Hémochromatose:

Suivi et Traitement

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Rennes

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Surveillance et traitement d'une hémochromatose HFe1

- Quelle est la définition d'une hémochromatose
- Connaître la classification des hémochromatoses
- Traiter et savoir surveiller un malade qui a une hémochromatose

Conflits d'intérêts

- Laboratoire NOVARTIS

Définition et Classifications

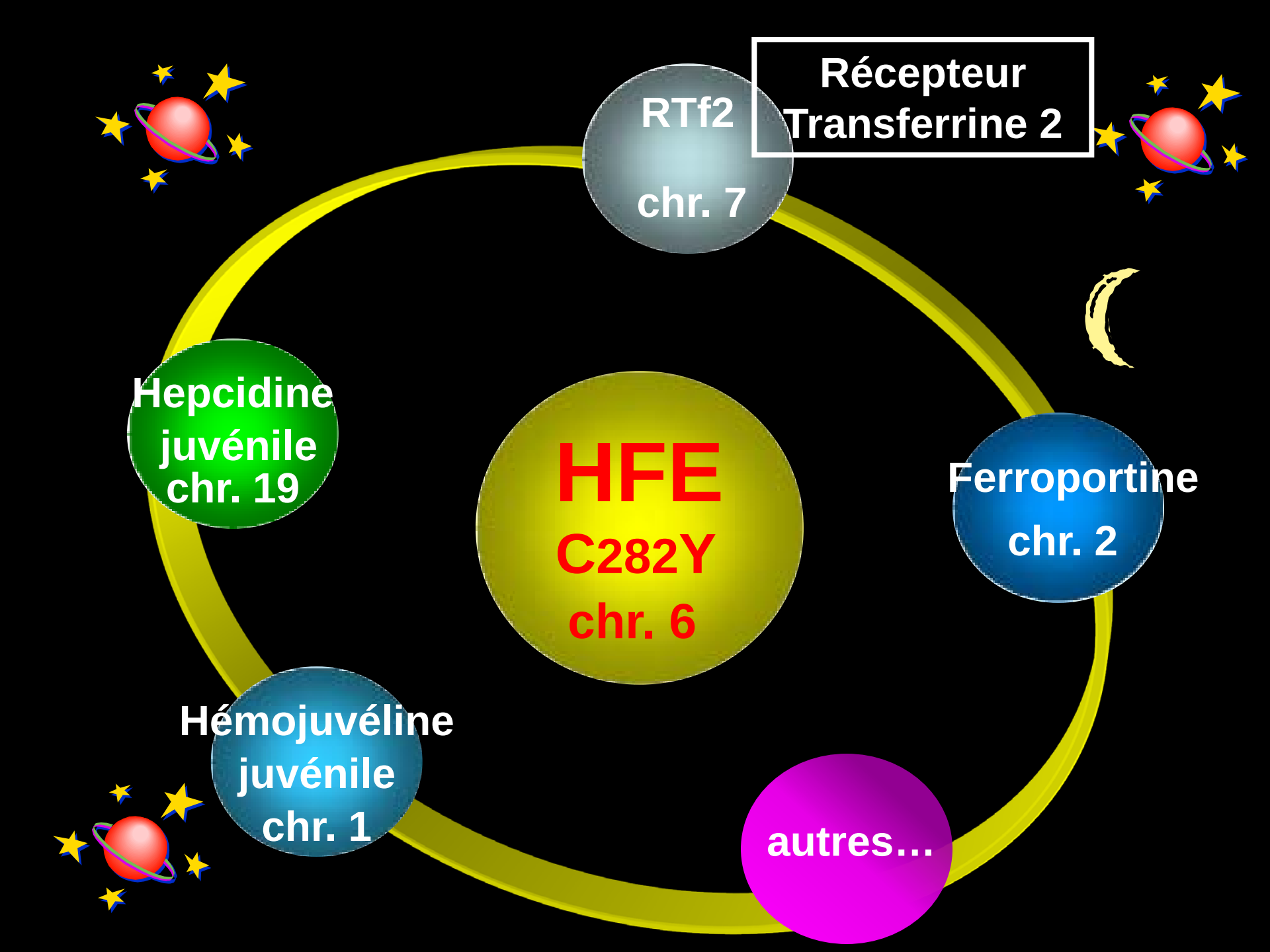
Traitement

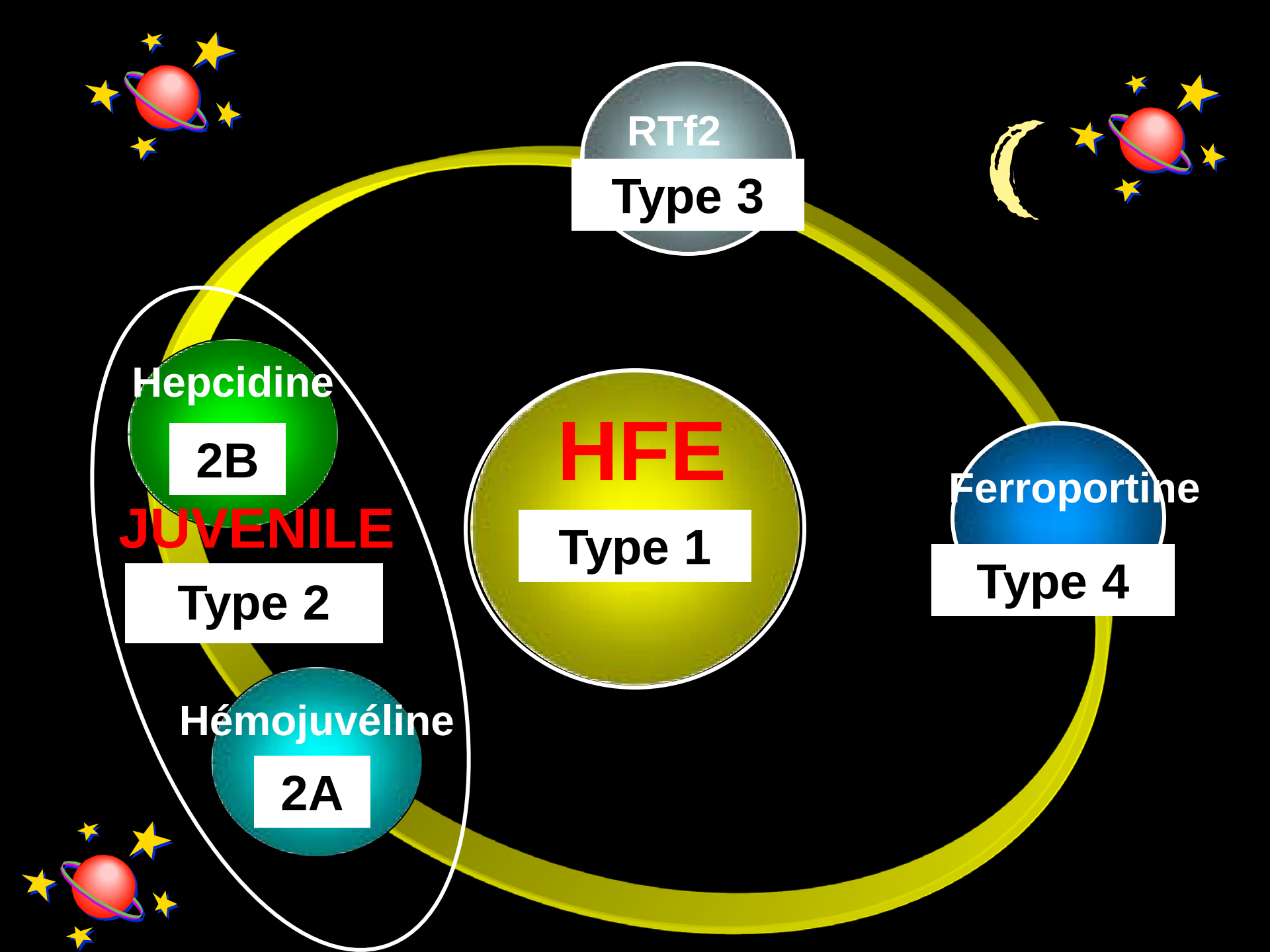
Surcharge en fer

chronique

génétique

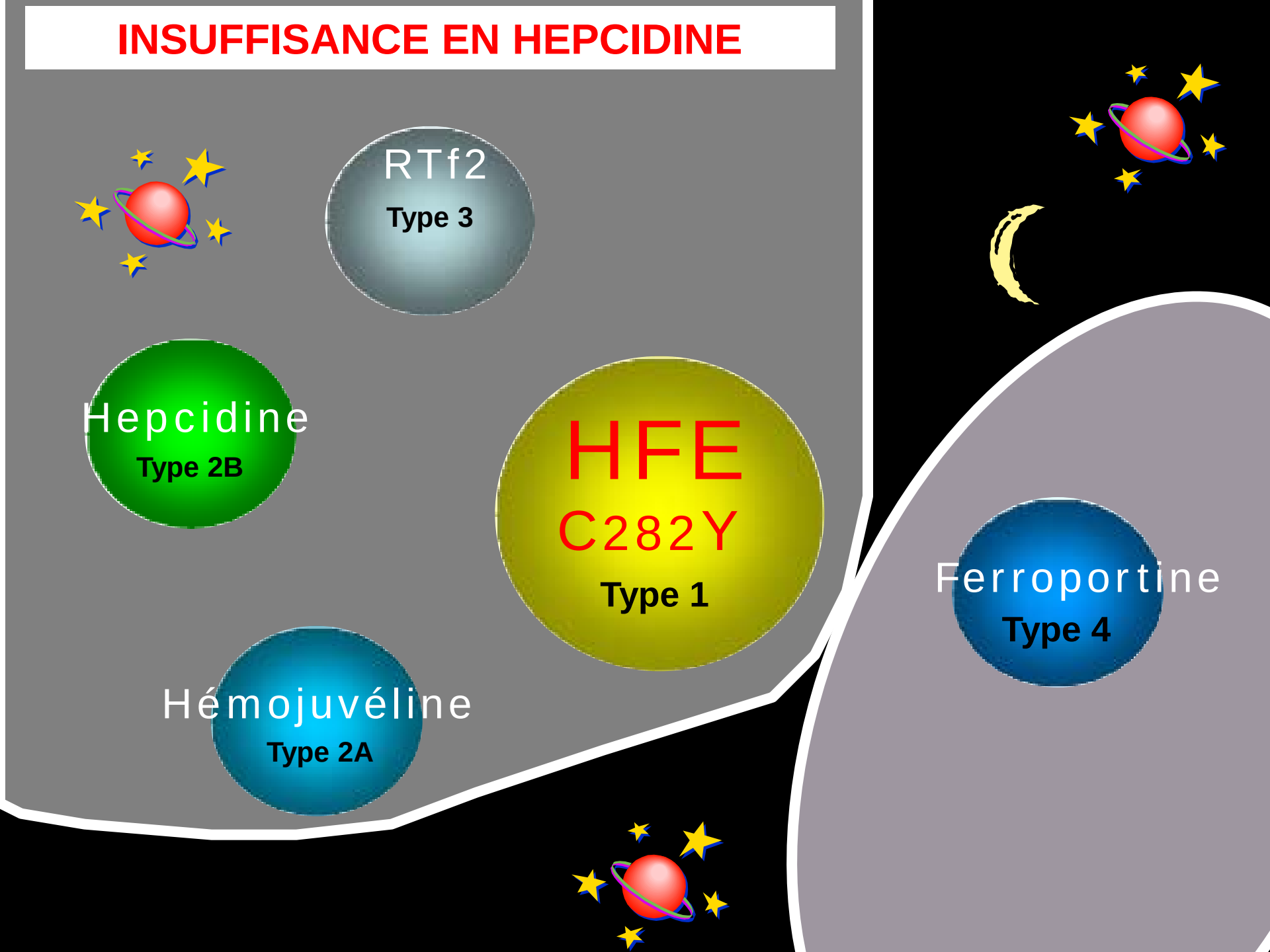
CLASSIFICATION GENETIQUE

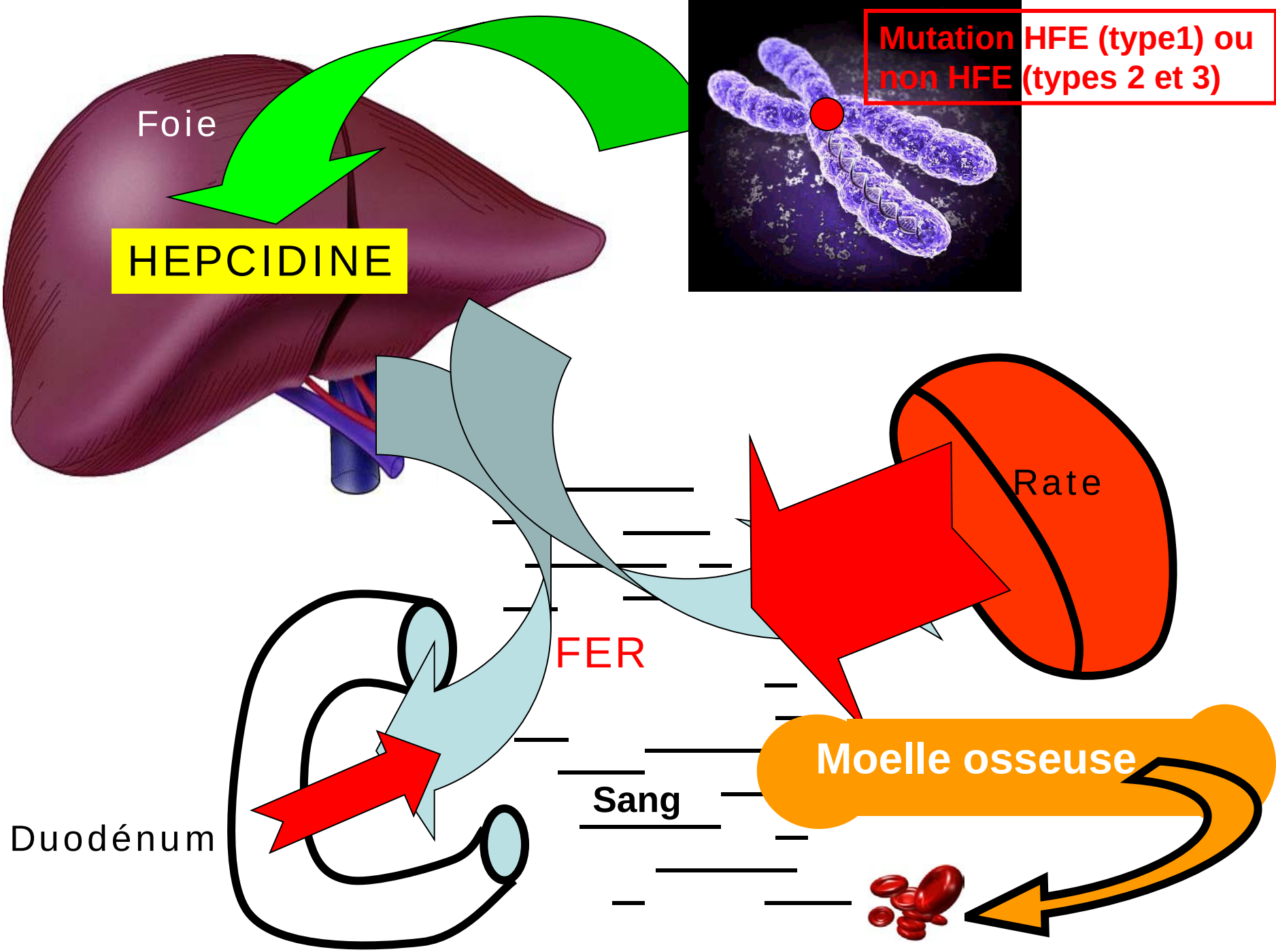

Récepteur Transferrine 2
RTf2
chr. 7
Hepcidine juvénile
chr. 19
HFE
C282Y
chr. 6
Ferroportine
chr. 2
Hémojuvéline juvénile
chr. 1
autres…

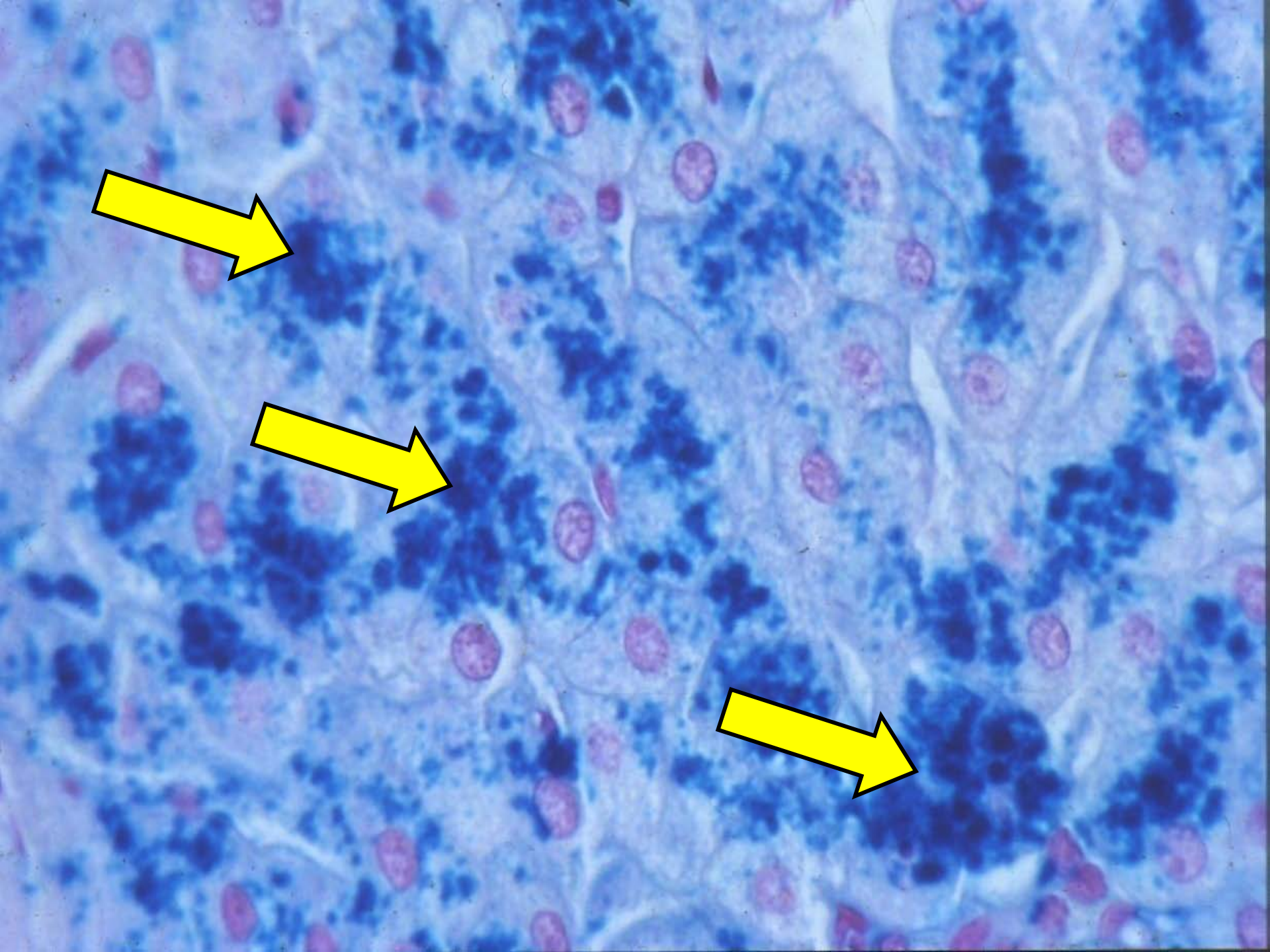

RTf2
Type 3
Hepcidine
2B
JUVENILE
Type 2
Hémojuvéline
2A
HFE
Type 1
Ferroportine
Type 4

CLASSIFICATION PHYSIOPATHOLOGIQUE

INSUFFISANCE EN HEPCIDINE




Mutation HFE (type1) ou non HFE (types 2 et 3)
Foie
HEPCIDINE
Rate
FER
Moelle osseuse
Sang
Duodénum



INSUFFISANCE EN HEPCIDINE

RTf2
Type 3

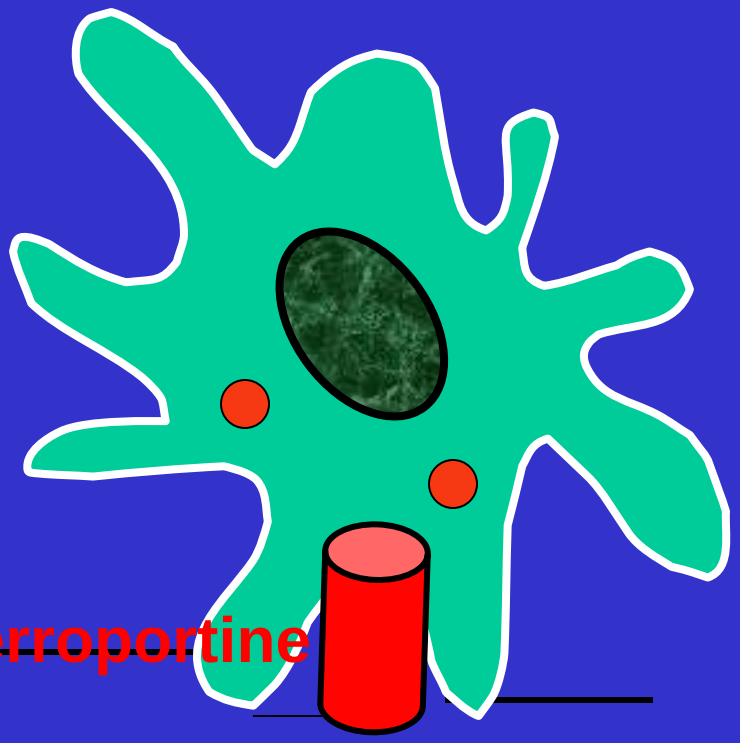
Hepcidine
Type 2B

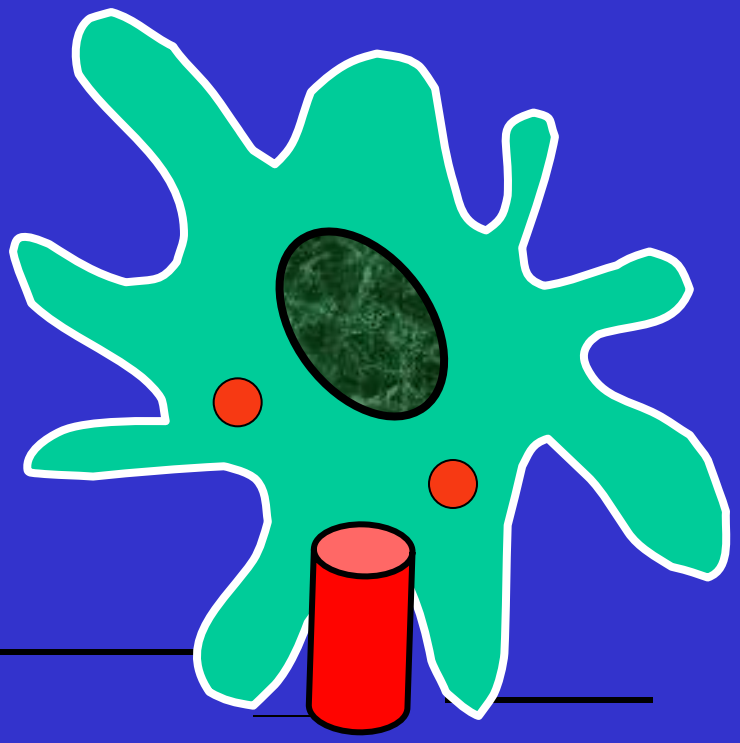
HFE
C282Y
Type 1

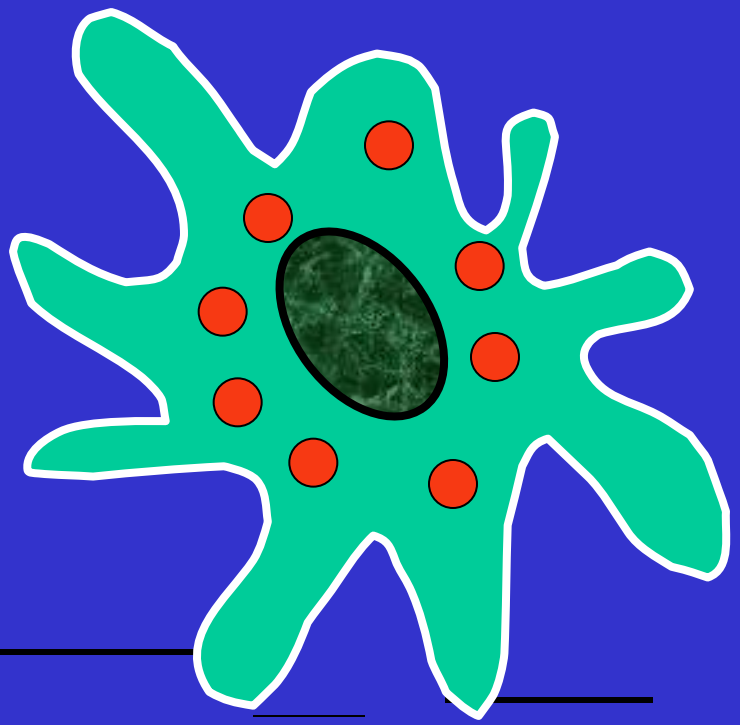
Hémojuvéline
Type 2A

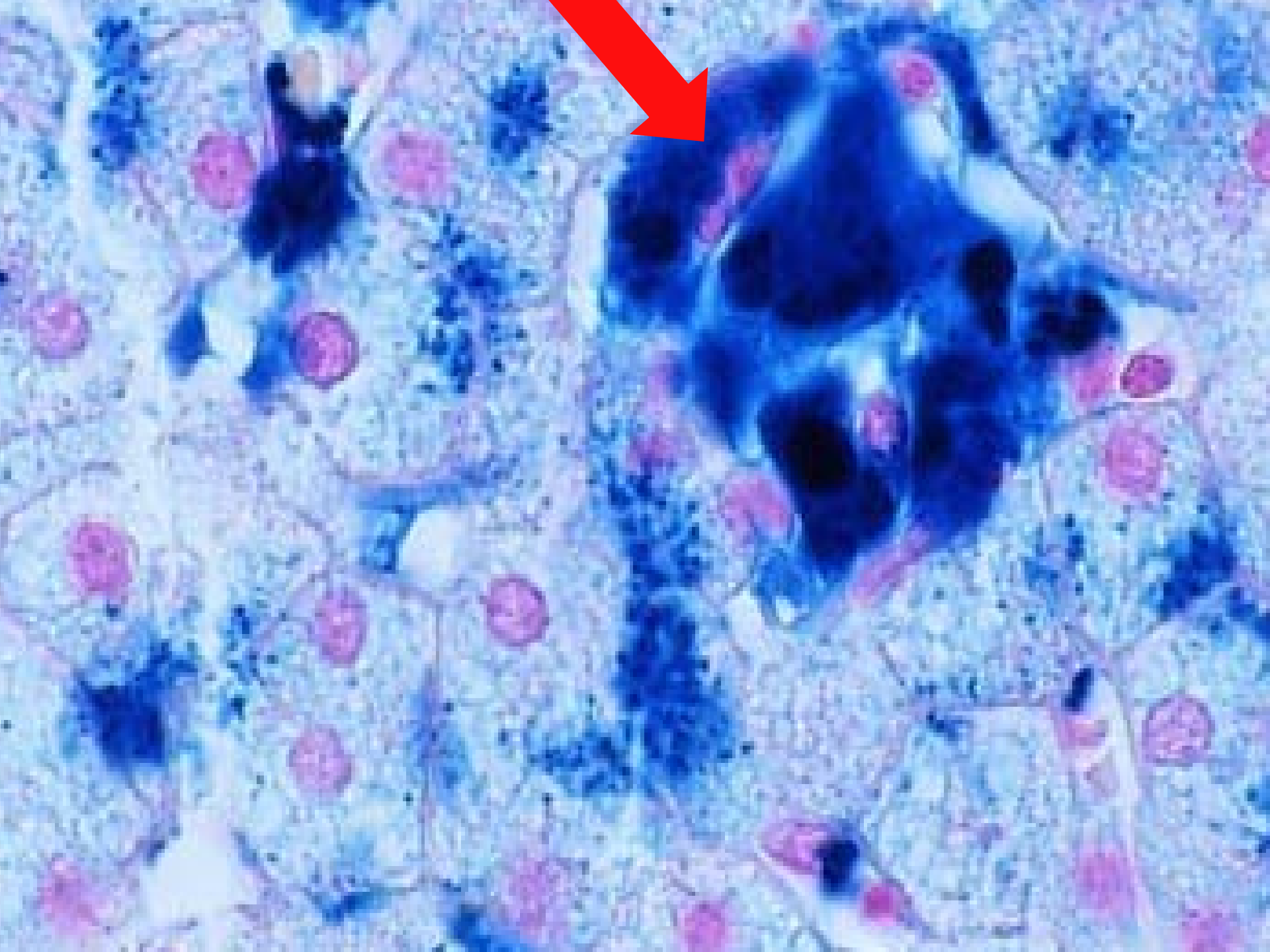
INSUFFISANCE EN FERROPORTINE

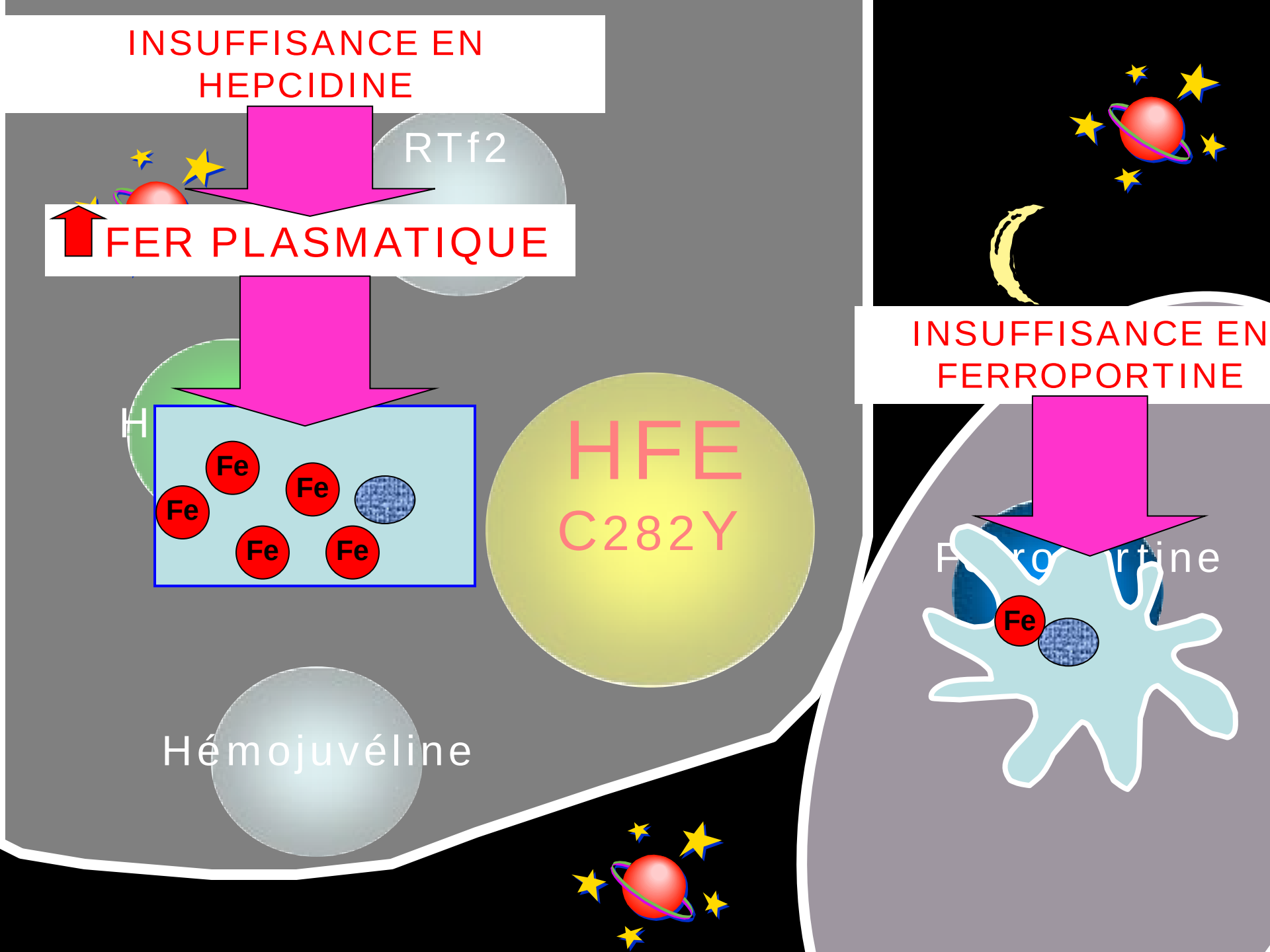
Ferroportine

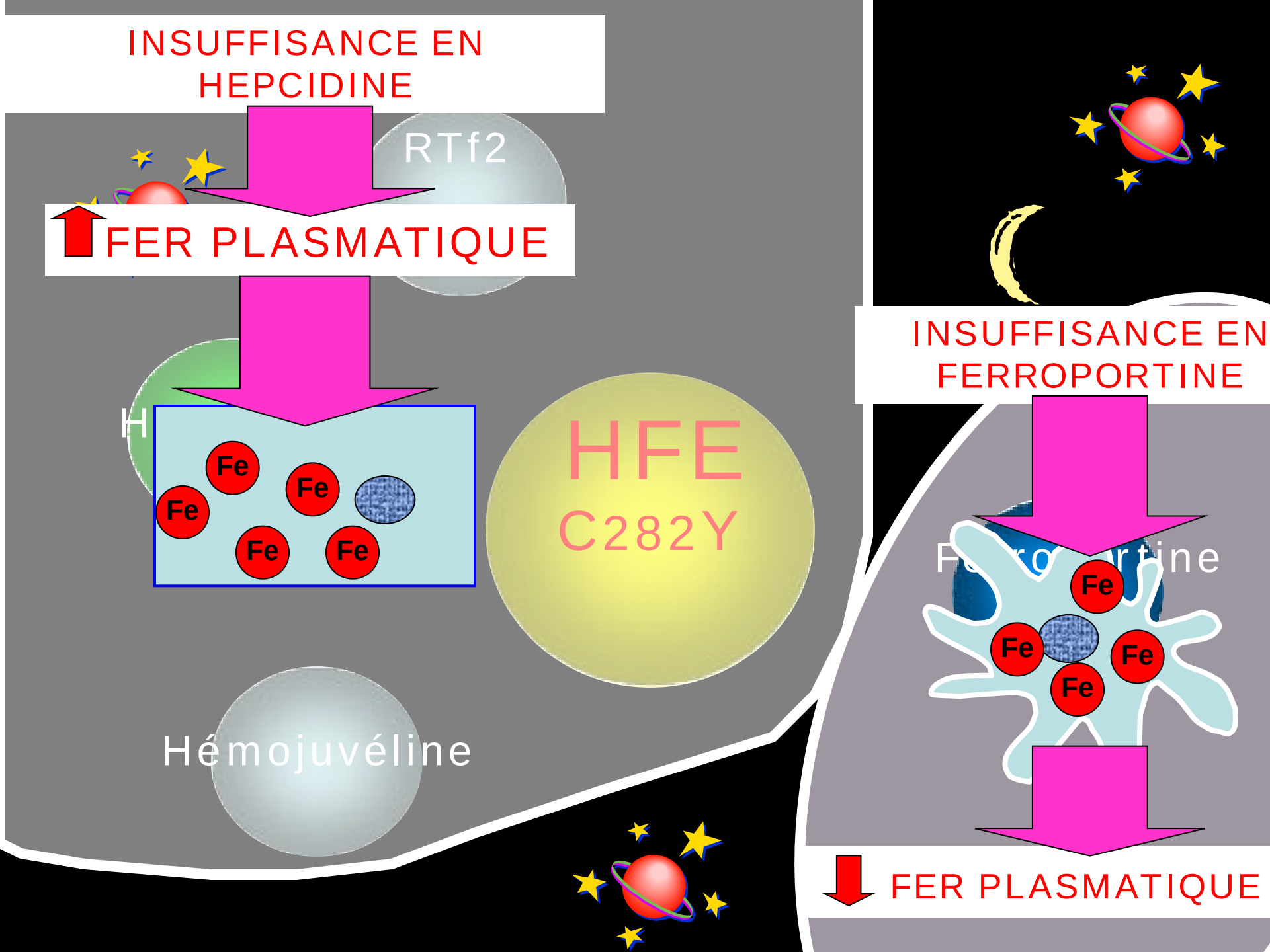

Ferroportine



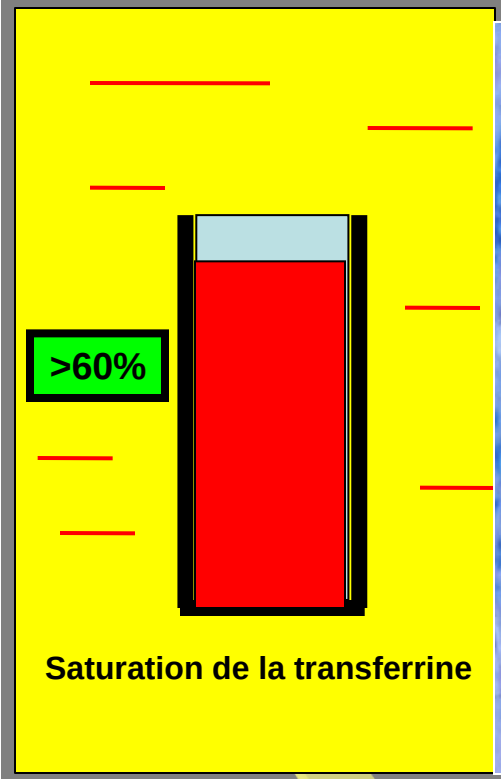


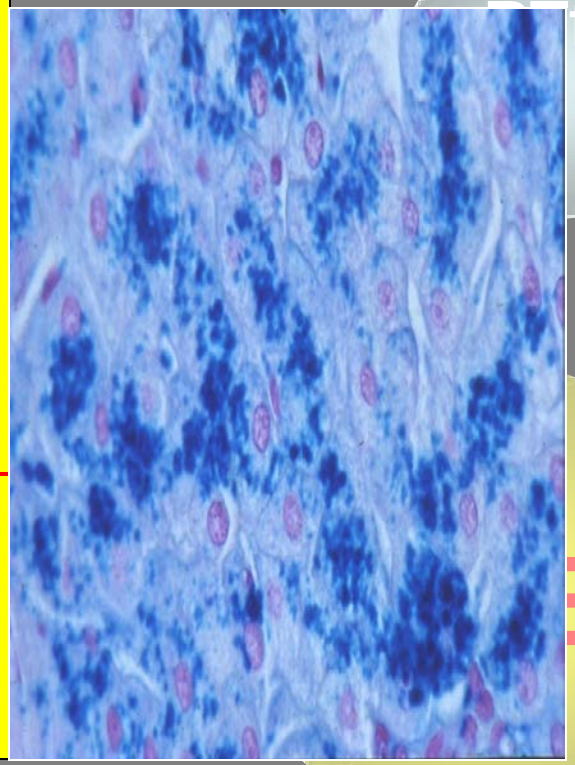


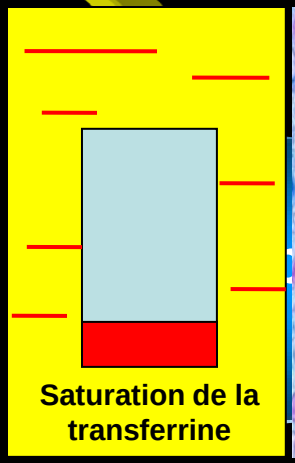

INSUFFISANCE EN HEPCIDINE
RTf2
FER PLASMATIQUE
Fe
Fe
Fe
Fe
Fe
HFE
C282Y
Hémojuvéline
INSUFFISANCE EN FERROPORTINE
Fe

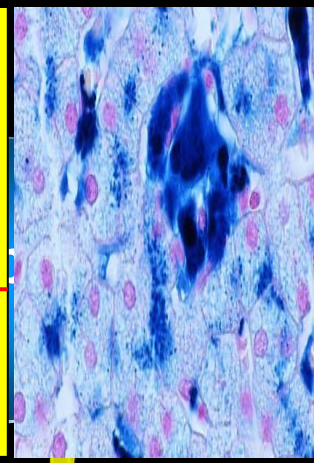

INSUFFISANCE EN HEPCIDINE
RTf2
FER PLASMATIQUE
Fe
Fe
Fe
Fe
Fe
HFE
C282Y
Hémojuvéline
INSUFFISANCE EN FERROPORTINE
Fe
Fe
Fe
Fe
FER PLASMATIQUE

CLASSIFICATION CLINIQUE





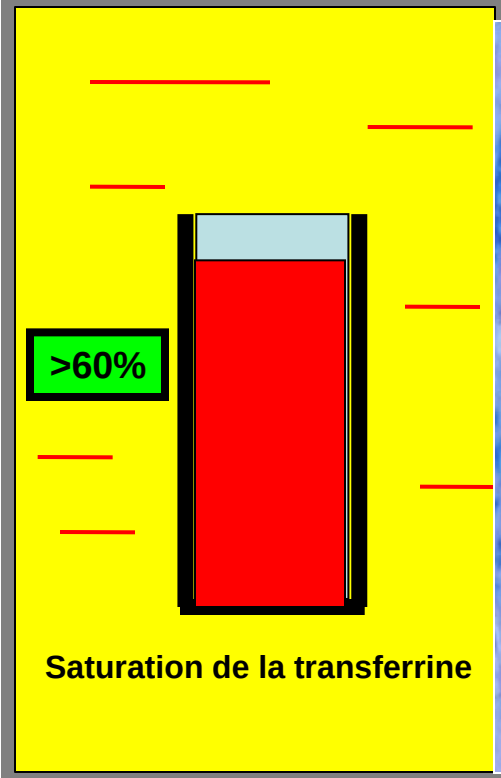


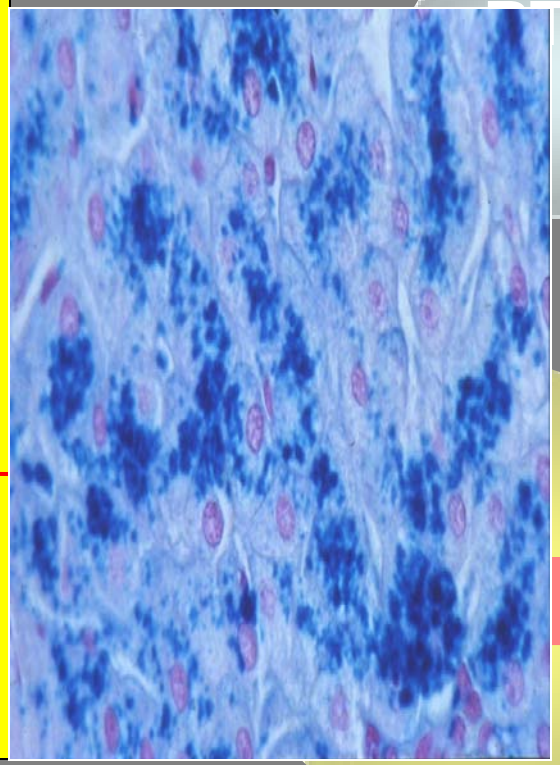


Hémojuvéline (H. juvénile)

Définition et Classifications

Suivi et Traitement

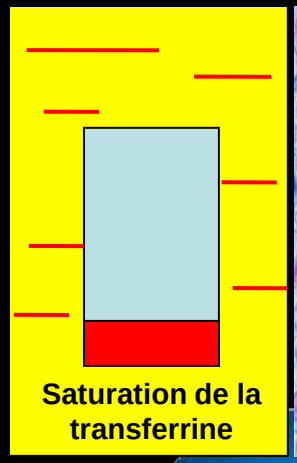


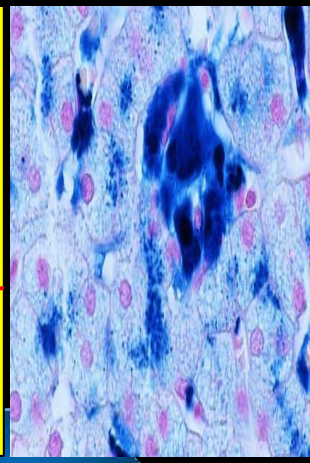


TTf2

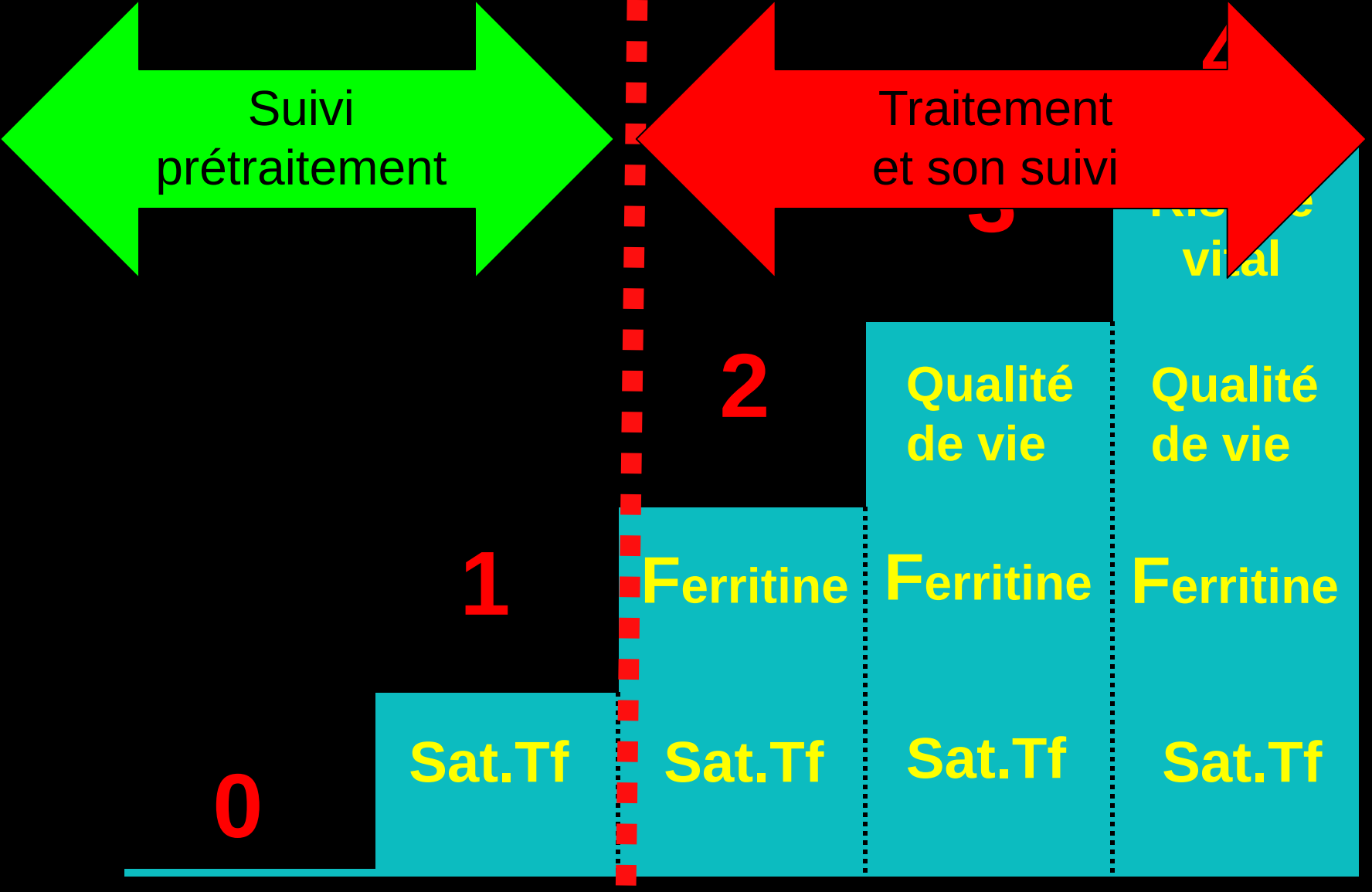
E

Hémojuvéline
(H. juvénile)





Ferroportine


Suivi
prétraitement
Traitement
et son suivi
0
1
2
Sat.Tf
Ferritine
Sat.Tf
Qualité
de vie
Ferritine
Sat.Tf
Qualité
de vie
Ferritine
Sat.Tf

Suivi prétraitement

0	1	2	3	4
	Sat.Tf	Ferritine Sat.Tf	Qualité de vie Ferritine Sat.Tf	Risque vital Qualité de vie Ferritine Sat.Tf

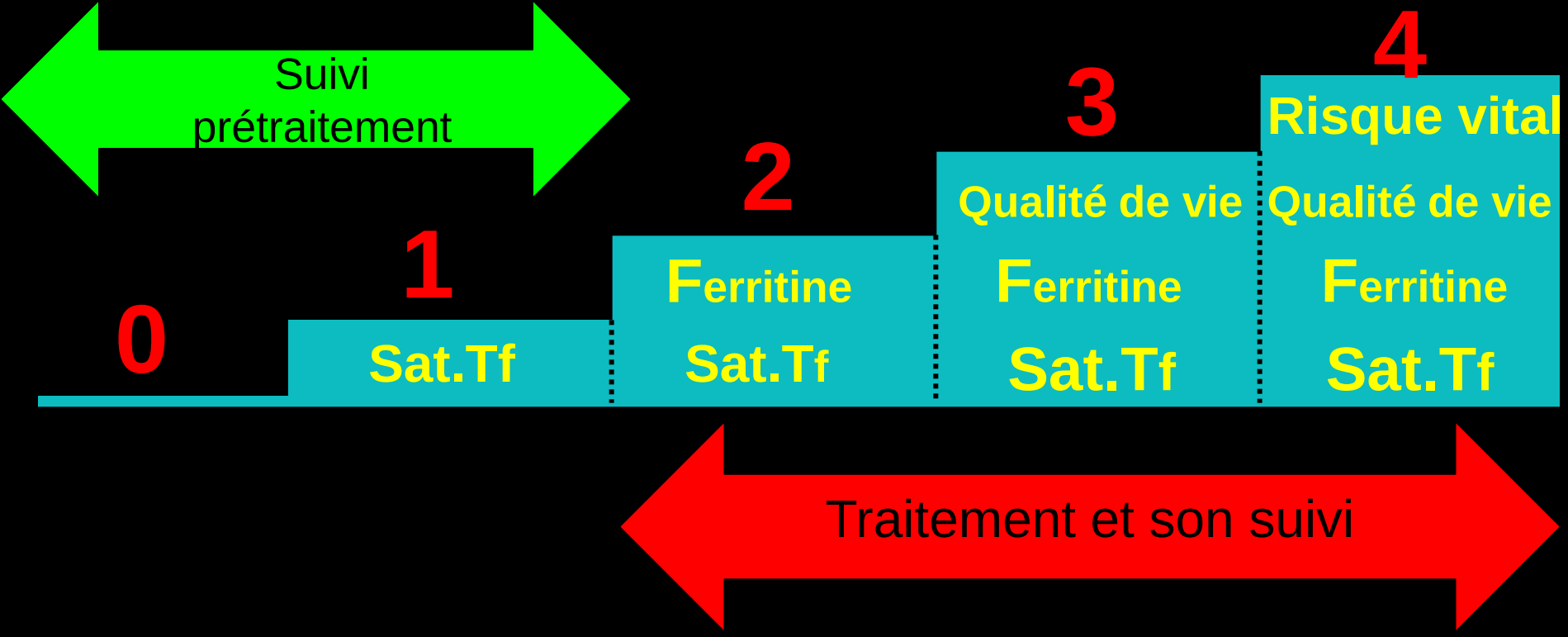
Evaluation clinique

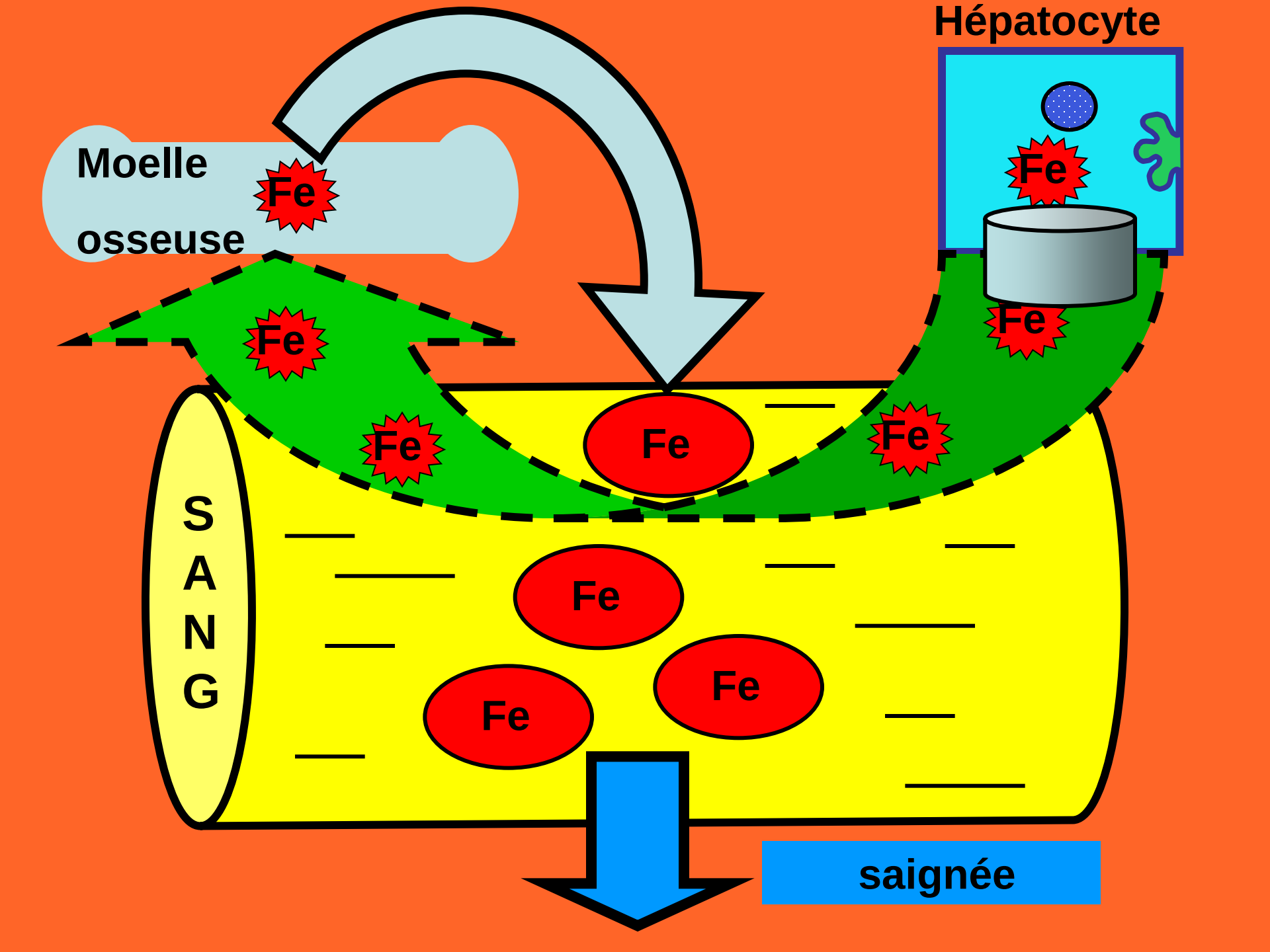
/ 3 ans	/ an

Evaluation biologique

STf + Ferritinémie

/ 3 ans	/ an

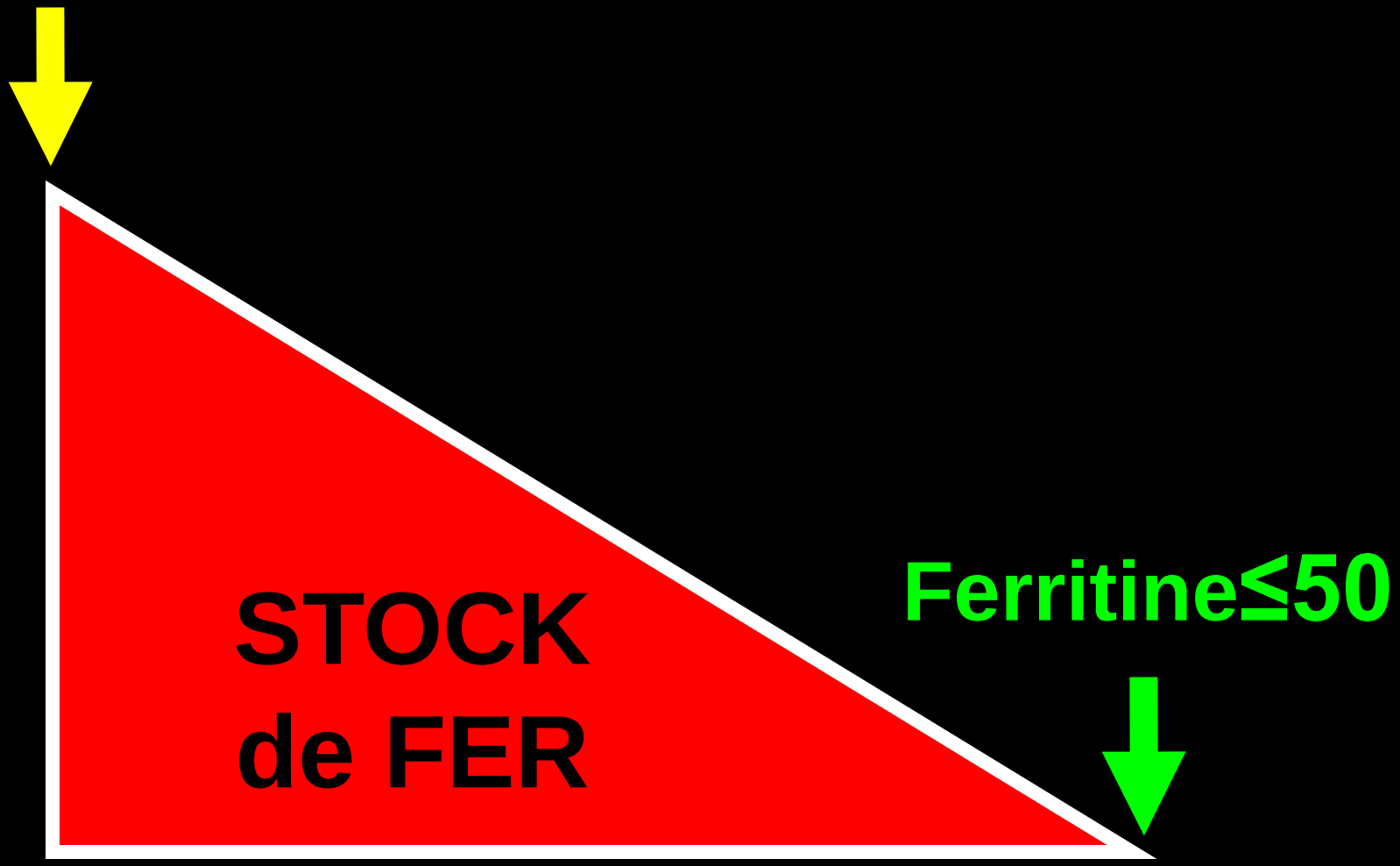

Suivi
prétraitement
0
1
Sat.Tf
2
Ferritine
Sat.Tf
3
Qualité de vie
Ferritine
Sat.Tf
4
Risque vital
Qualité de vie
Ferritine
Sat.Tf
Traitement et son suivi

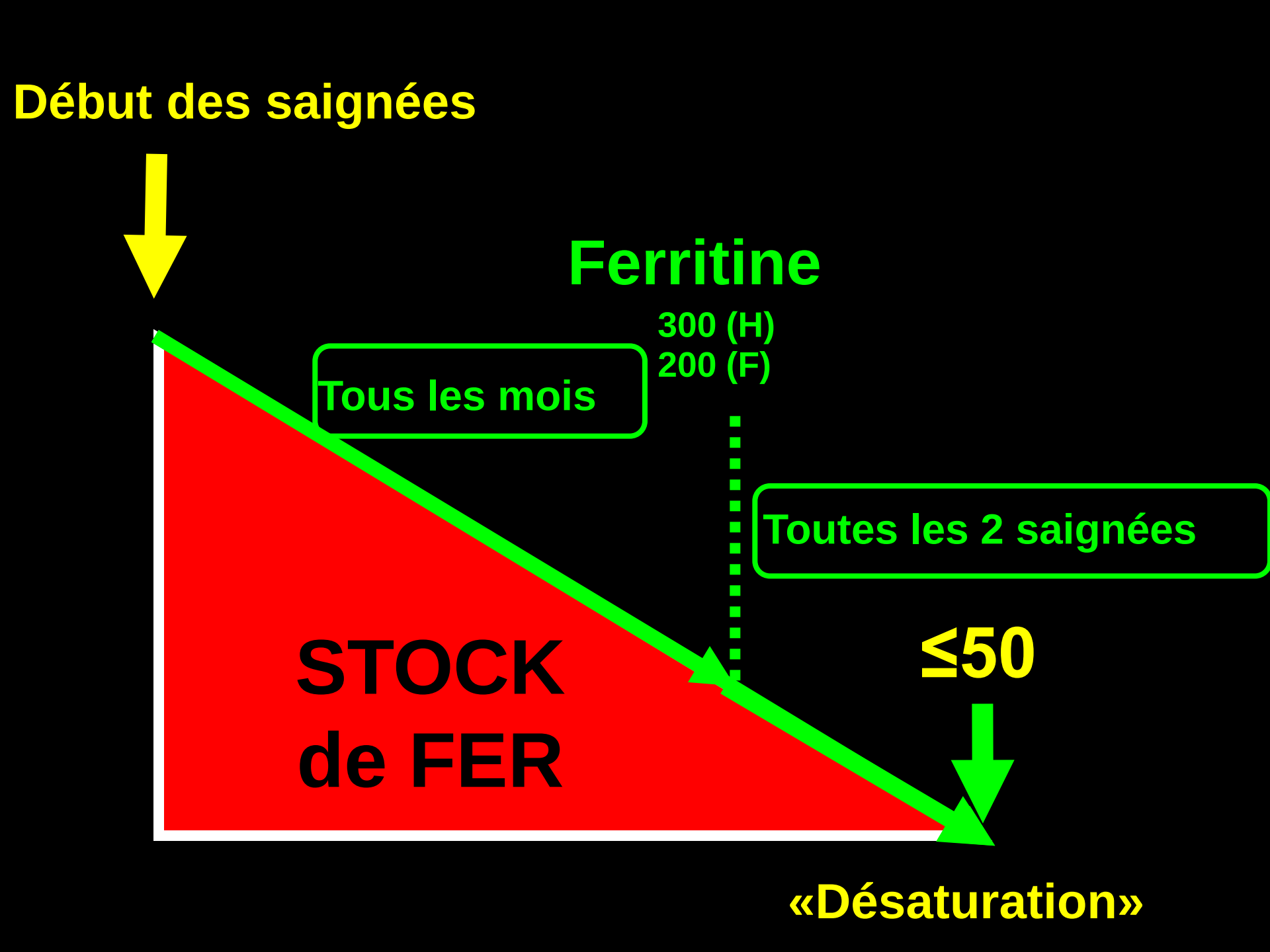

Hépatocyte
Moelle
osseuse
Fe
Fe
Fe
Fe
Fe
Fe
Fe
Fe
Fe
S
A
N
G
saignée

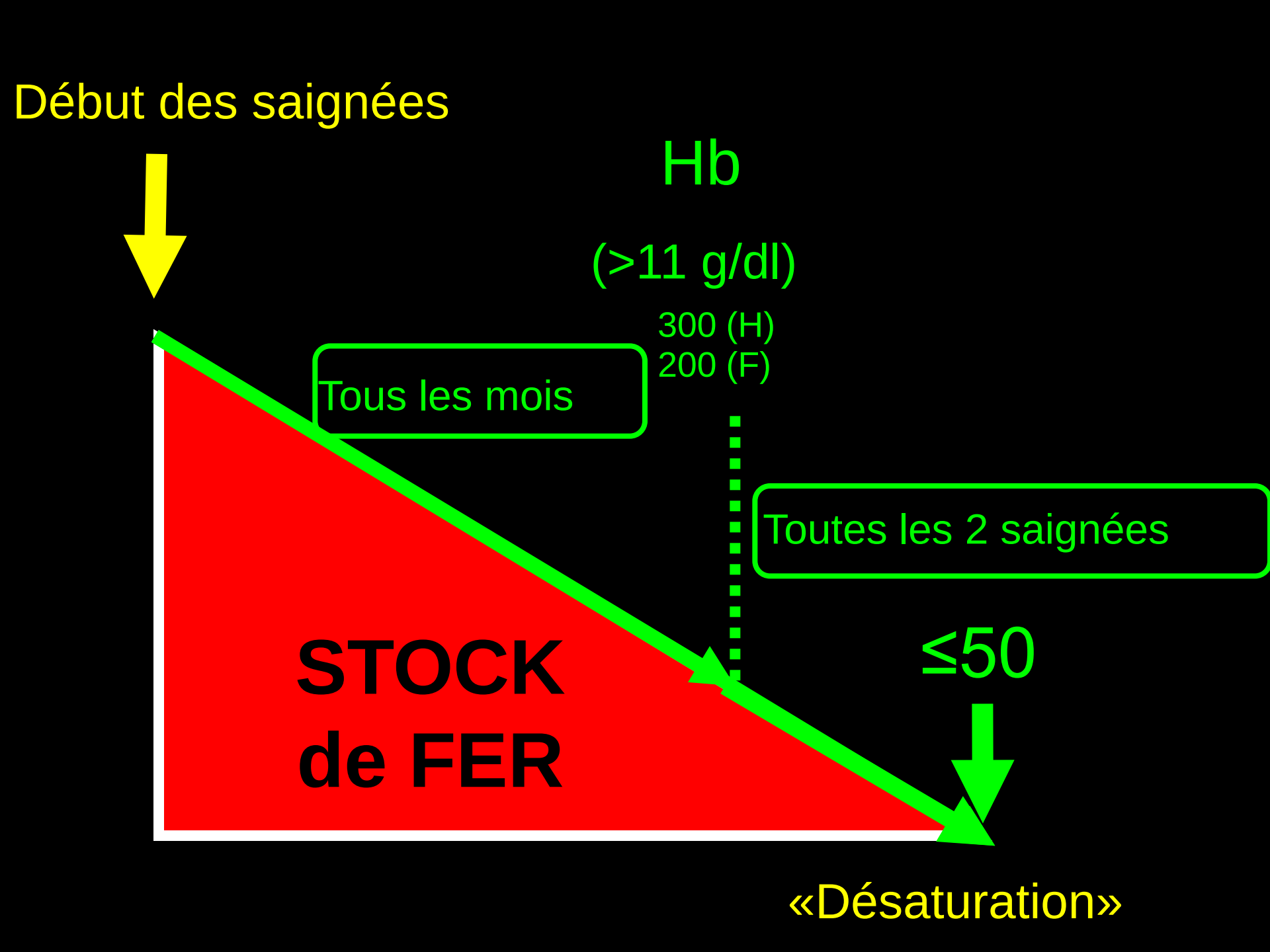
Phase d’induction

7ml/kg (≤550ml)

7ml/kg/semaine (≤550ml)



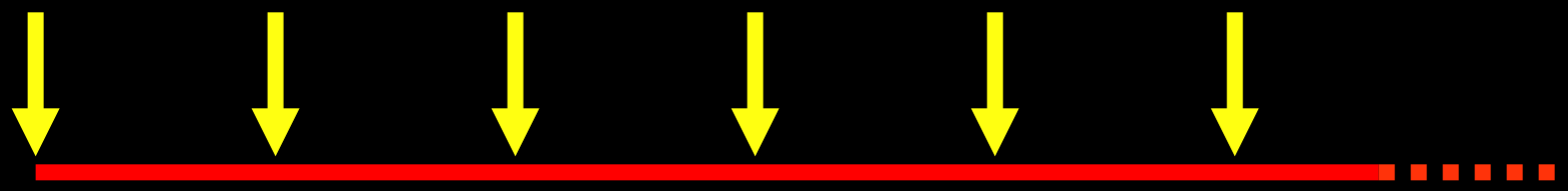

Début des saignées
Ferritine
300 (H)
200 (F)
Tous les mois
Toutes les 2 saignées
STOCK
de FER
≤50
«Désaturation»


Début des saignées
Hb
(>11 g/dl)
300 (H)
200 (F)
Tous les mois
Toutes les 2 saignées
STOCK
de FER
≤50
«Désaturation»

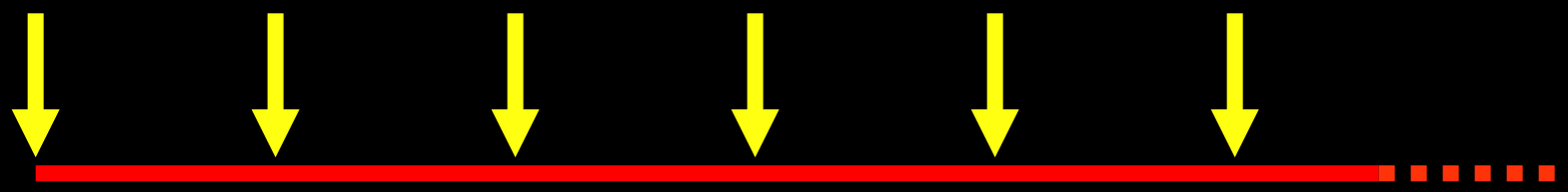
Phase d’entretien

Objectif

Ferritine ≤ 50



Ferritine ≤ 50



Ferritine toutes les 2 saignées

Hb avant saignée

Hôpital

EFS

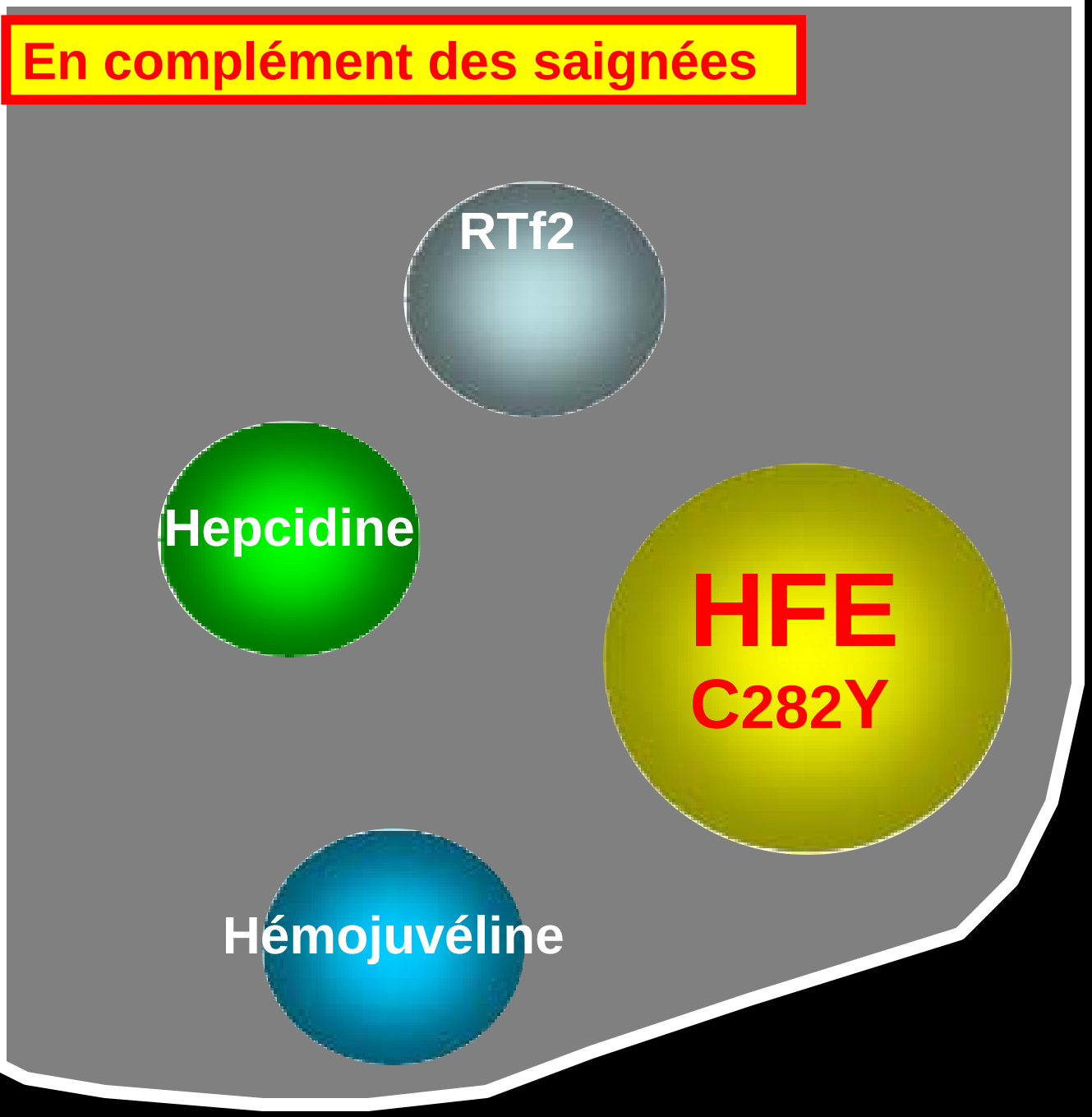
Cabinet médical

Cabinet IDE

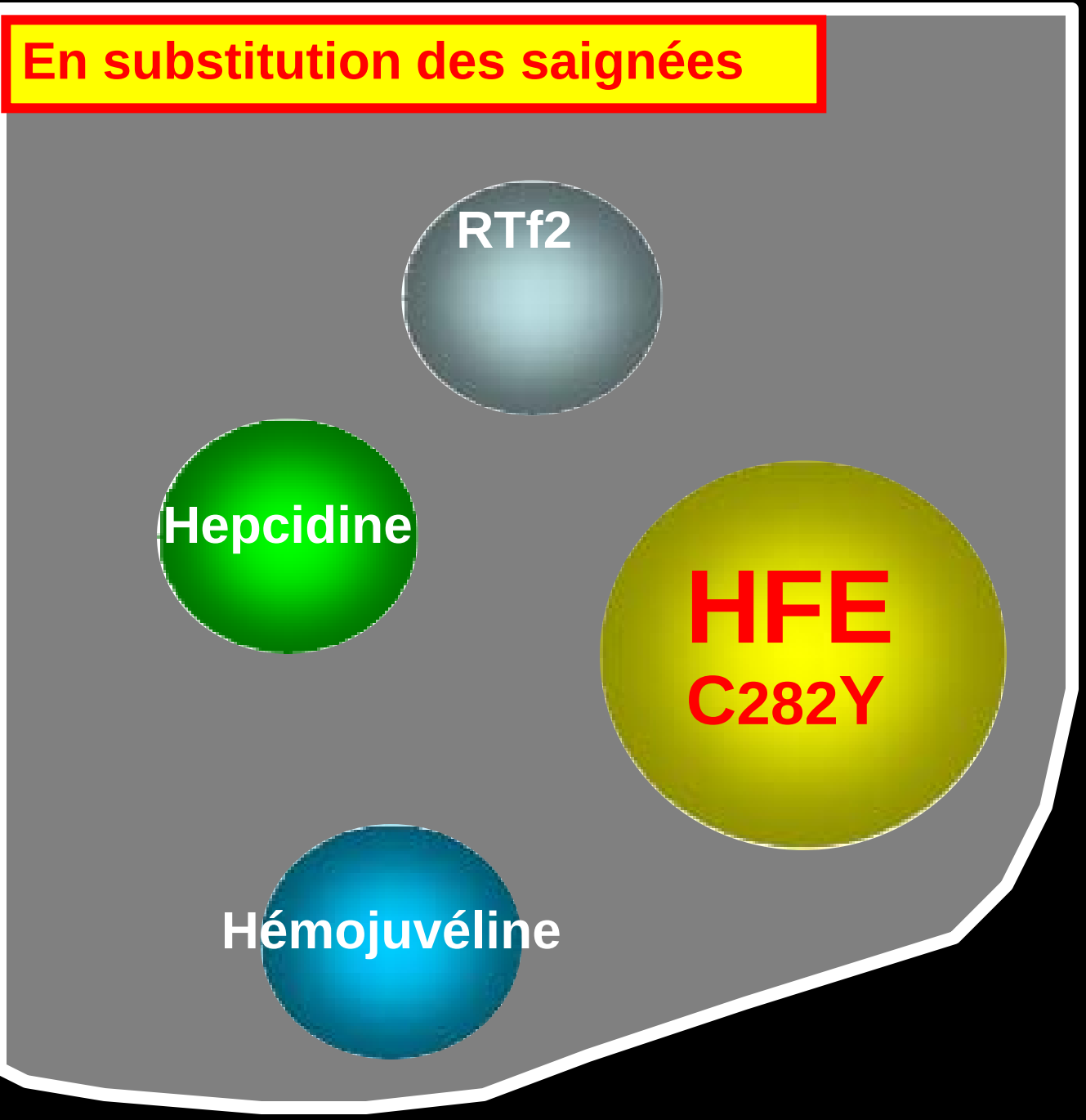
Domicile

Déférasirox (Exjade®) ?
CICL670A0107
ICL670 500mg
08/2003

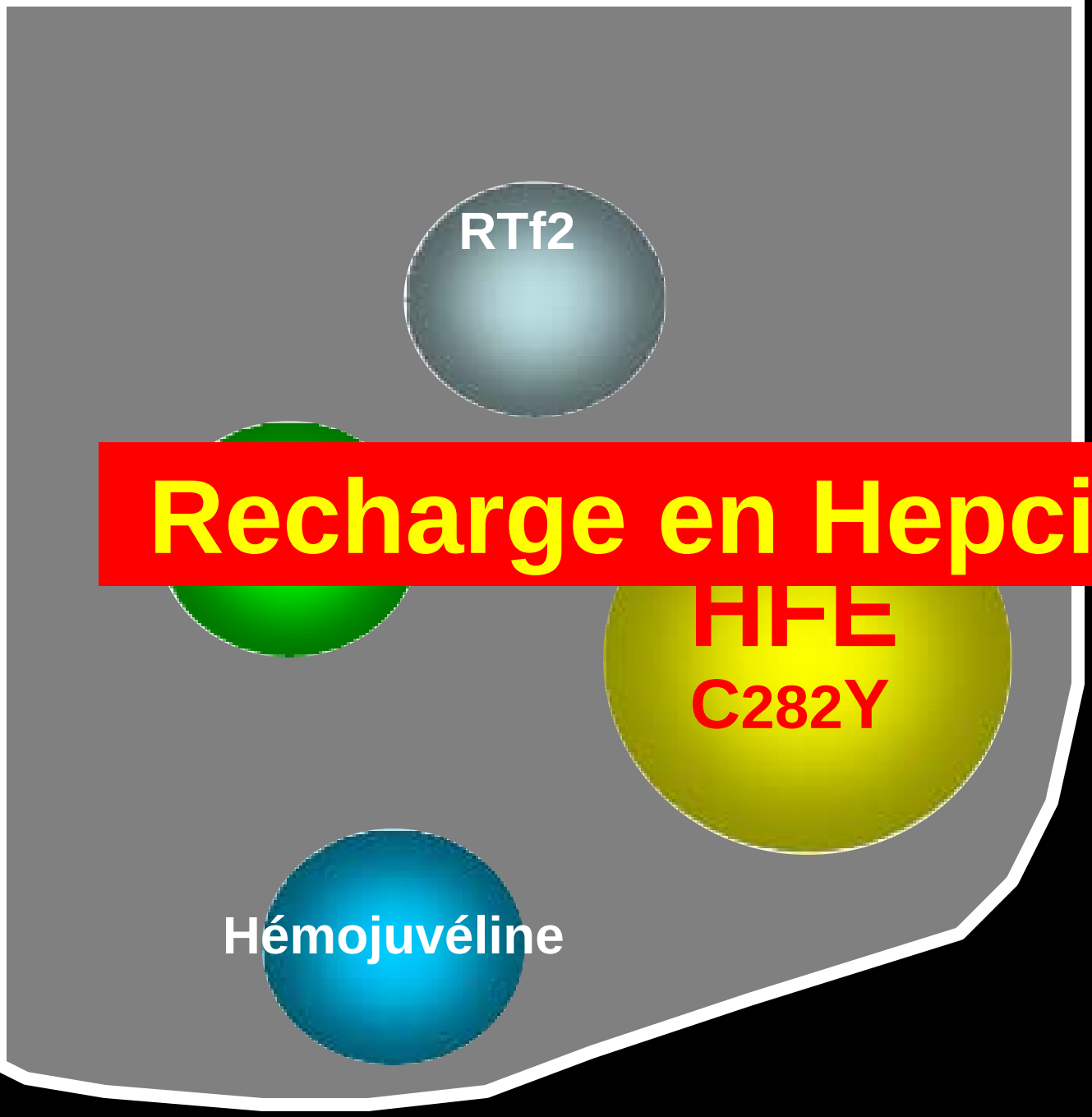
En complément des saignées



En substitution des saignées



Recharge en Hepcidine…



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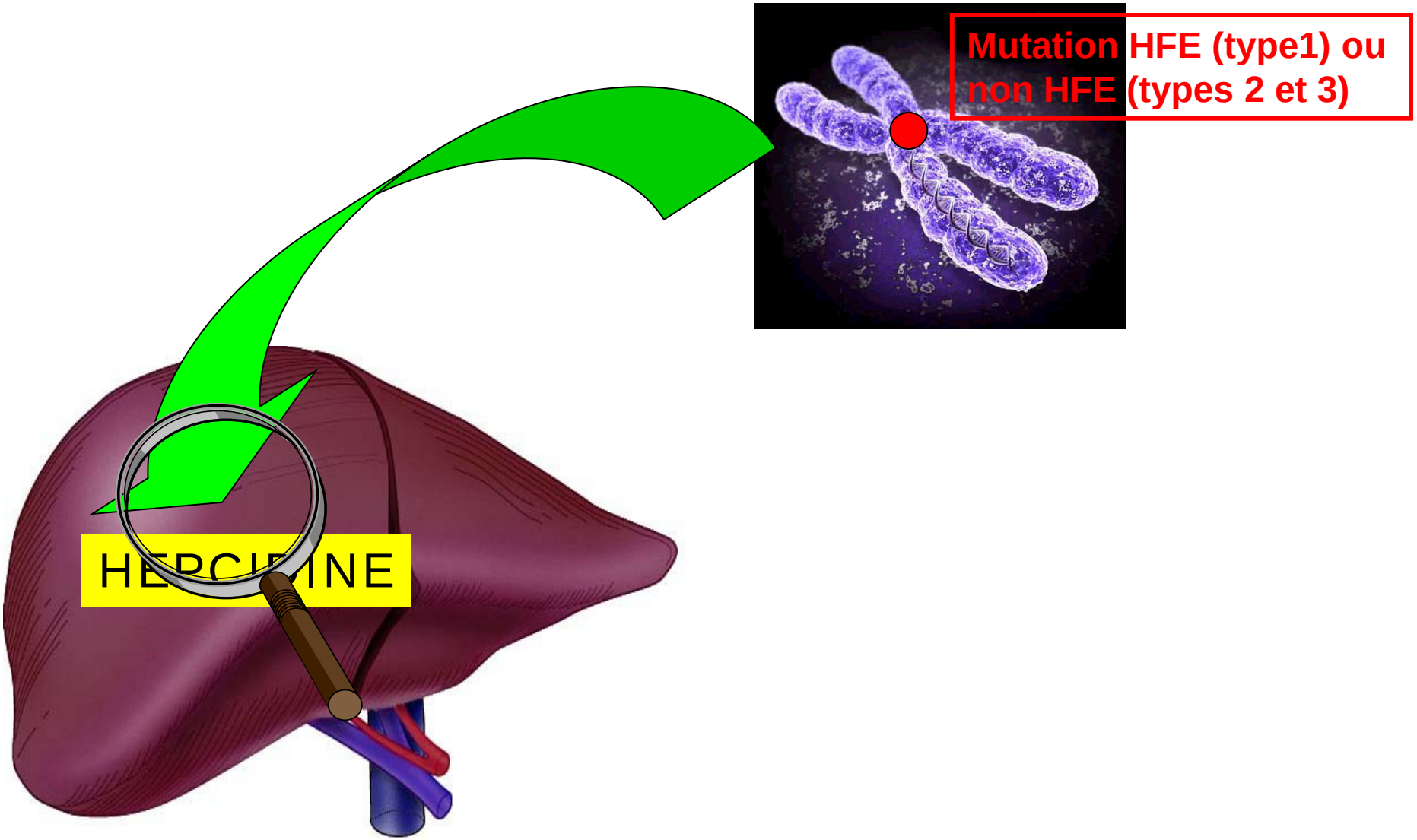
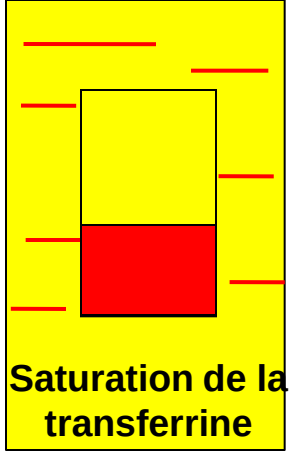
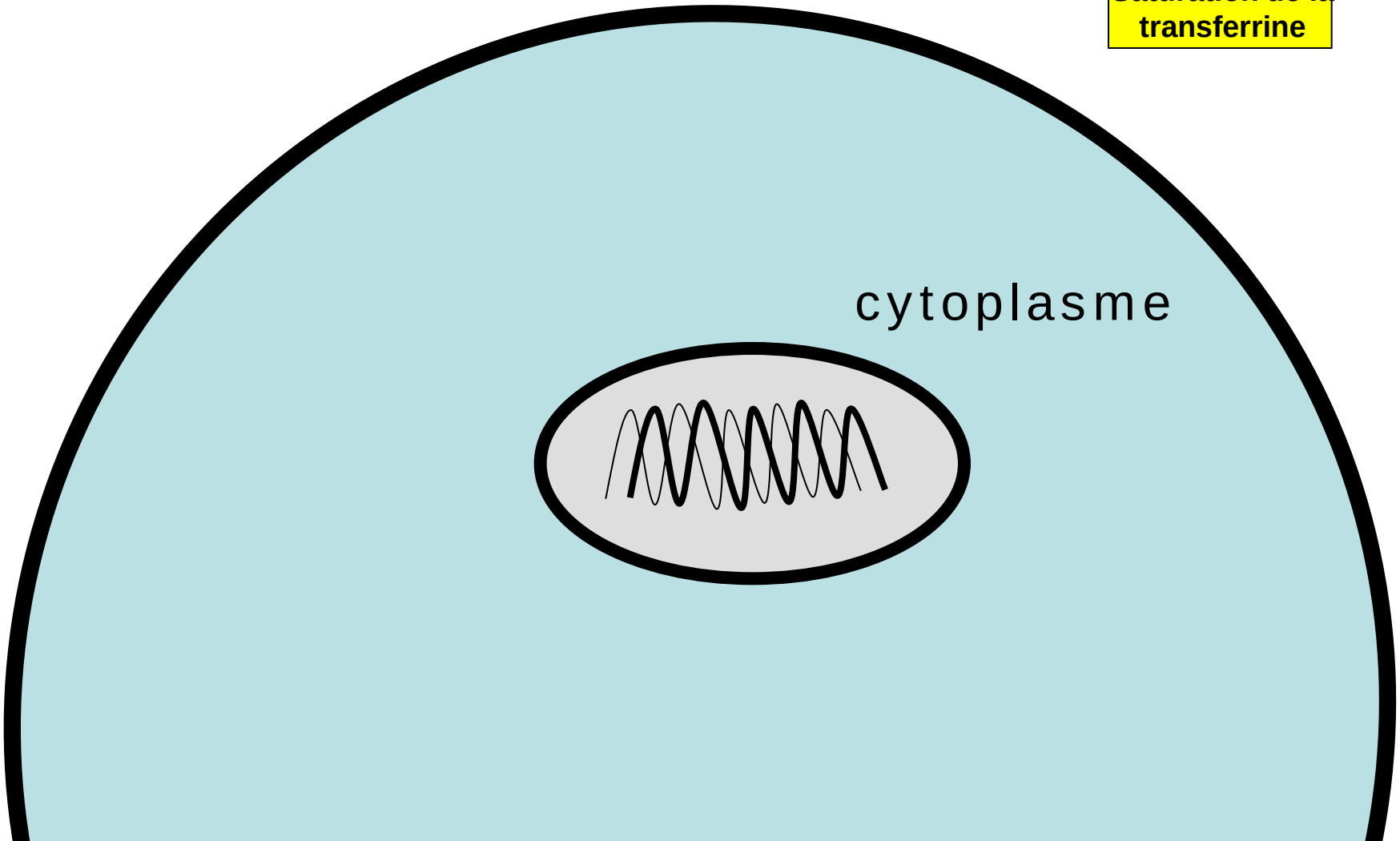
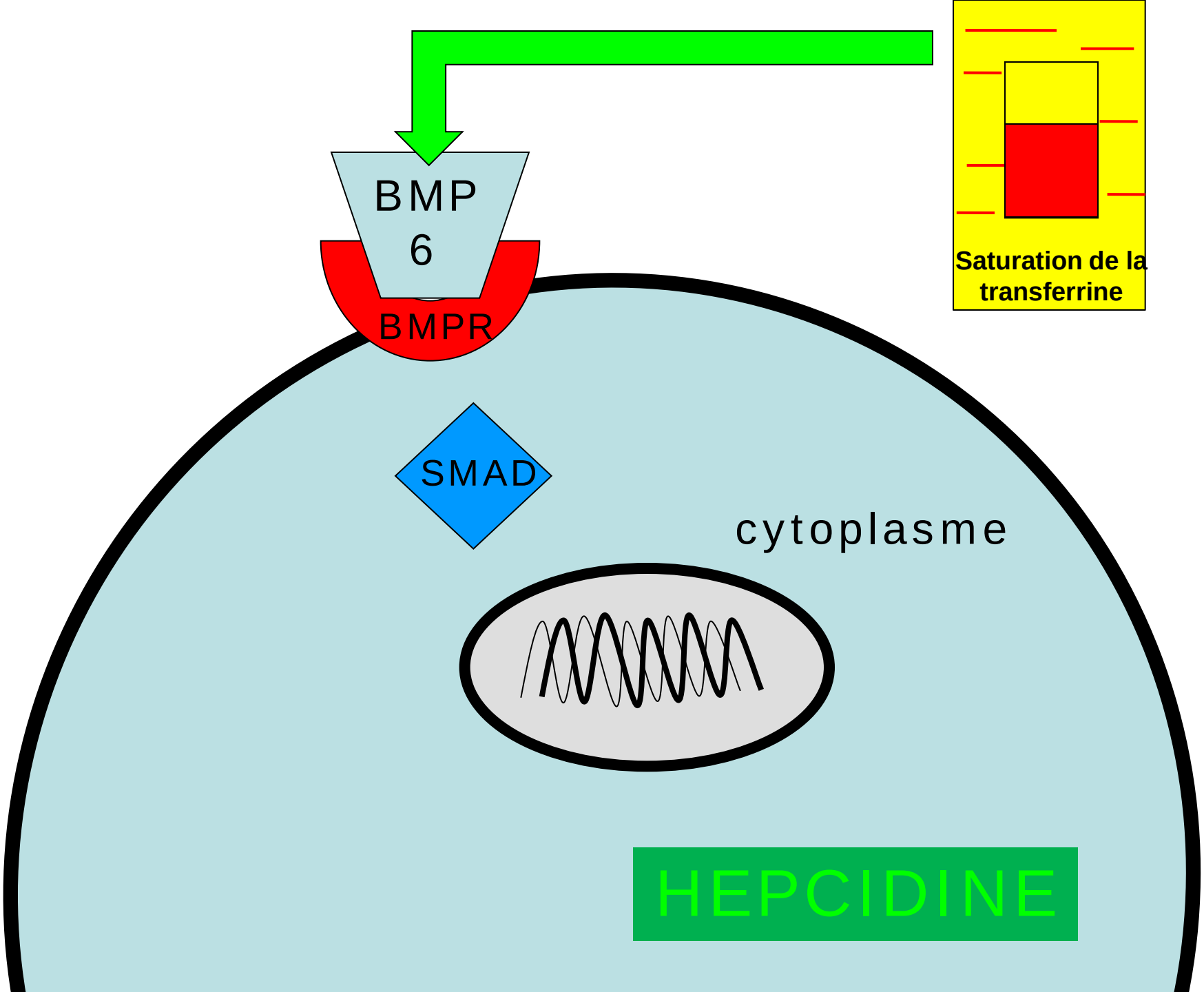
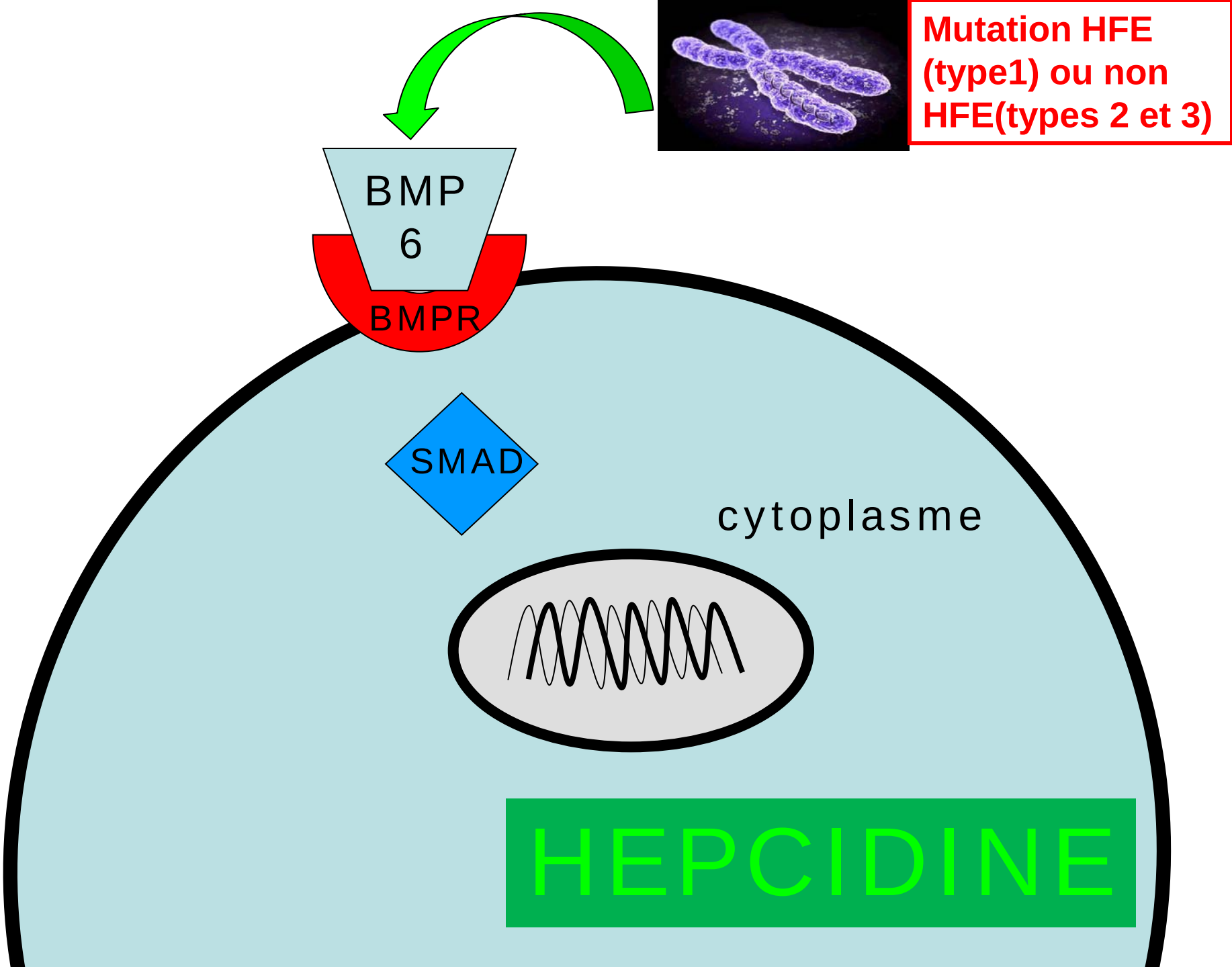
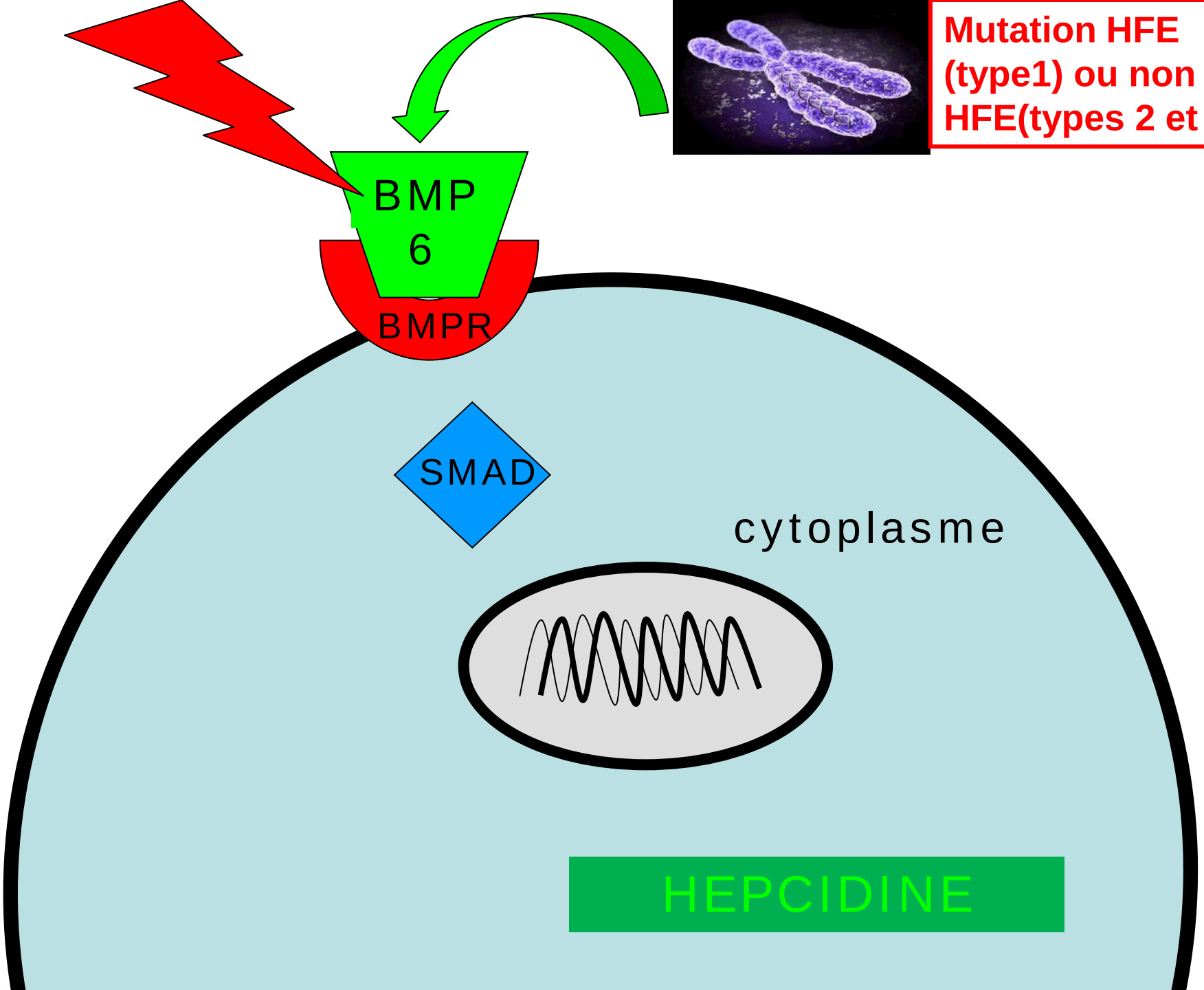

Mutation HFE (type1) ou non HFE (types 2 et 3)
HEPCIDINE

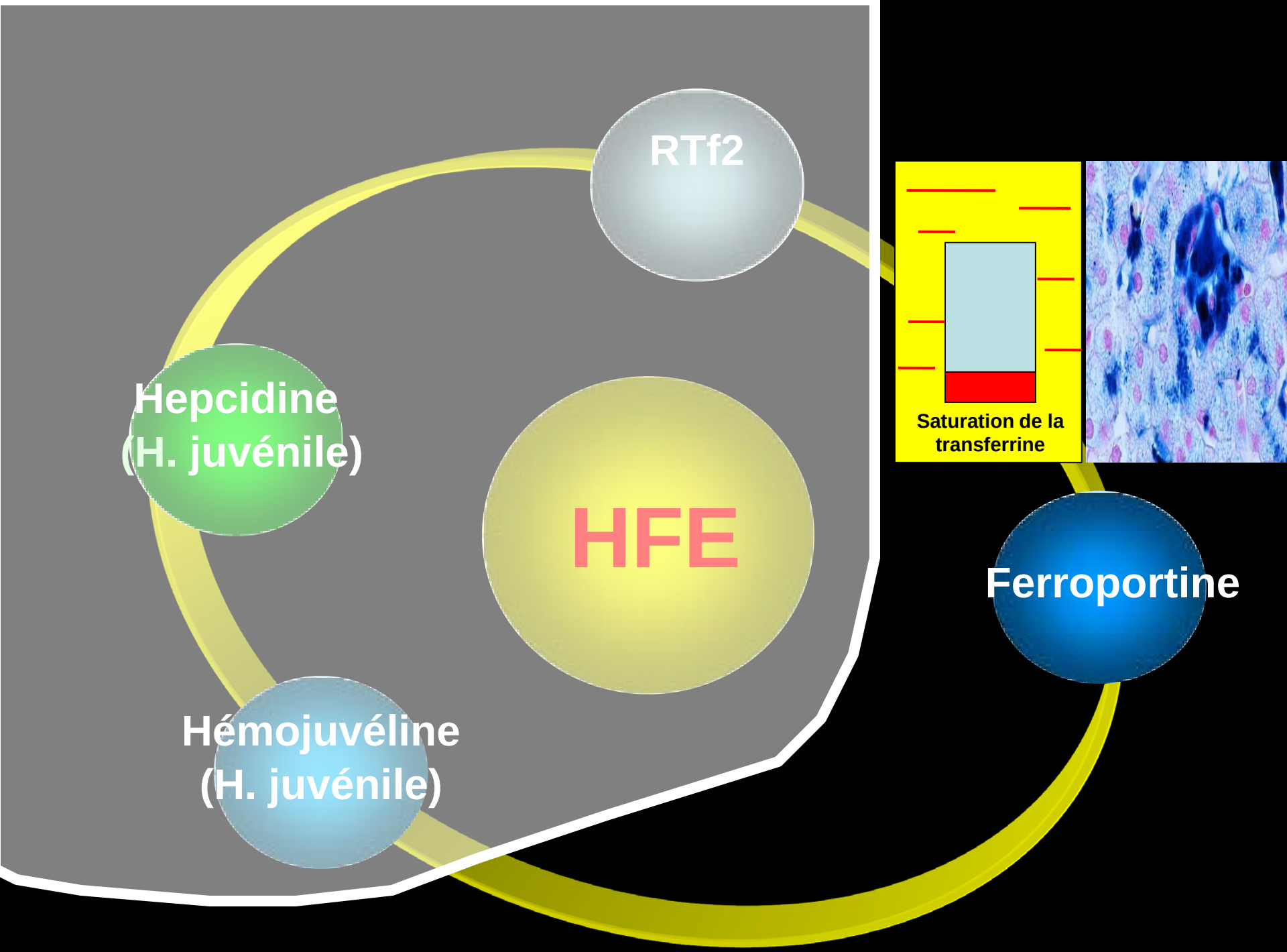

Saturation de la transferrine

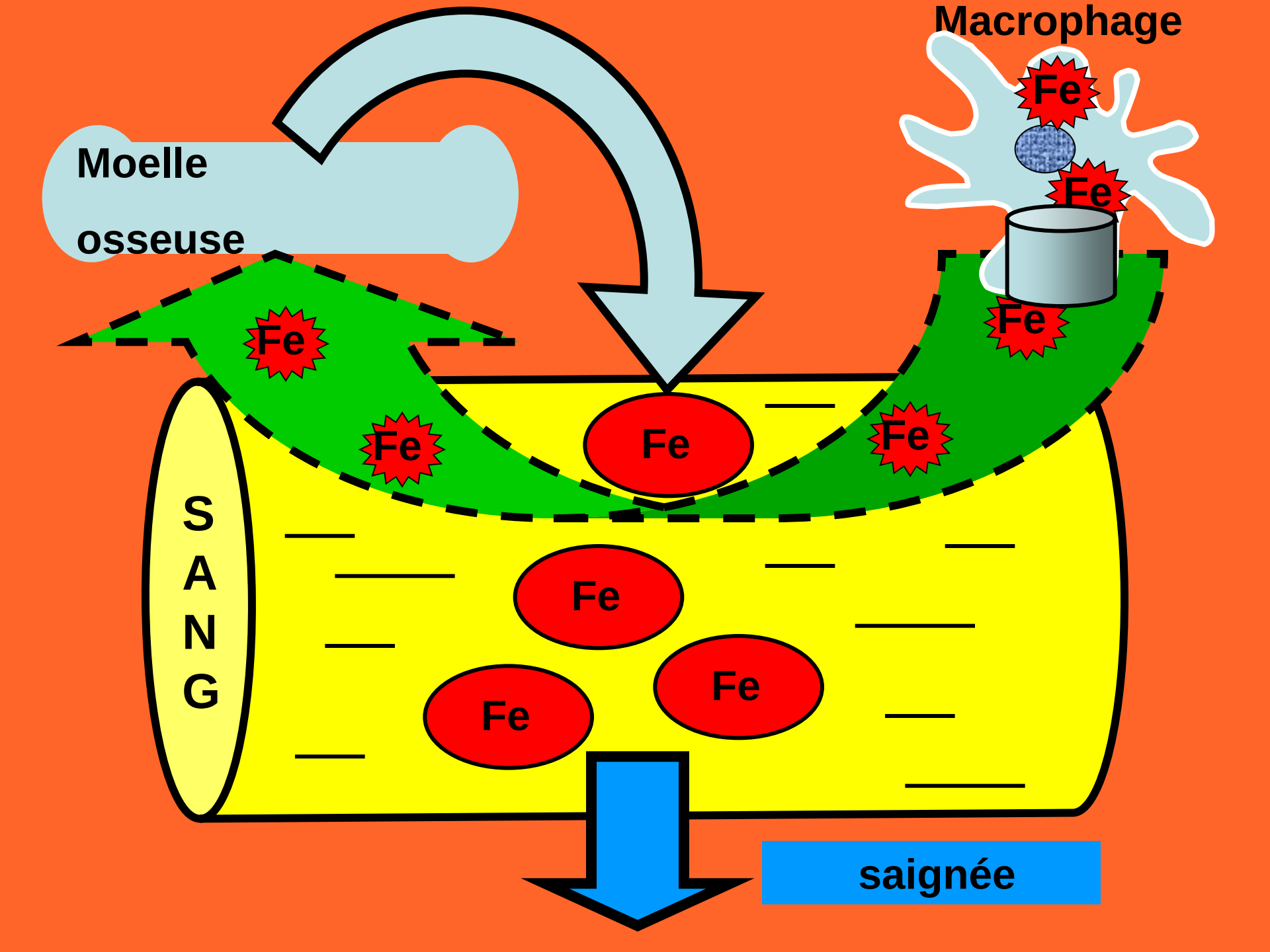

cytoplasme


BMP
6
BMPR
SMAD
cytoplasme
HEPCIDINE
Saturation de la
transferrine

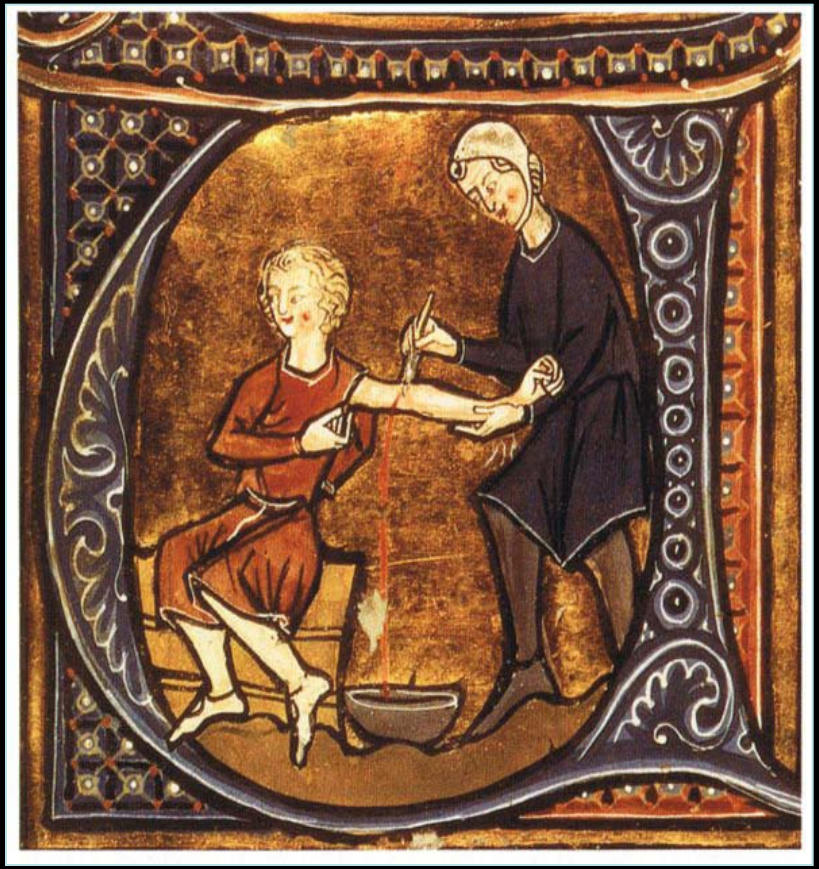

Mutation HFE (type1) ou non HFE(types 2 et 3)
BMP 6
BMPR
SMAD
cytoplasme
HEPCIDINE

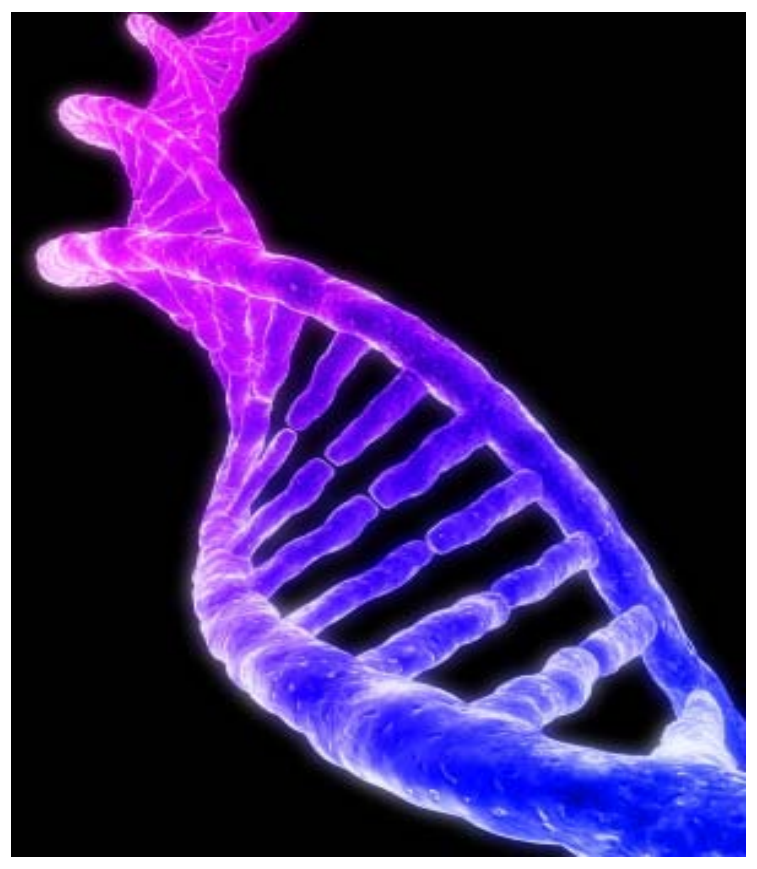

Mutation HFE (type1) ou non HFE(types 2 et 3)
BMP 6
BMPR
SMAD
cytoplasme
HEPCIDINE


RTf2
Hepcidine
(H. juvénile)
HFE
Hémojuvéline
(H. juvénile)
Saturation de la
transferrine
Ferroportine


Macrophage
Fe
Fe
Moelle
osseuse
Fe
Fe
Fe
Fe
Fe
Fe
Fe
S
A
N
G
saignée

Déférasirox
CICL670A0107
ICL670 500mg
FC05 0902
08/2003





Les Points Forts

1 -Surcharge en fer chronique génétique, le plus souvent par hepcidinodéficience

2 - Prise en charge Hémochromatose HFE (= Type1) : www.has-sante.fr

3 - Saignées : option thérapeutique principale

4 - Chélation orale (déférasirox) : en voie de positionnement

5- Recharge en hepcidine: l'avenir…