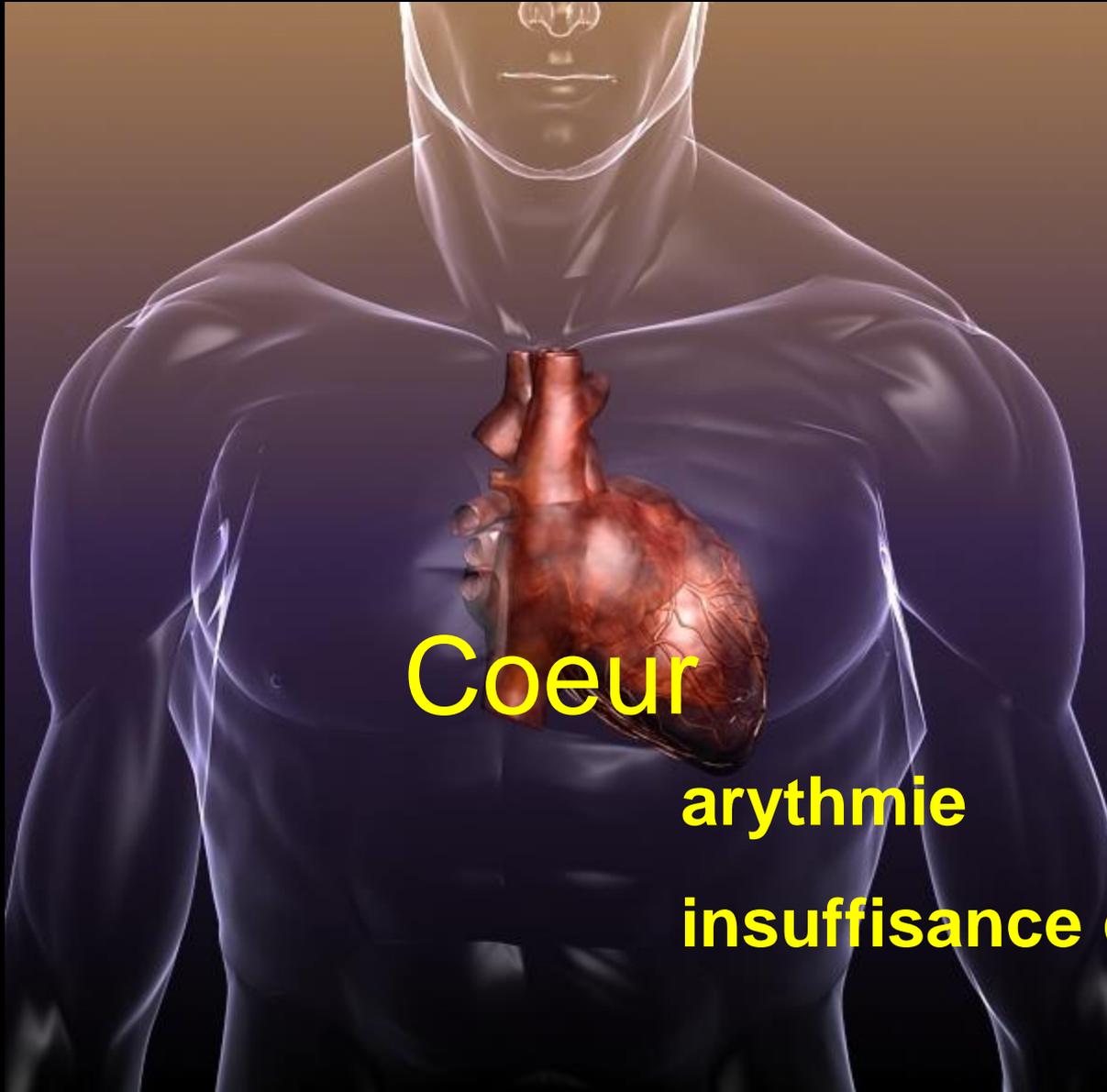
Cancer





Pancréas

diabète

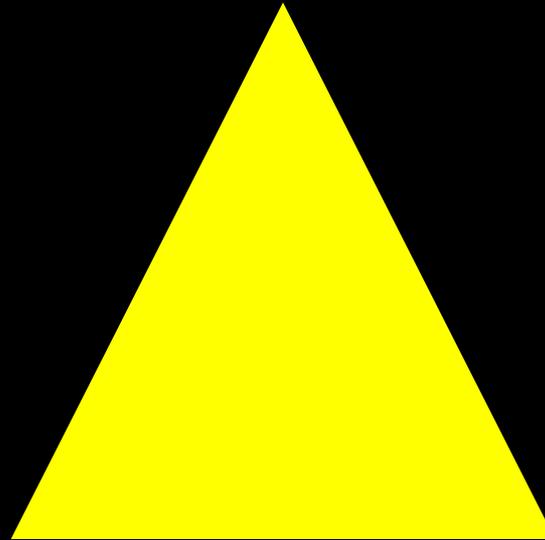


Coeur

arythmie

insuffisance cardiaque

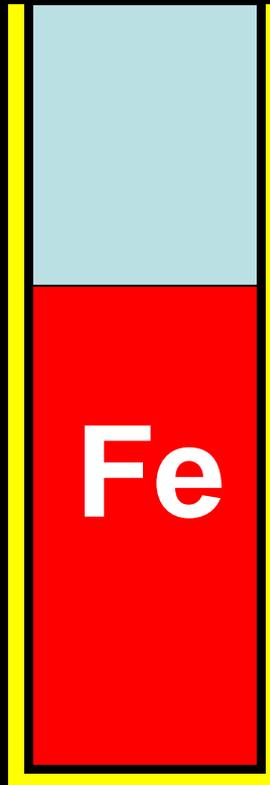
Expression clinique



Imagerie

Biologie

- **Saturation transferrine**
- **Ferritine**
- **Test génétique**



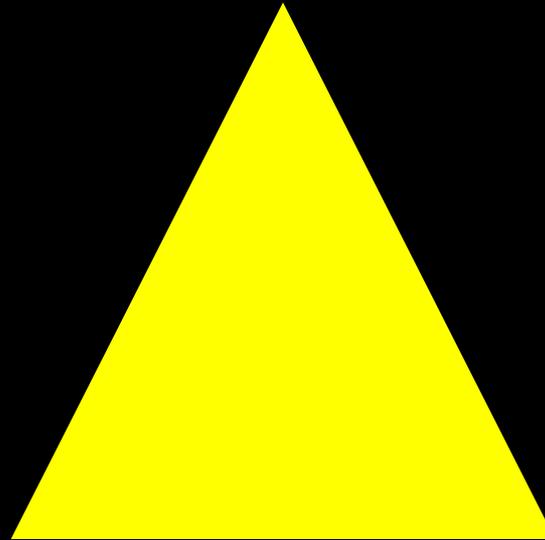
Saturation transferrine > 45%

**> 60 % (H)
> 50 % (F)**

À doser 2 fois !

**Une saturation de transferrine normale
exclut une hémochromatose**

Expression clinique



Imagerie

Biologie

- **Saturation transferrine**
- **Ferritine**
- **Test génétique**

> 300 $\mu\text{g/L}$ (H)

> 200 $\mu\text{g/L}$ (F)

↑ Ferritine

Syndrome métabolique



Inflammation



Alcoolisme



> 300 µg/L (H)

> 200 µg/L (F)



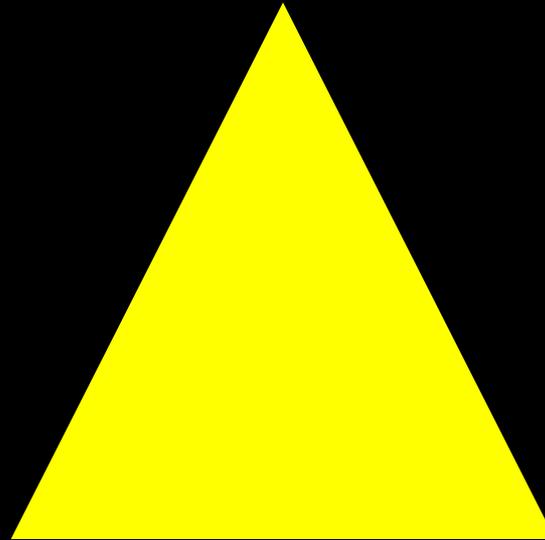
↑ Ferritine

Syndrome métabolique

Inflammation

Alcoolisme

Expression clinique



Imagerie

Biologie

- **Saturation transferrine**
- **Ferritine**
- **Test génétique**

C282Y/C282Y

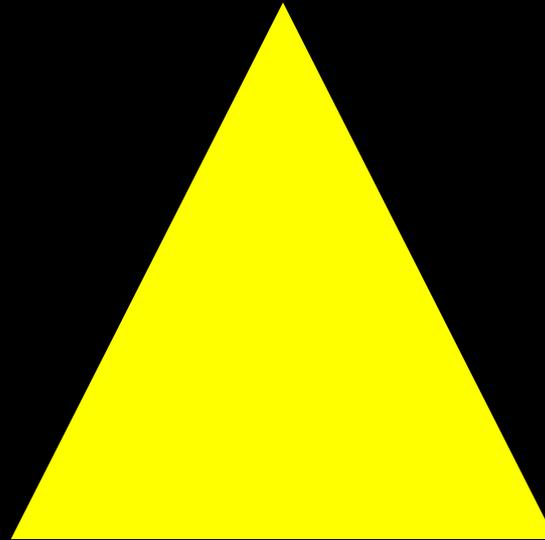
~~**H63D**~~

~~**H63D/H63D**~~

~~**S65C**~~

C282Y/H63D...

Expression clinique



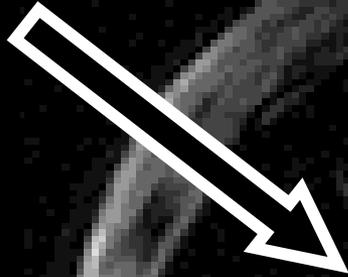
Imagerie

Biologie

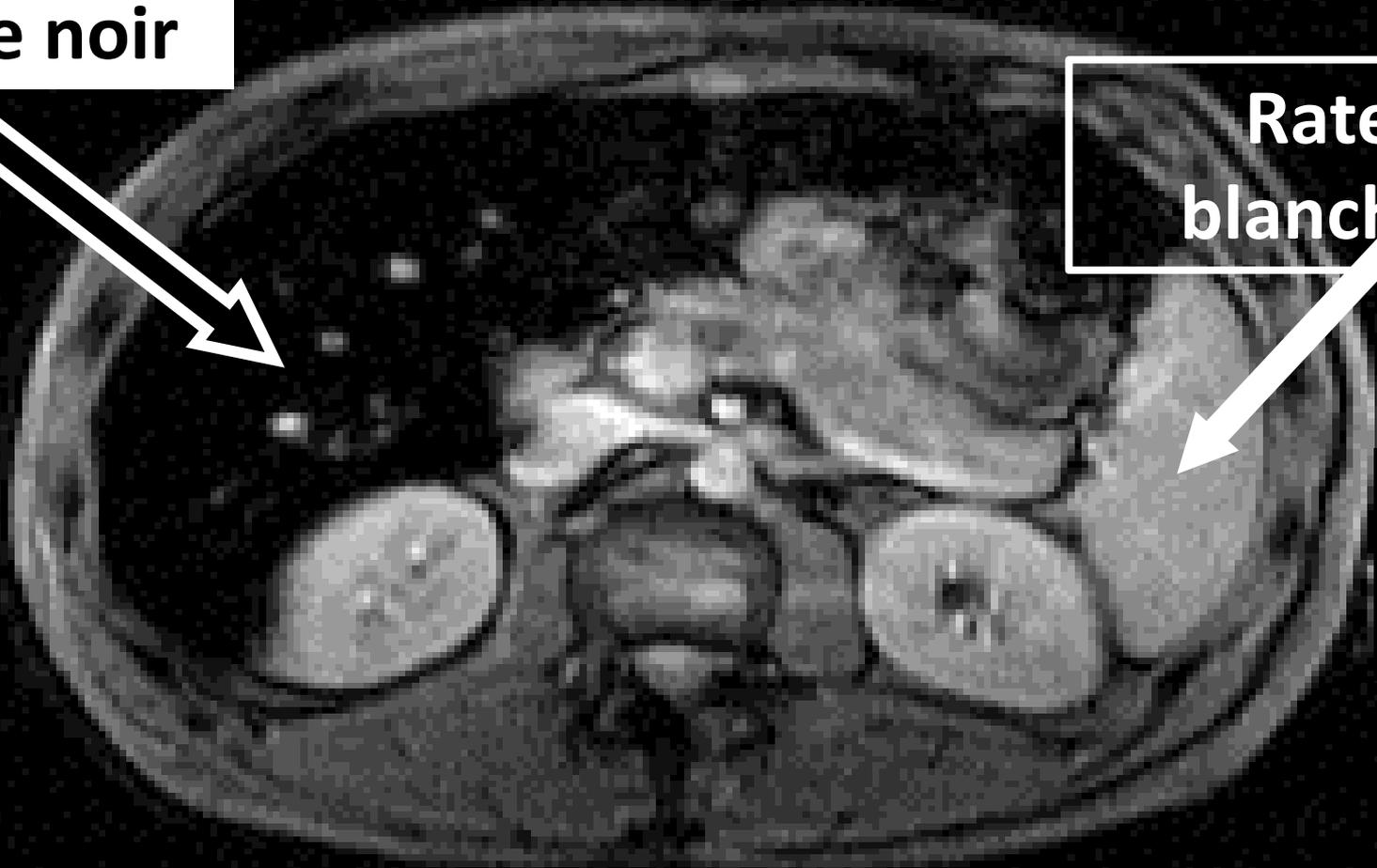
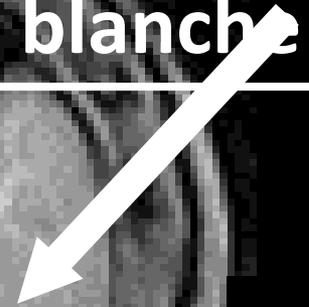
- **Saturation transferrine**
- **Ferritine**
- **Test génétique**

IRM

Foie noir



Rate
blanche



Hémochromatose

Définition et physiopathologie

Diagnostic

Hémochromatose liée à HFE

Hémochromatose non liée à HFE

Autres surcharges génétiques

Traitement

Prévention

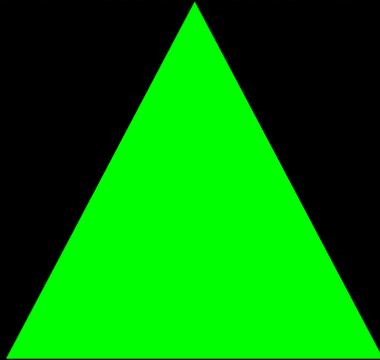
Hémochromatose non liée à *HFE* (= non *C282Y/C282Y*)

(hémoujuvéline, TFR2, hepcidine...)

Similarités

« Syndrome d'hepcidino-déficience »

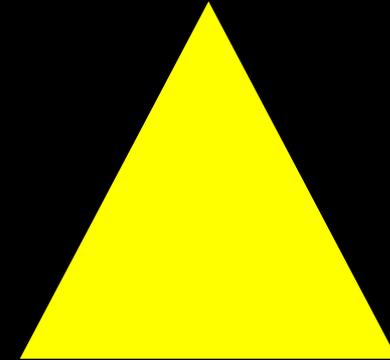
↑ Saturation transferrine



Pas de surcharge
splénique

Surcharge
hépatique

Expression clinique



Imagerie

Biologie

Différences...

Hémochromatose non liée à *HFE* (= non *C282Y/C282Y*)

(hémoujuvéline, TFR2, hepcidine...)

Différences

- Rare ...mais multi-ethnique
- < 30 ans
- Plus sévère
- Centre expert

Hémochromatose

Définition et physiopathologie

Diagnostic

Hémochromatose liée à HFE

Hémochromatose non liée à HFE

Autres surcharges génétiques

Traitement

Prévention

Surcharges génétiques en fer non hémochromatosiques

Surcharge en fer

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graph TD; A[Surcharge en fer] --> B[Génétique]; A --> C[Déficiency en hepcidine];
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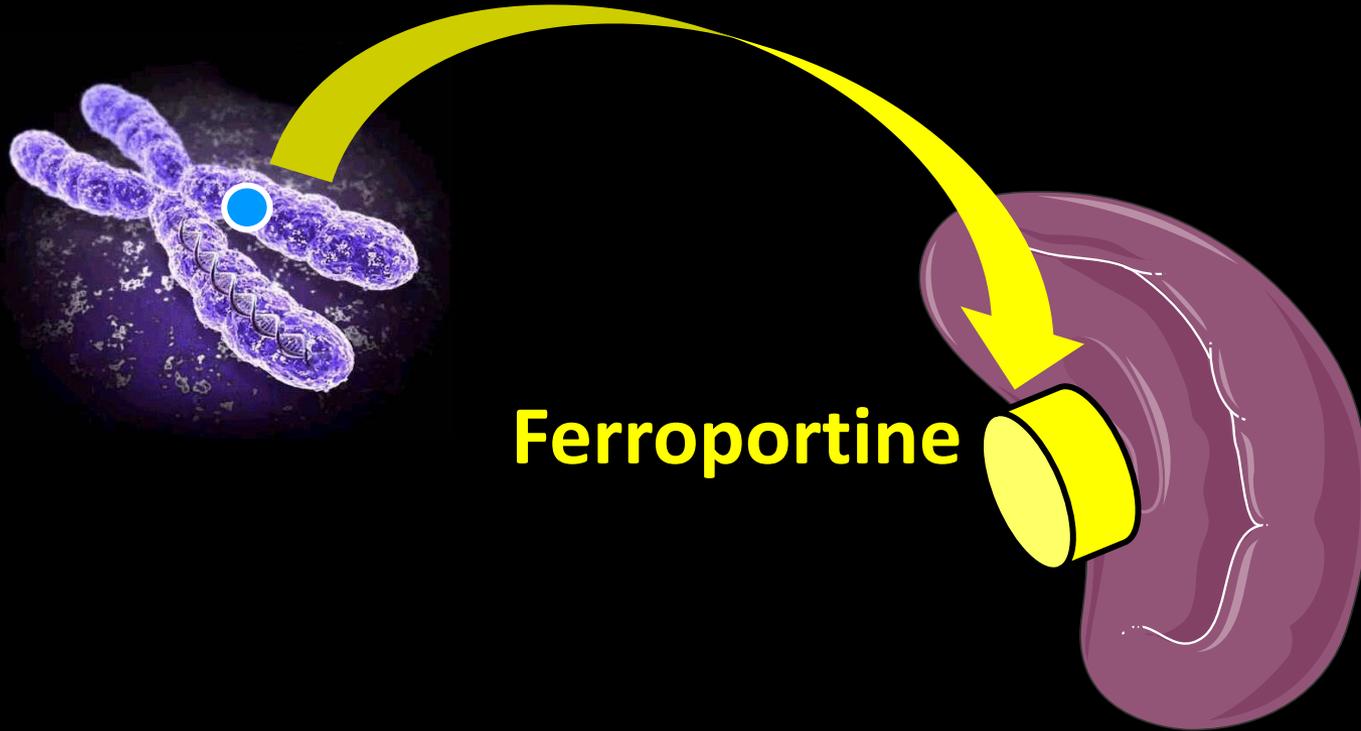
Génétique

~~Déficiency en
hepcidine~~

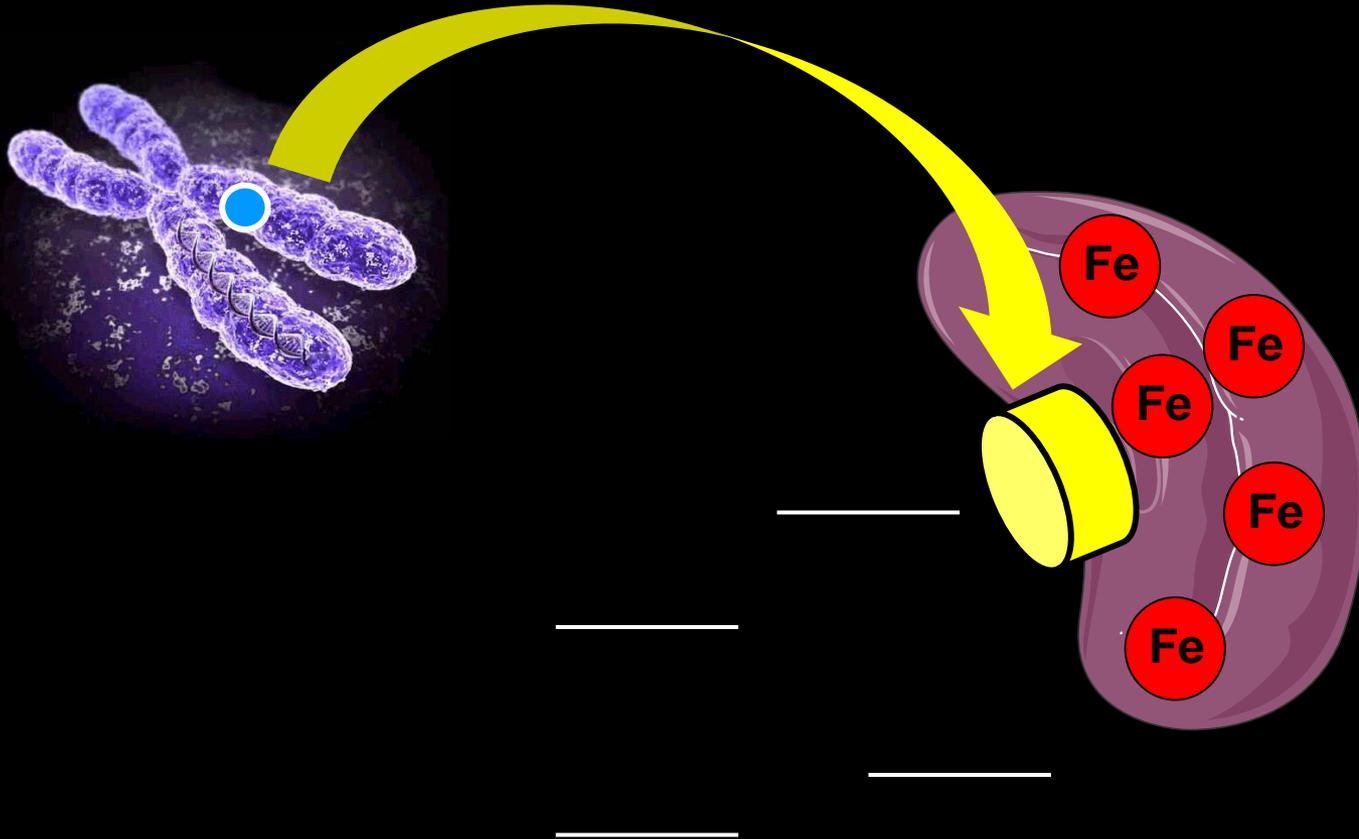
Maladie de la ferroportine

Acéruloplasminémie héréditaire

Maladie de la ferroportine



Maladie de la ferroportine



Normalité de la saturation
de la transferrine

Hyperferritinémie
très marquée

Acéruloplasminémie héréditaire

- Anémie
 - Saturation de la transferrine basse
 - Hyperferritinémie



- Céruloplasmine : 0

**Une saturation de transferrine normale
exclut une hémochromatose**

**mais n'exclut pas certaines formes, très rares, de
surcharges génétiques en fer**

Hémochromatose

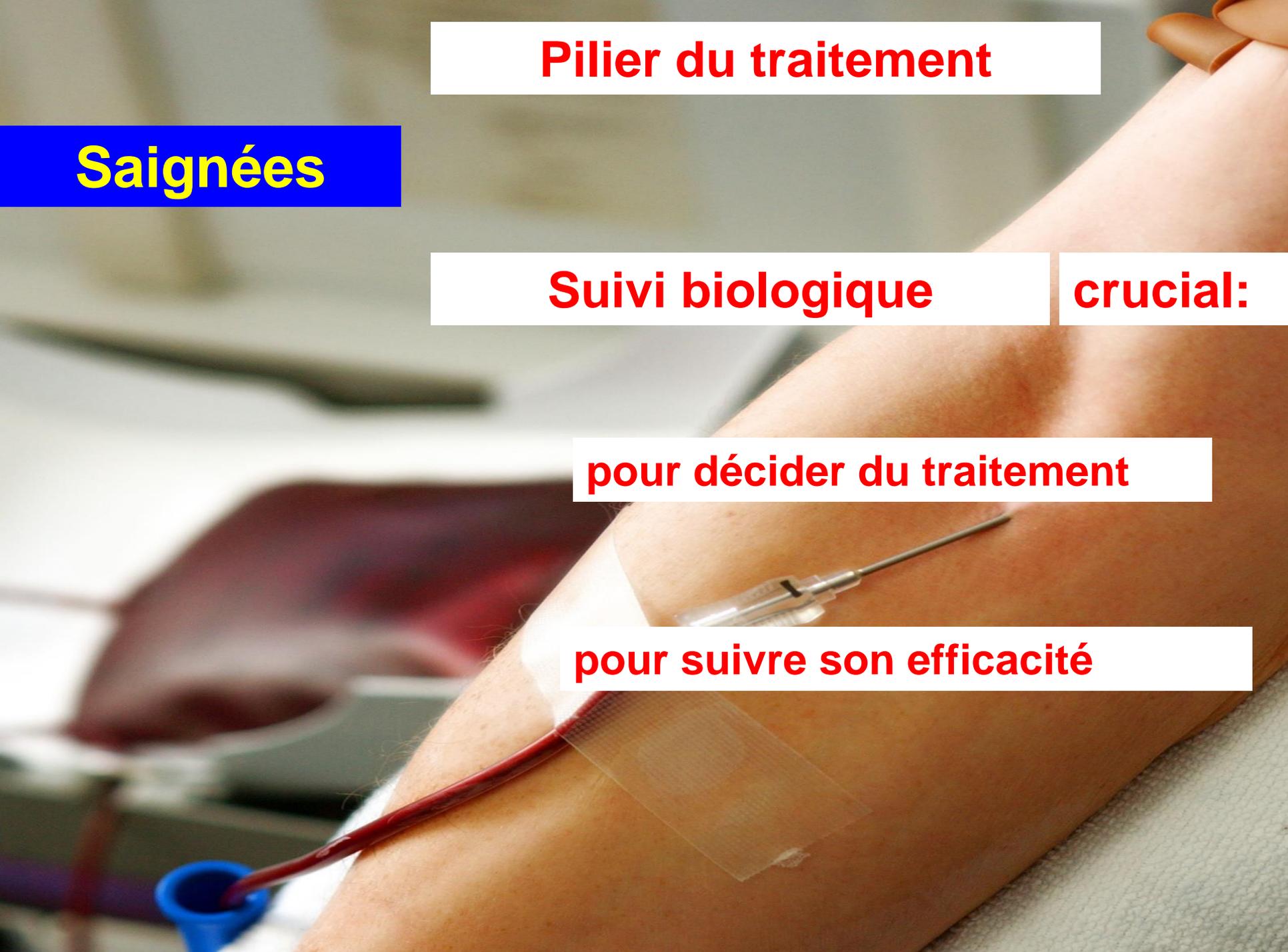
Définition et physiopathologie

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Traitement

Aujourd'hui
Demain

Prévention



Pilier du traitement

Saignées

Suivi biologique

crucial:

pour décider du traitement

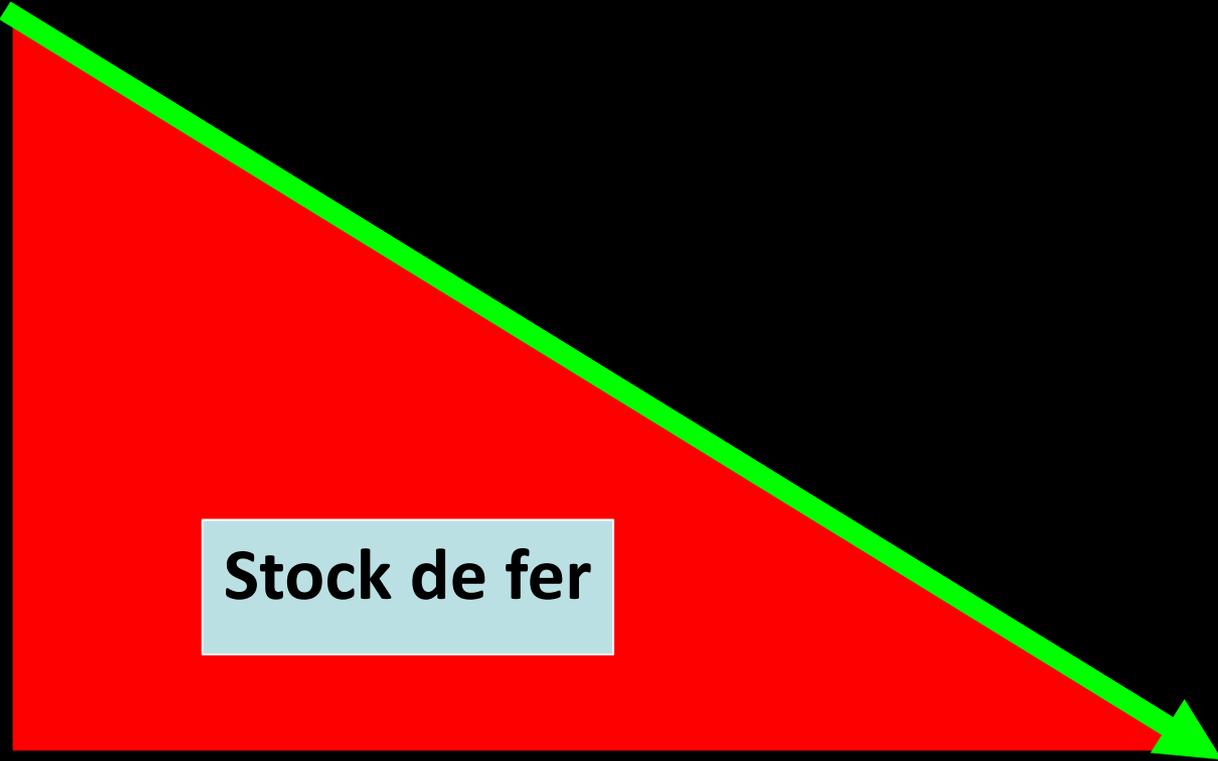
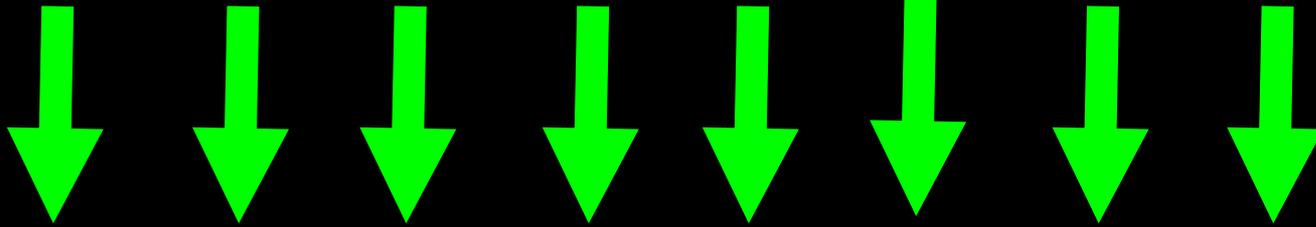
pour suivre son efficacité

Ferritine \geq 300 $\mu\text{g/L}$ (H)

Ferritine \geq 200 $\mu\text{g/L}$ (F)

Phase d'induction

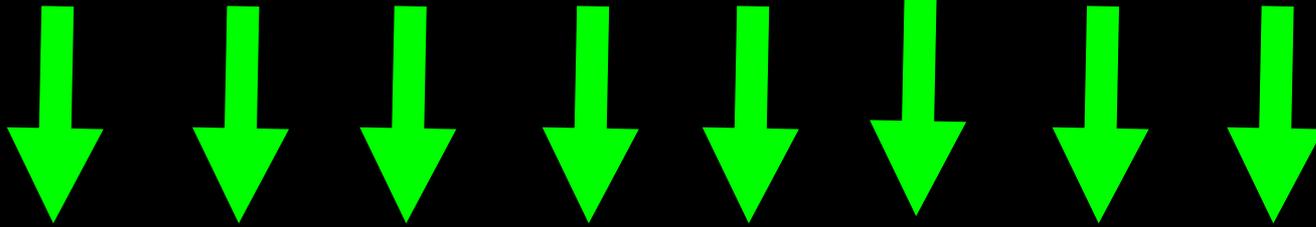
Saignées



Stock de fer

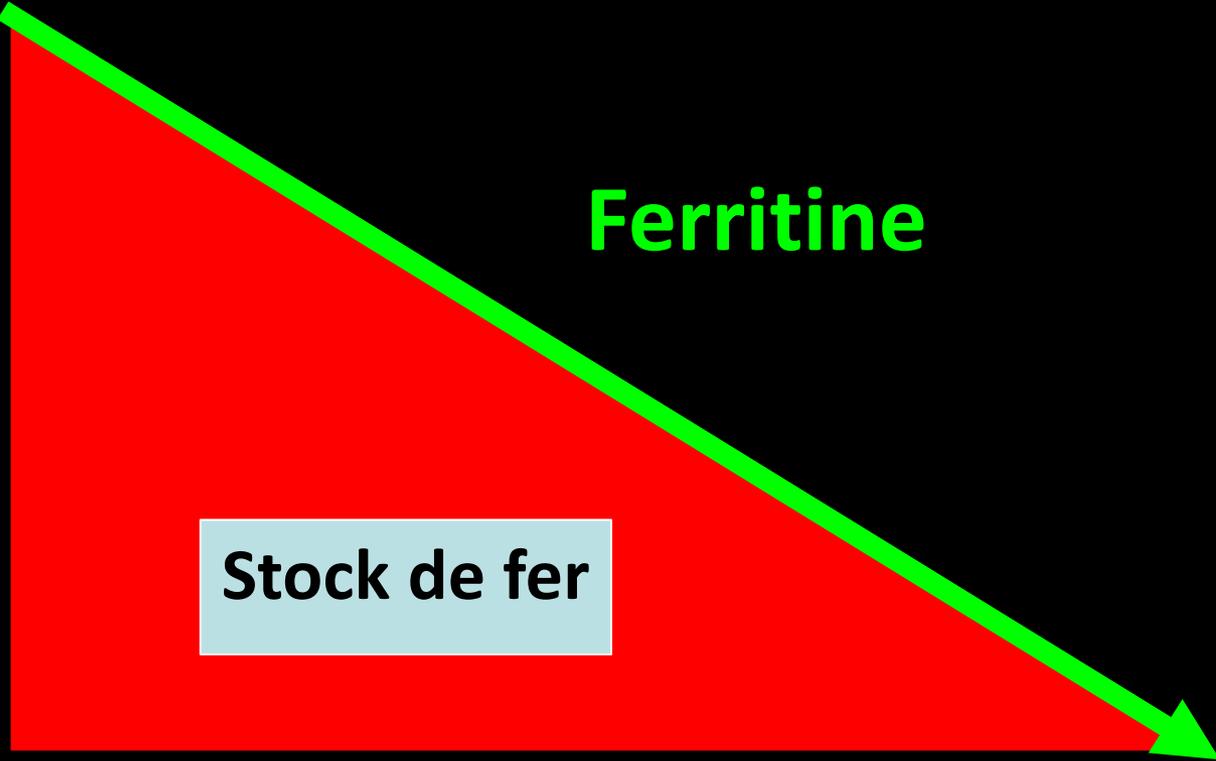
Ferritine = efficacité

Saignées

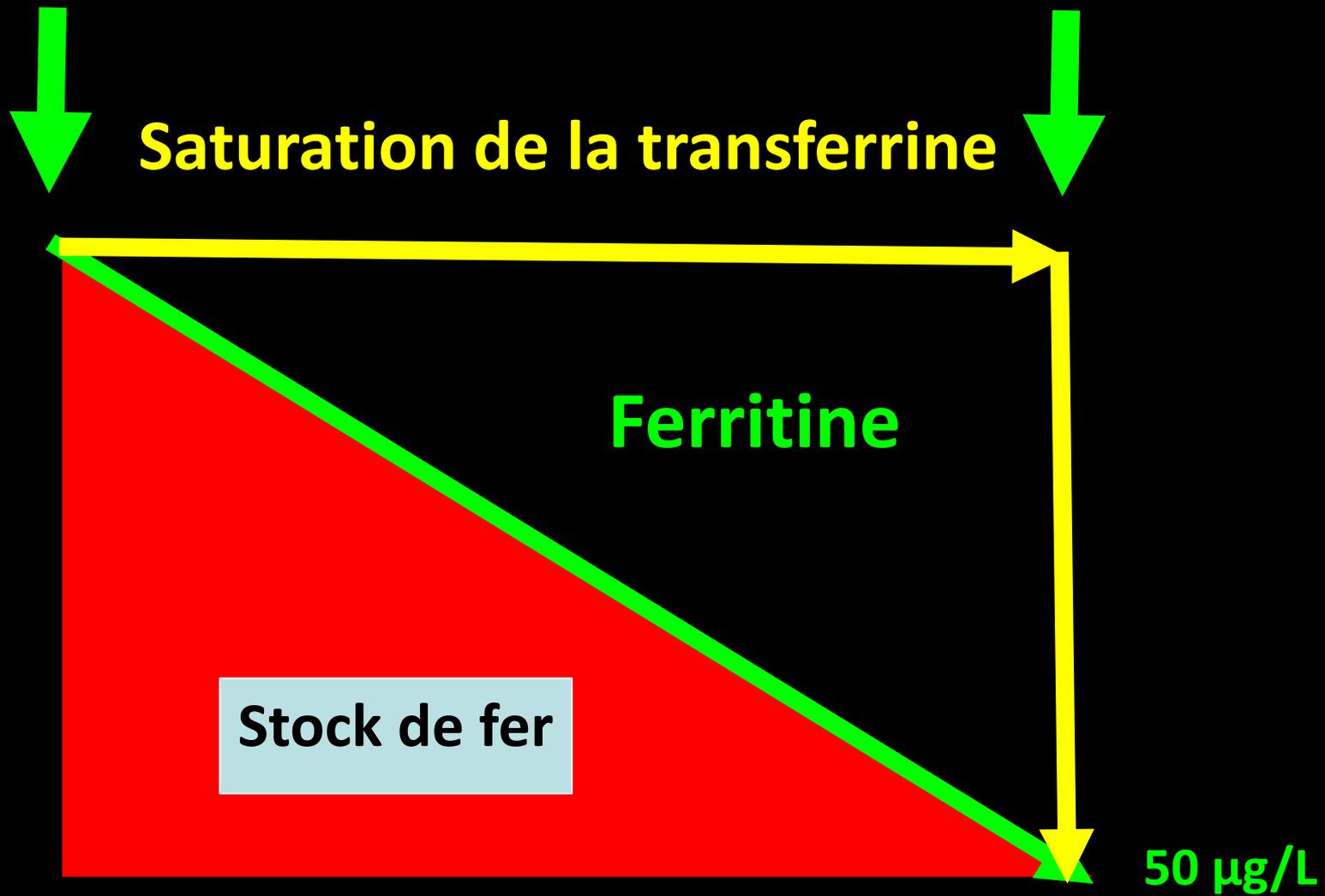


Ferritine

Stock de fer



**Se méfier de la
saturation de la transferrine !**



Traitement d'entretien...

Ferritine
≤50



ST ≤ 50

**Ferritine
 ≤ 50**



« Règle des « 50 » »

ST ≤ 50
Ferritine
≤ 50



« Règle des « 50 » »

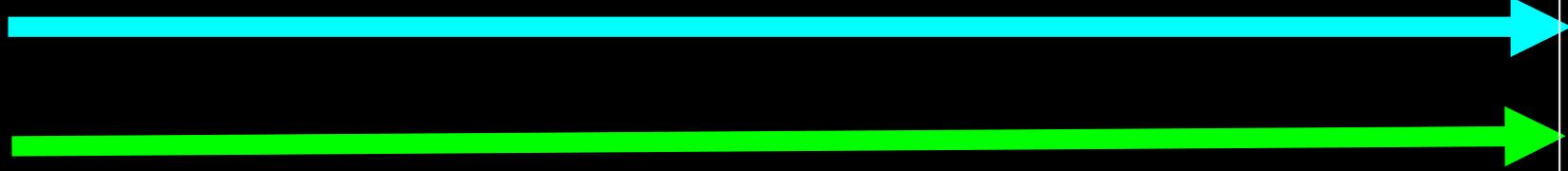
ST ≤ 50

Ferritine
 ≤ 50



TS
 > 75

Ferritine
 ≤ 50



« Normaliser » la ferritine reste l'objectif essentiel

**mais ne pas oublier la saturation de la transferrine
(2 fois par an)**

Hémochromatose

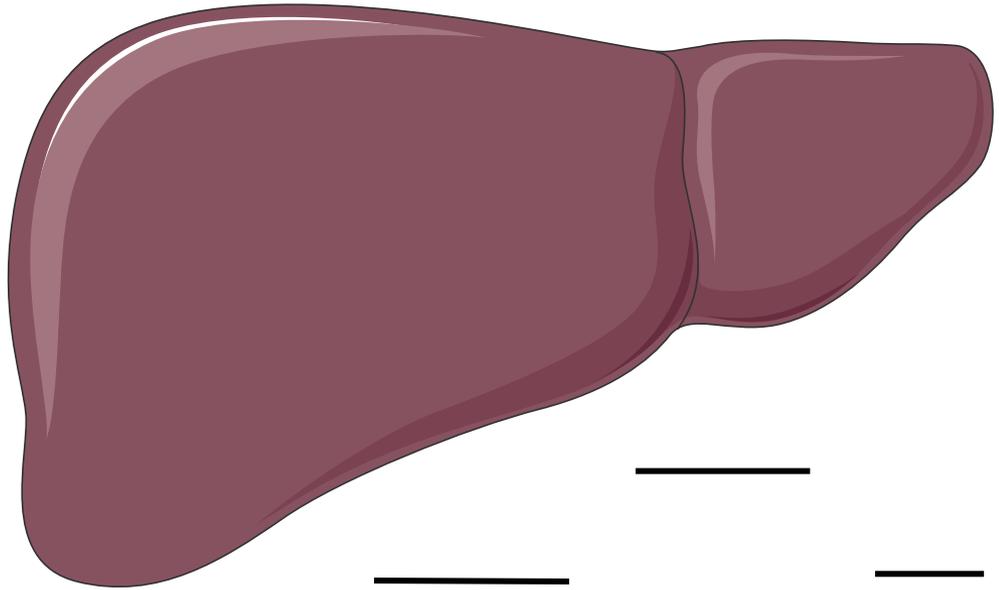
Définition et physiopathologie

Diagnostic

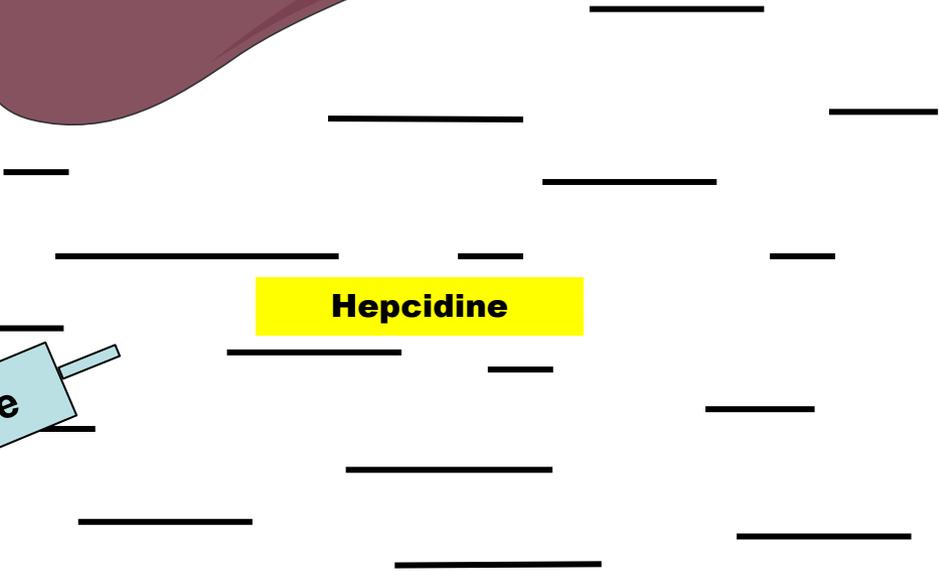
Traitement

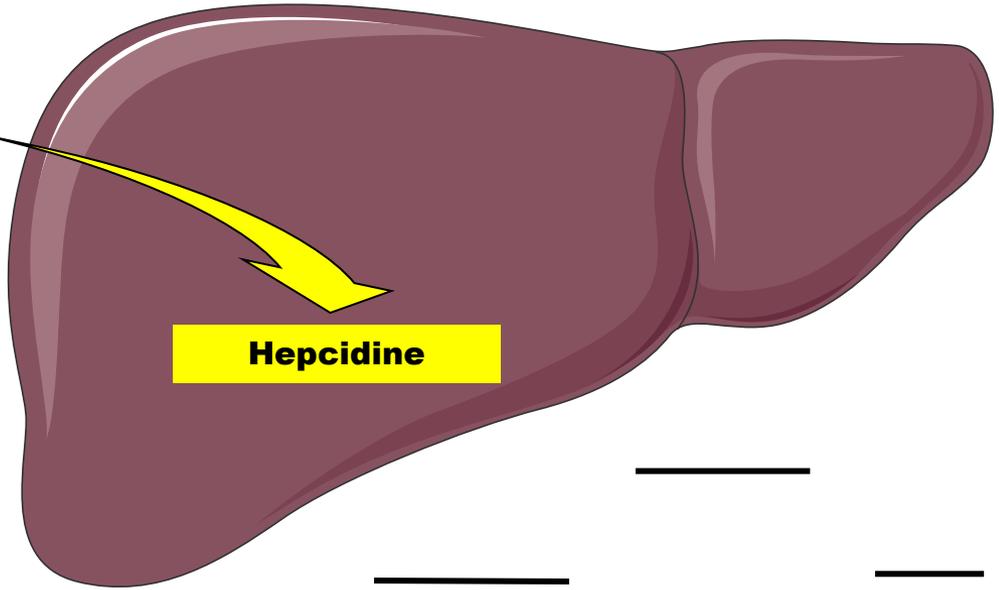
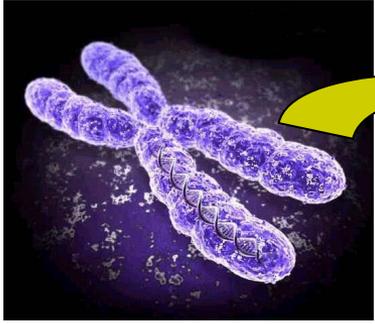
Aujourd'hui
Demain

Prévention



Hepcidine





Hepcidine



Hémochromatose

Définition et physiopathologie

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Traitement

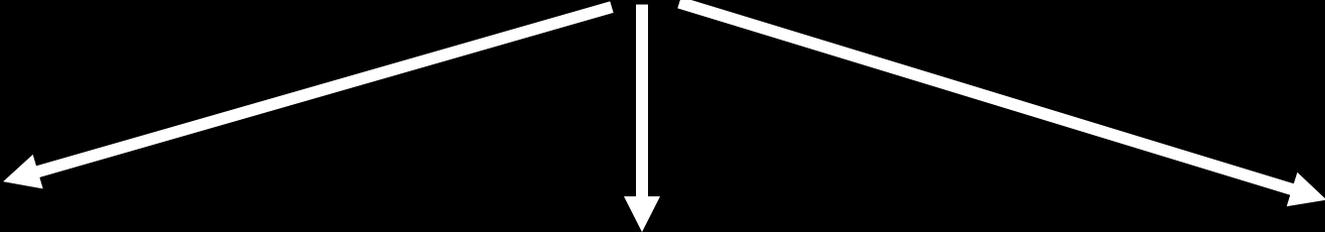
Prévention

Famille

Population

Test génétique en premier !

C282Y



C282Y

C282Y

C282Y/C282Y

Pas de transmission

Risque de transmission

Transmission

Pas de maladie

Pas de maladie

Risque de maladie

Hémochromatose

Définition et physiopathologie

Diagnostic

Traitement

Prévention

Famille

Population

Paramètres du fer en premier !

**Saturation
transferrine**

+

Ferritine