



**Service des Maladies du Foie
et Inserm U-991**

**Centre de Référence des Surcharges
en Fer Rares d'origine Génétique**

CHU Pontchaillou

L'Hémochromatose C282Y en 2014

Pierre Brissot

RENNES



Hémochromatose liée au gène HFE

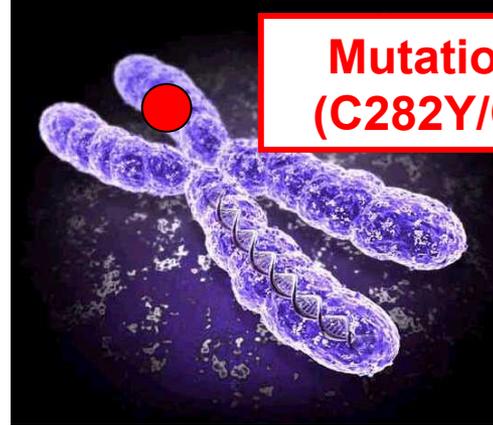
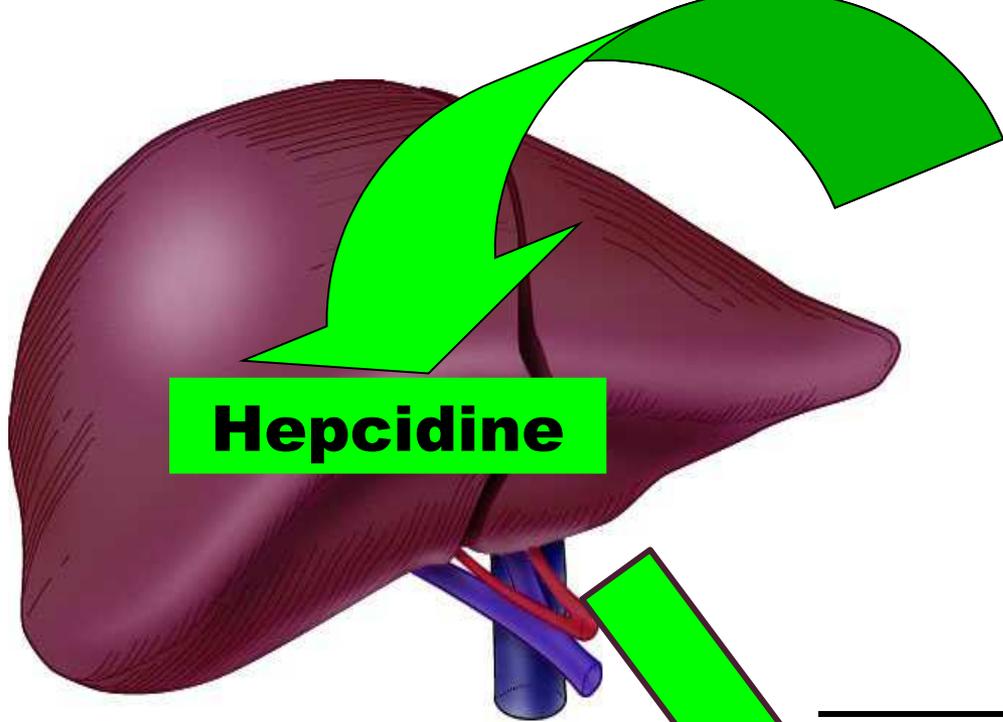
Hémochromatose de type 1

L'évolution des idées et des pratiques

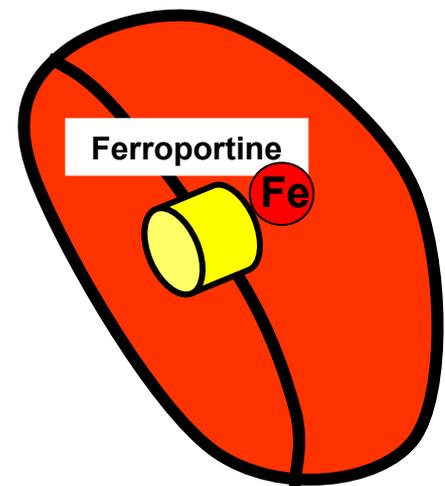
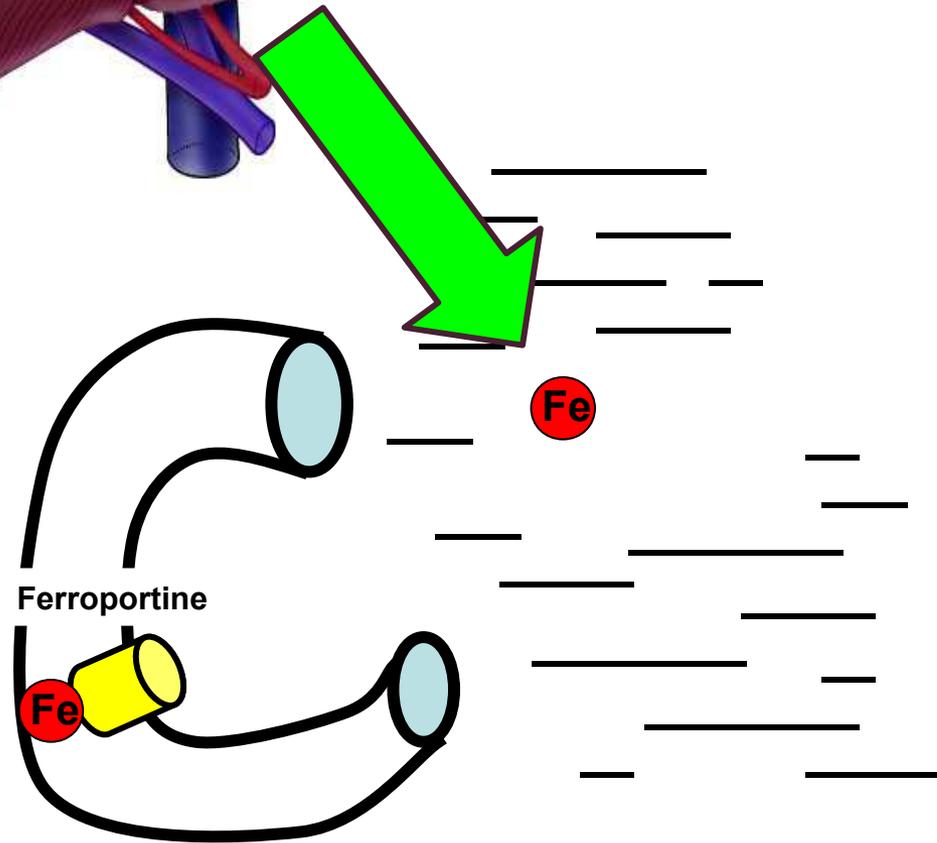
Physio-pathologie

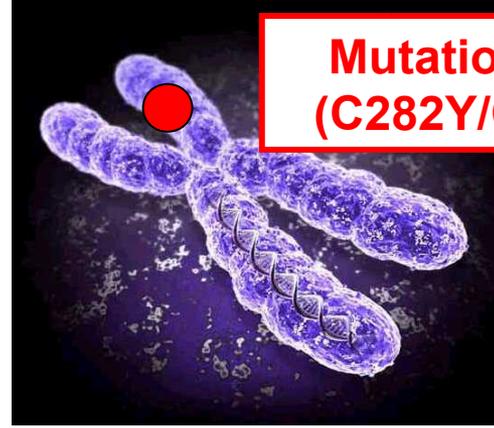
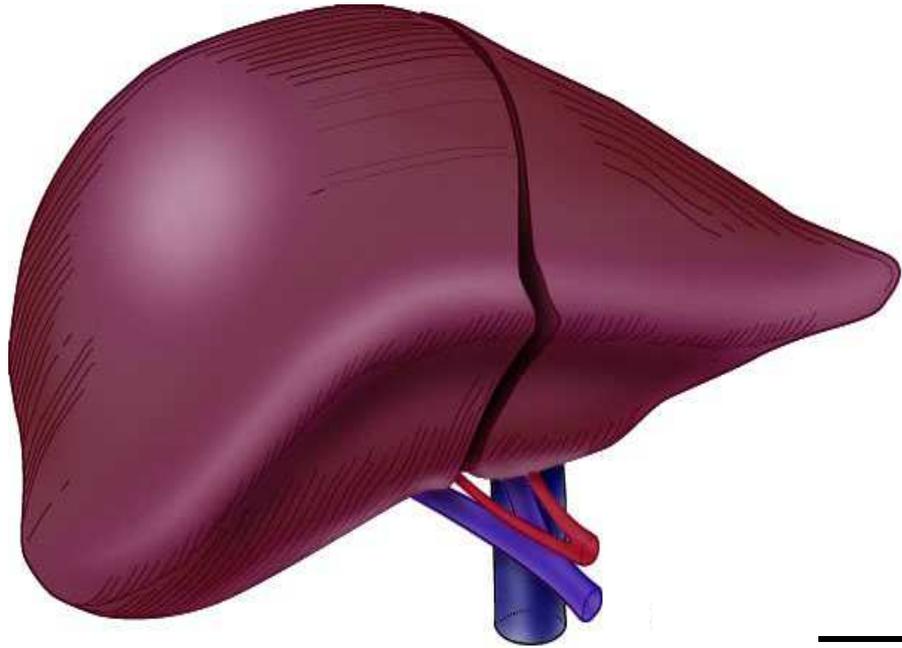
Diagnostic

Traitement

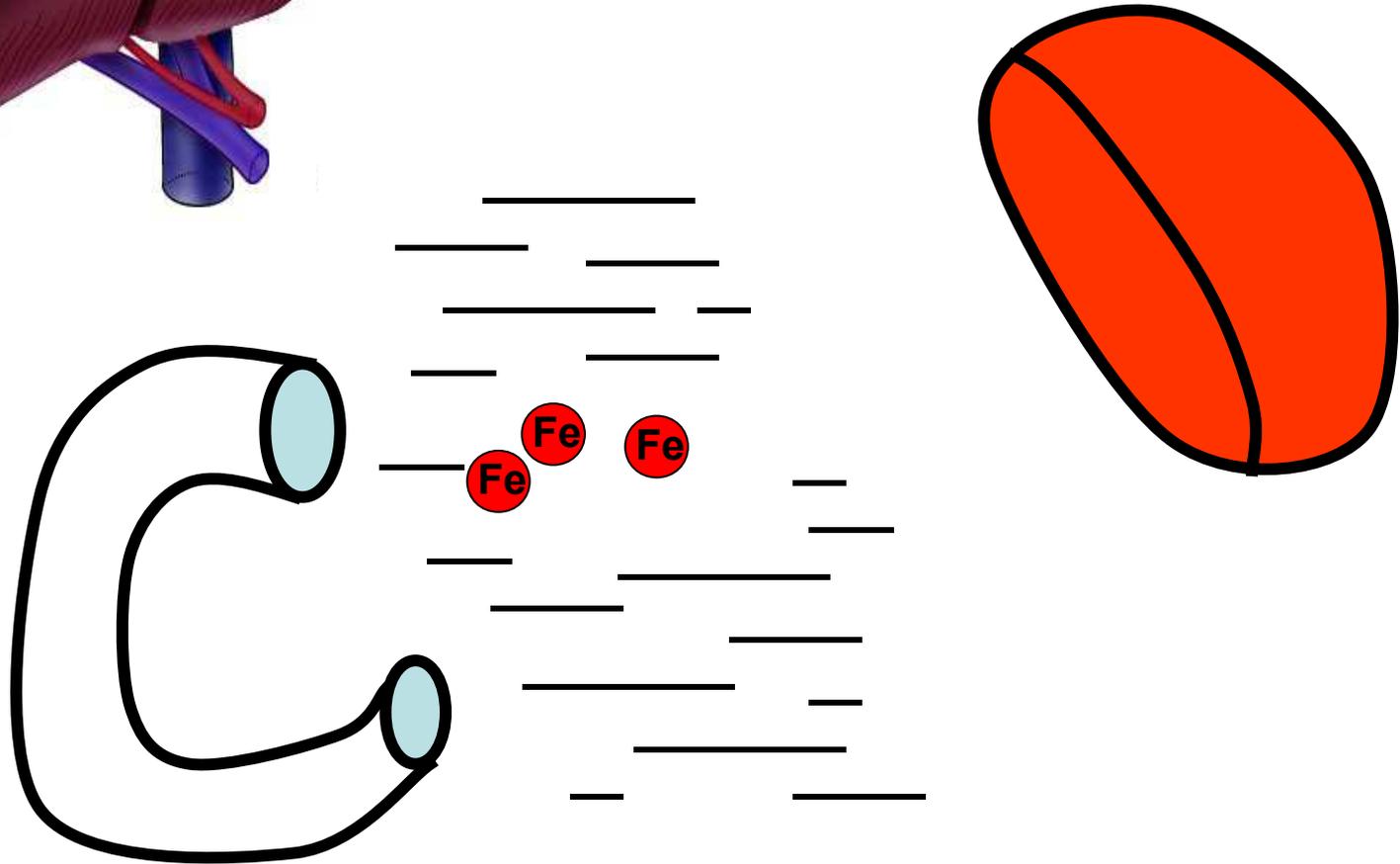


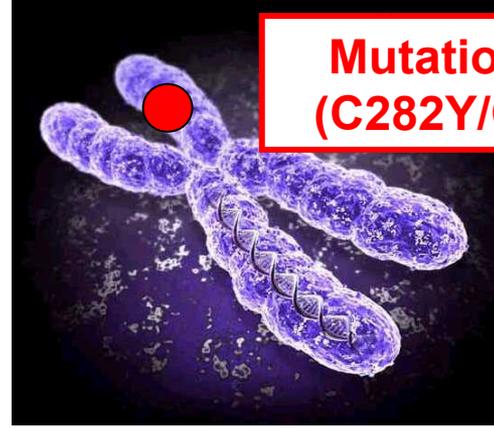
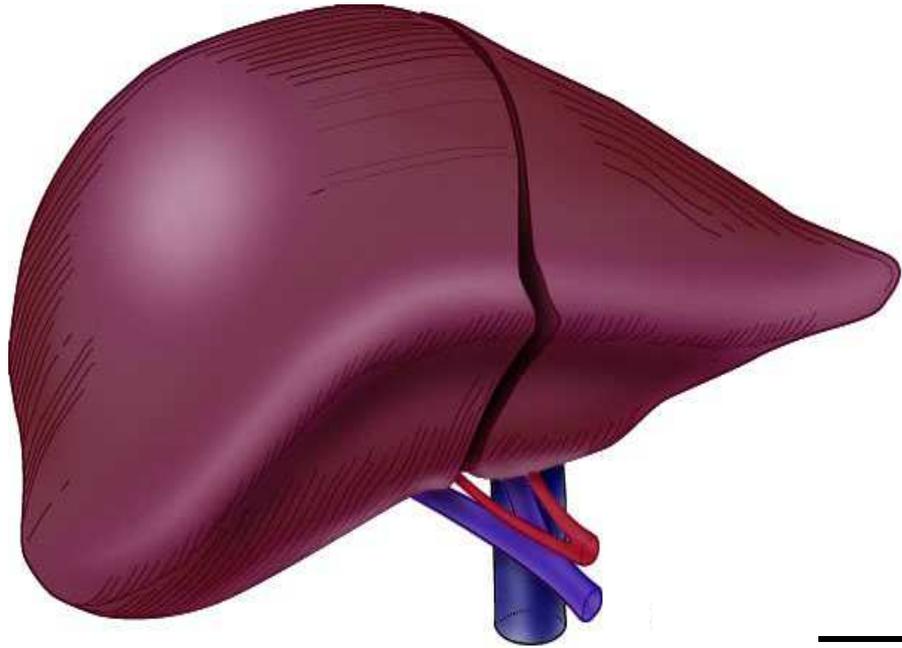
**Mutation HFE
(C282Y/C282Y)**



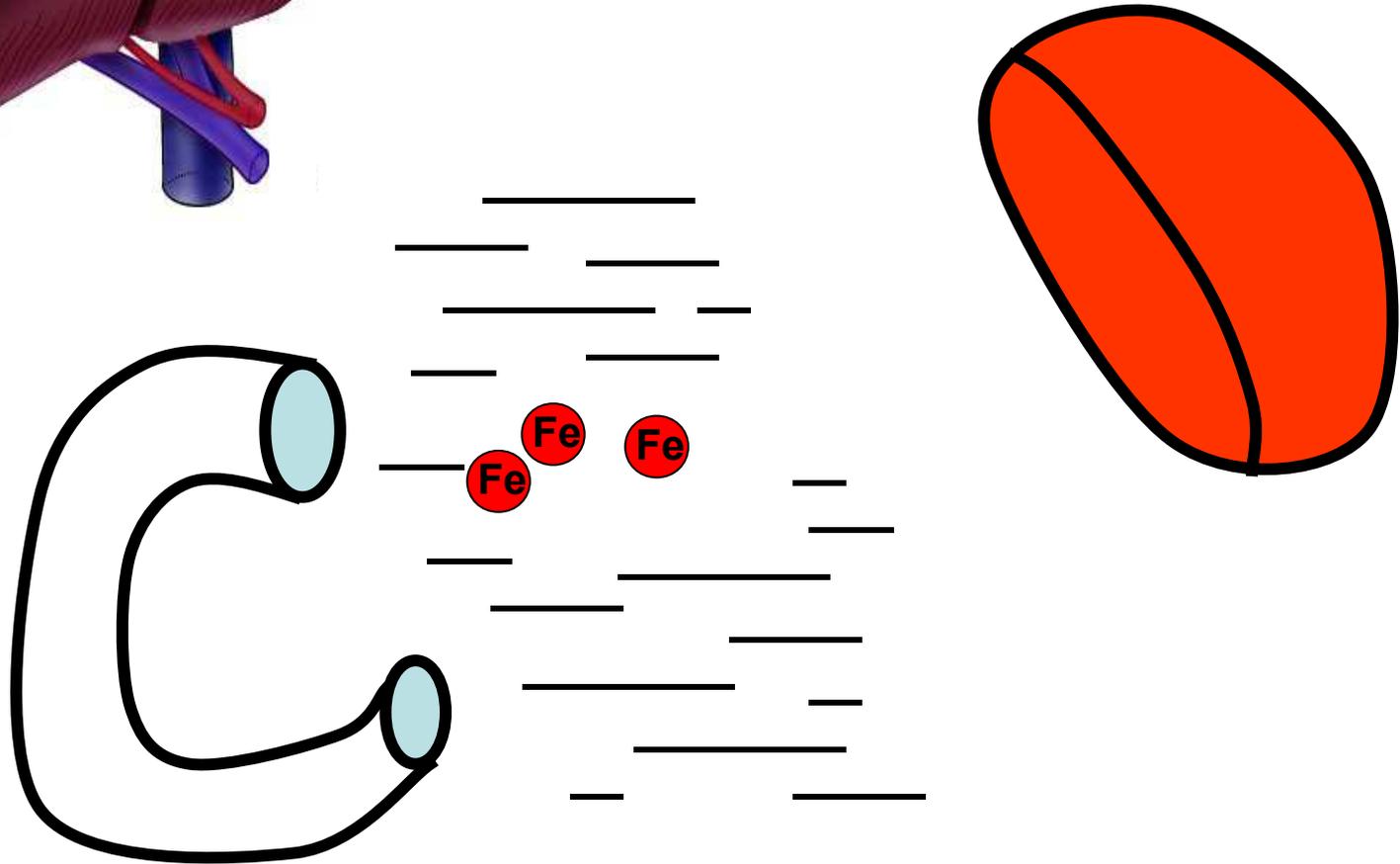


**Mutation HFE
(C282Y/C282Y)**





**Mutation HFE
(C282Y/C282Y)**

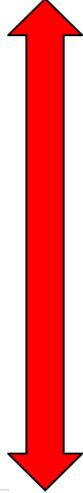






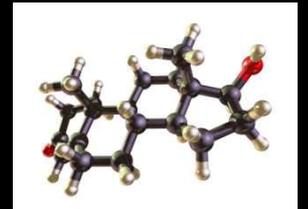
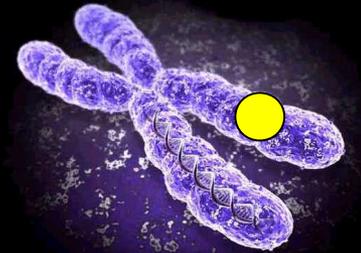
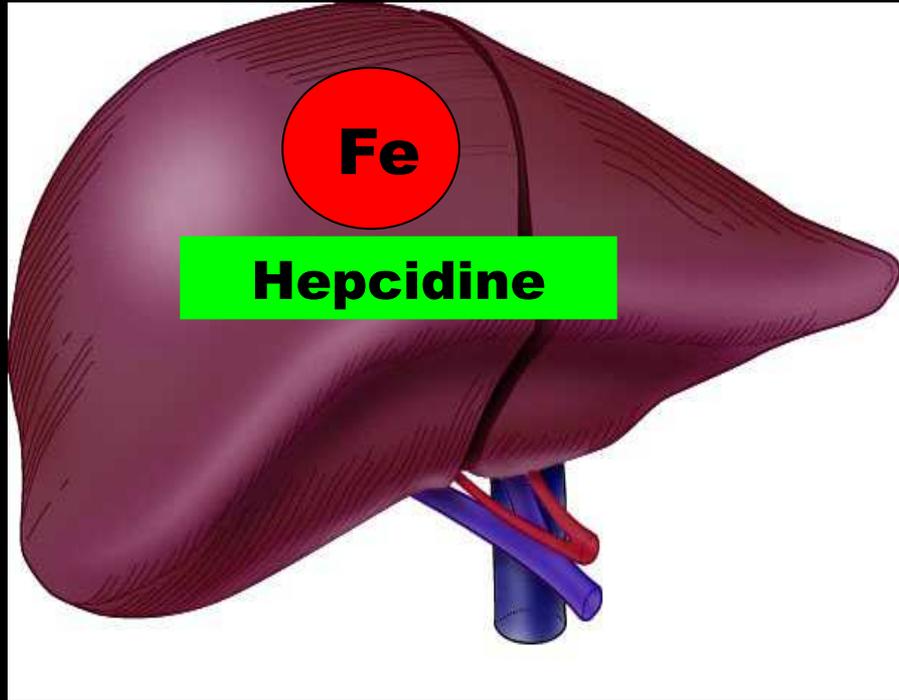
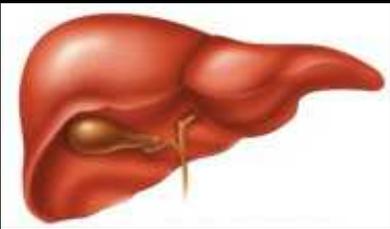
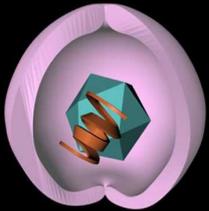
30% H

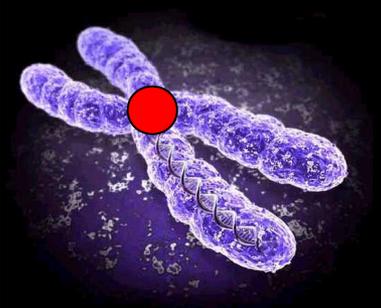
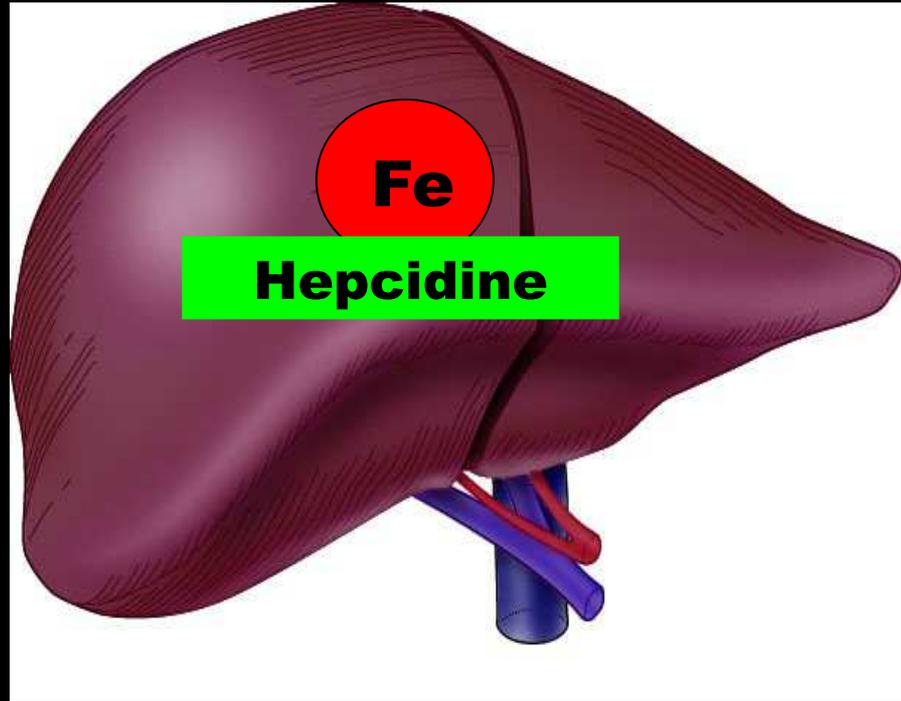
1% F

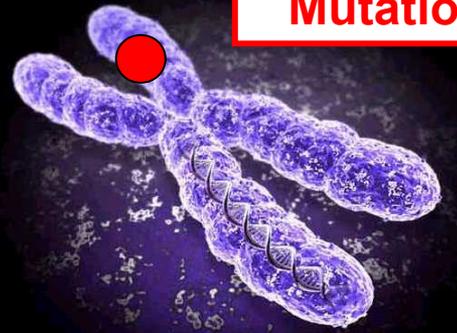
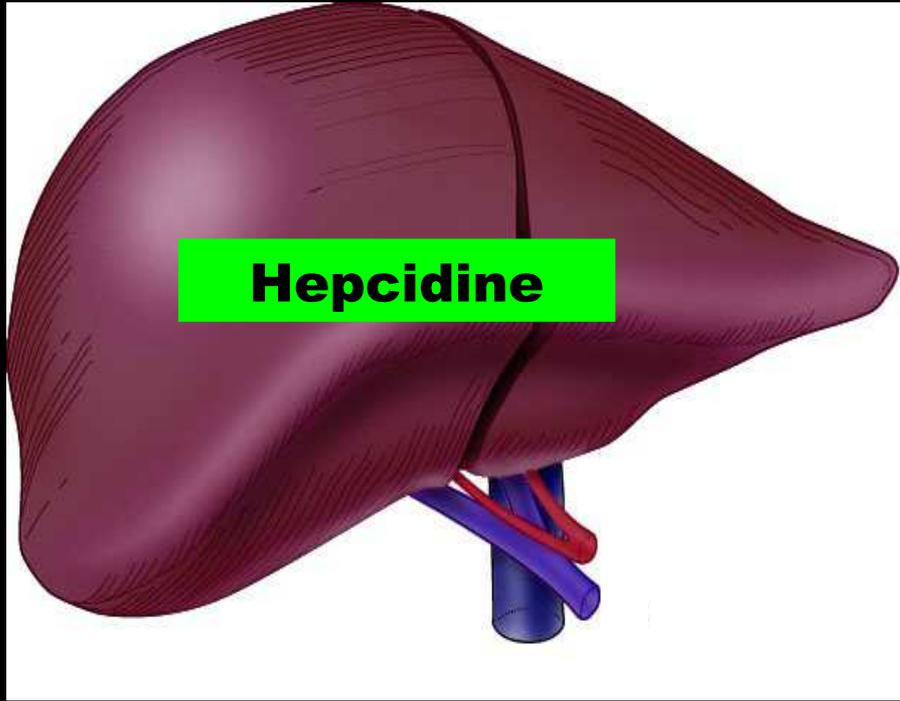


70% H

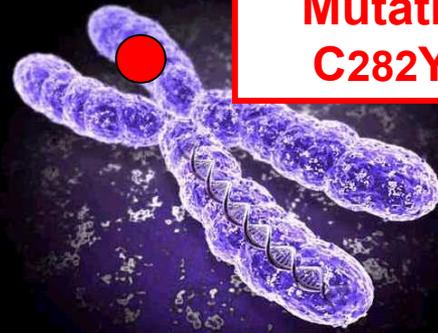
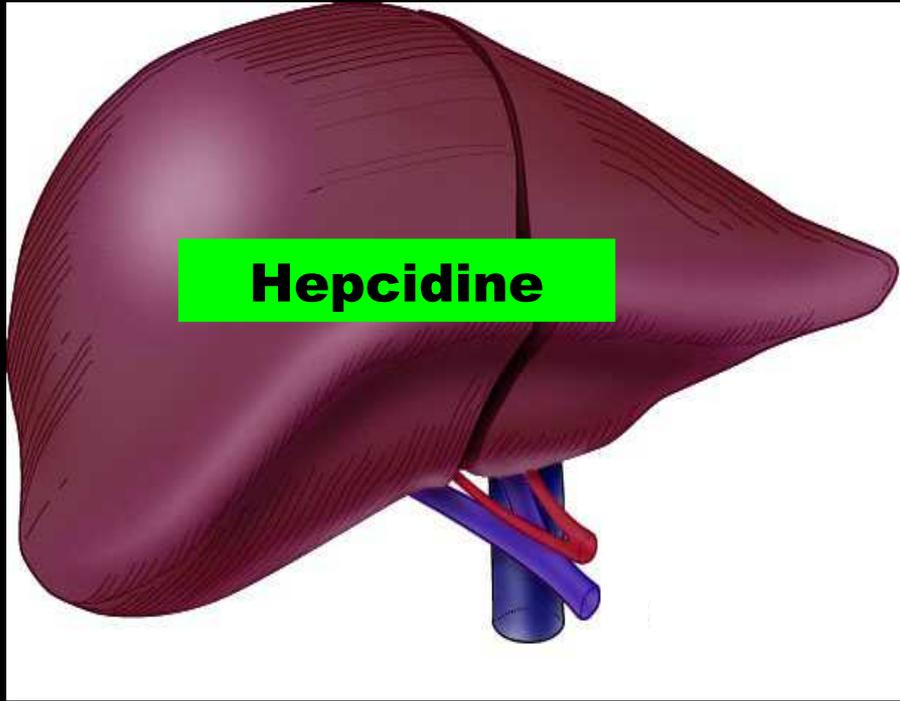
99% F



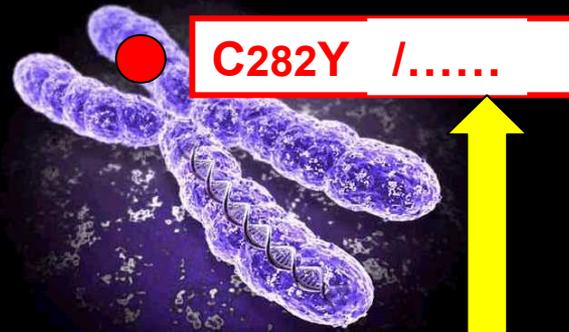
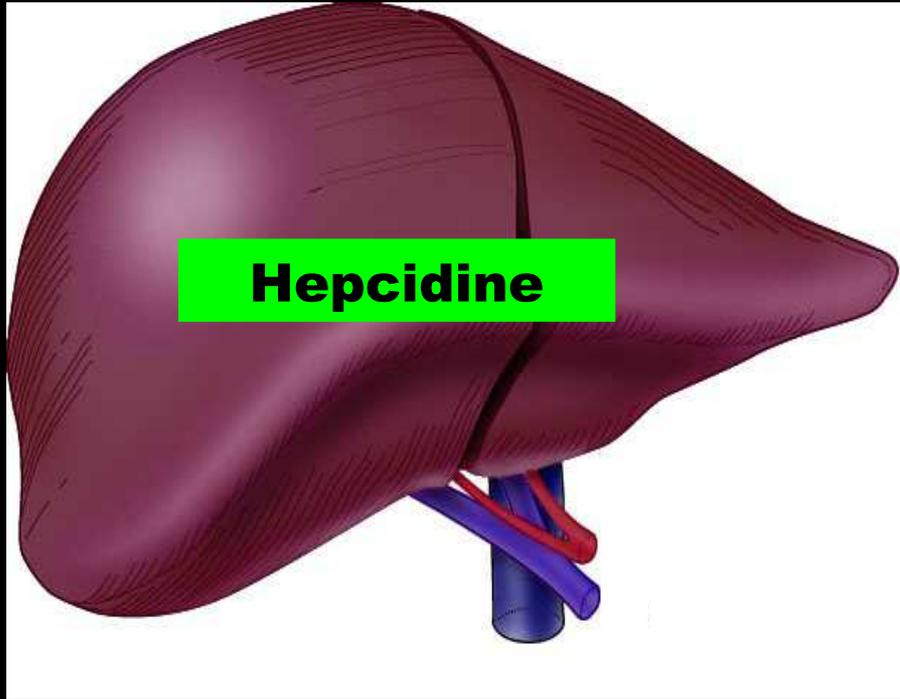




Mutations HFE

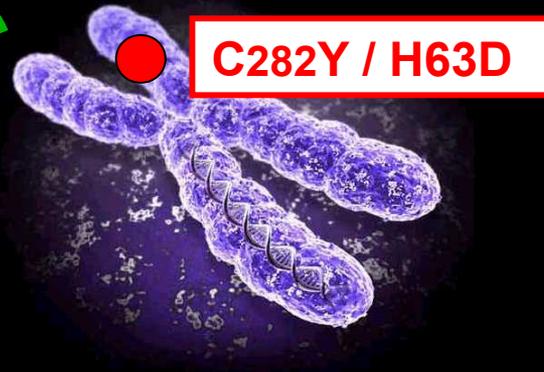
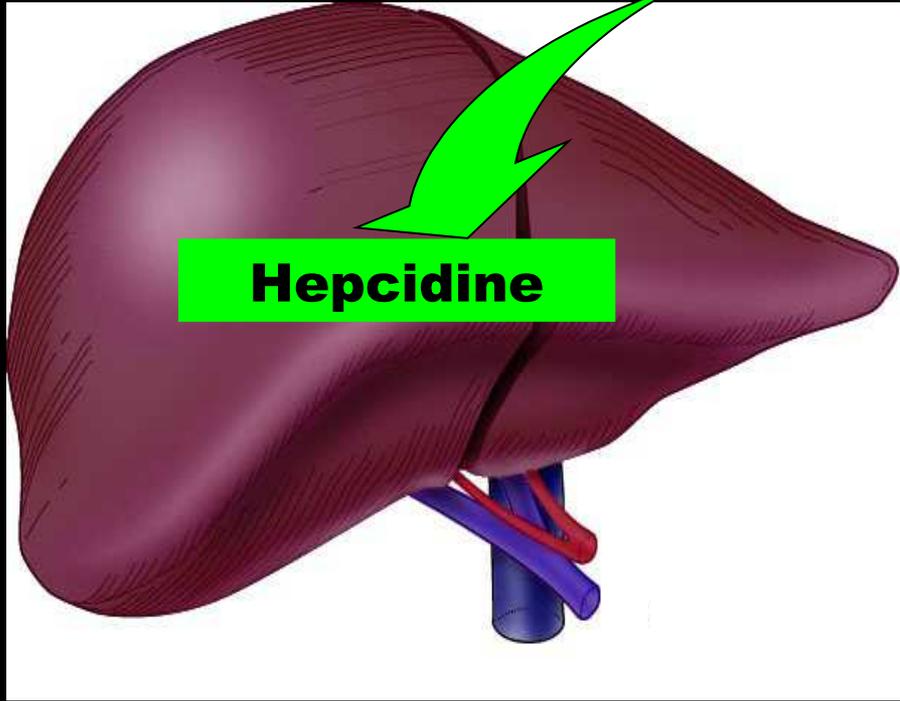


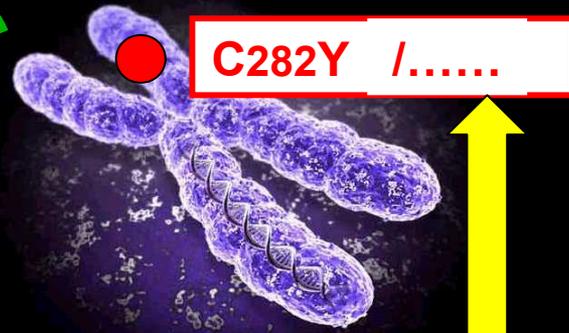
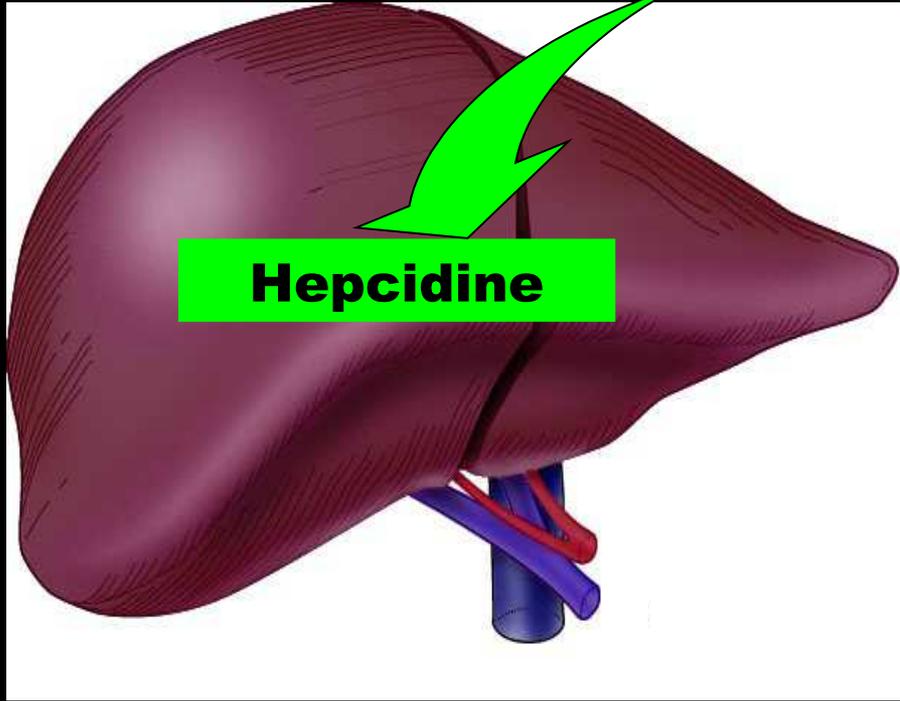
**Mutation HFE
C282Y/C282Y**



**Mutation
HFE
fréquente**

H63D



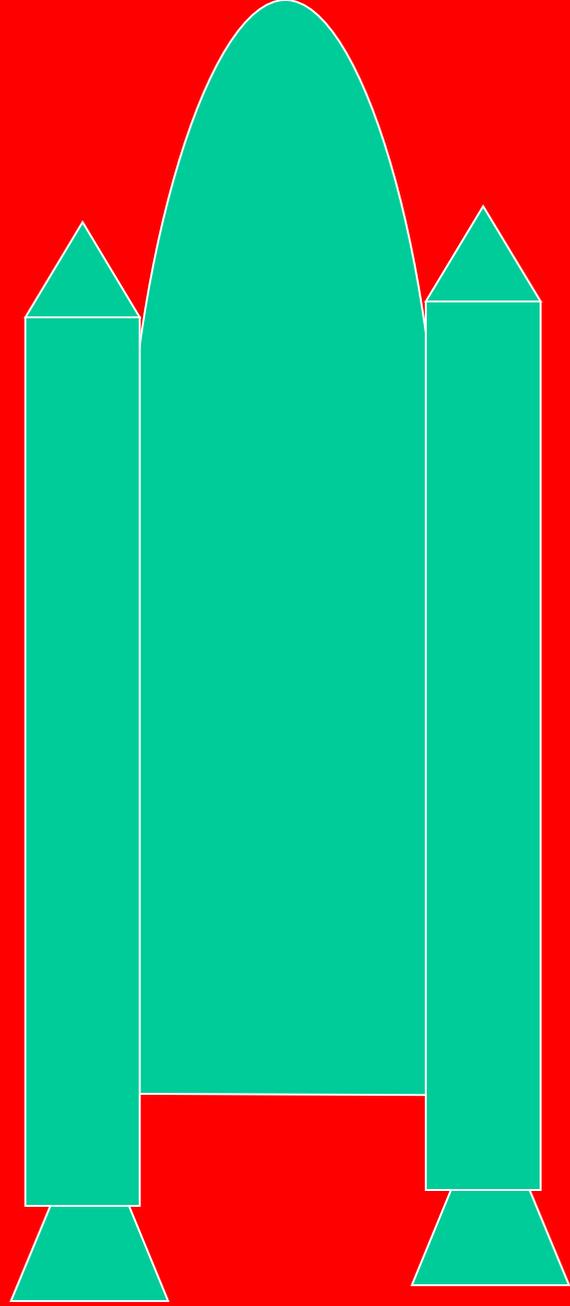


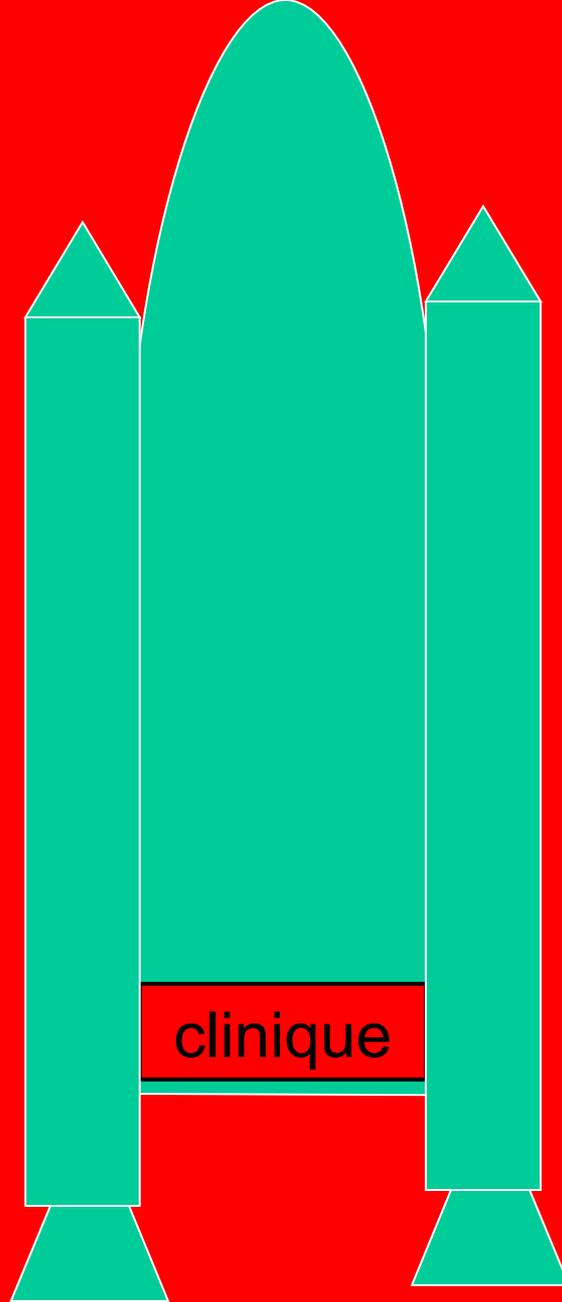
Mutation
HFE rare

Physio-pathologie

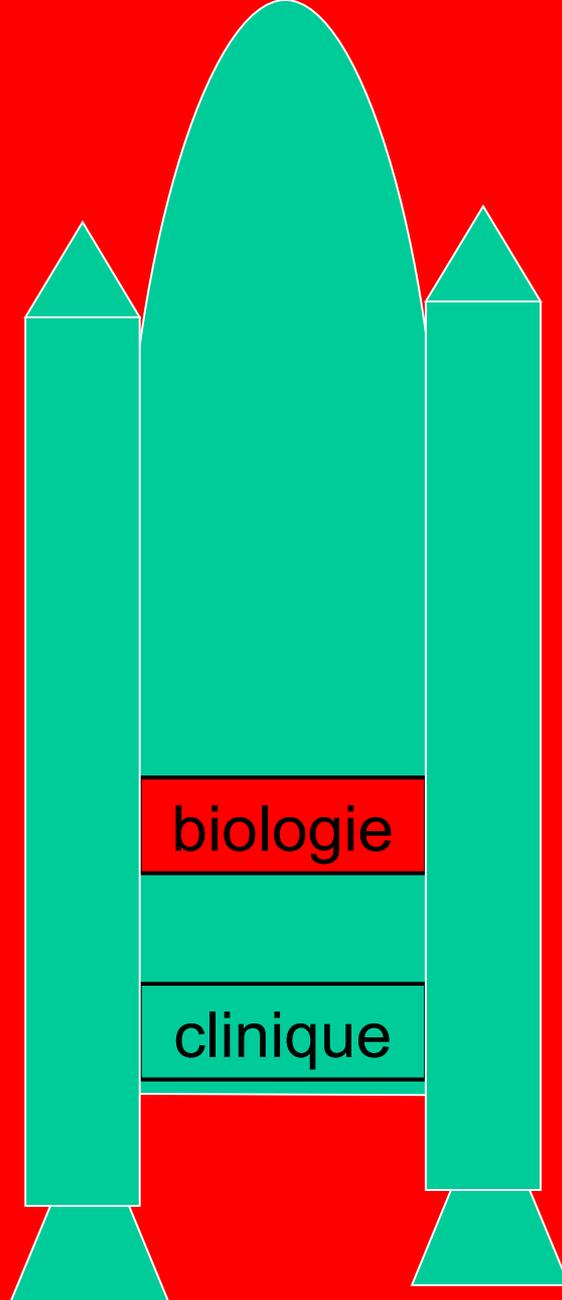
Diagnostic

Traitement



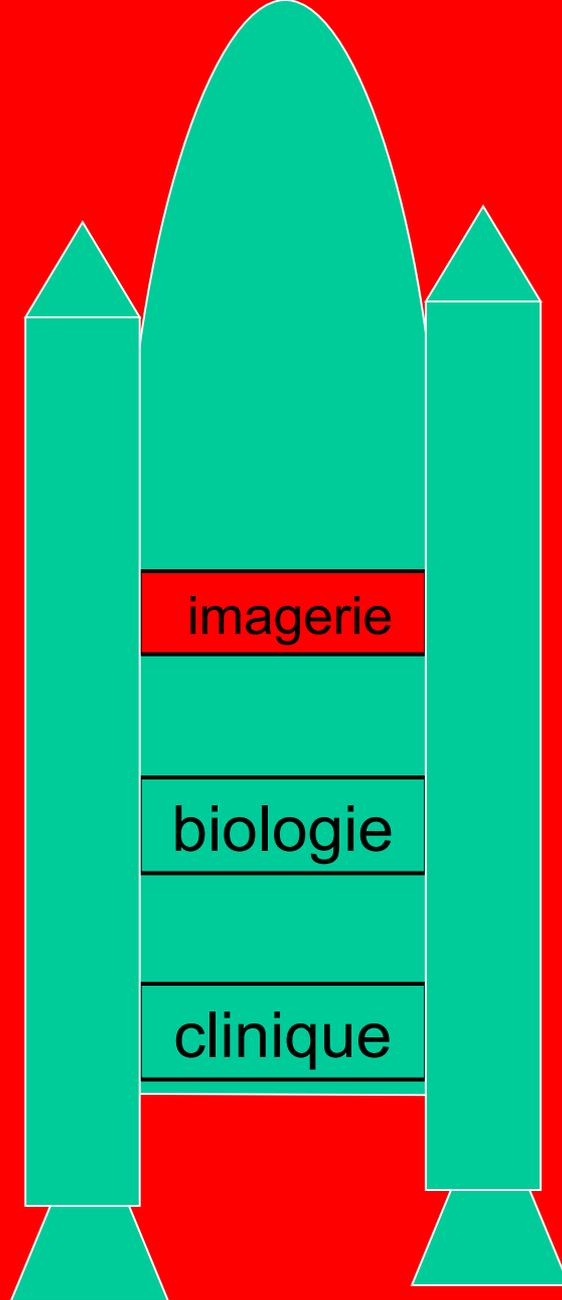


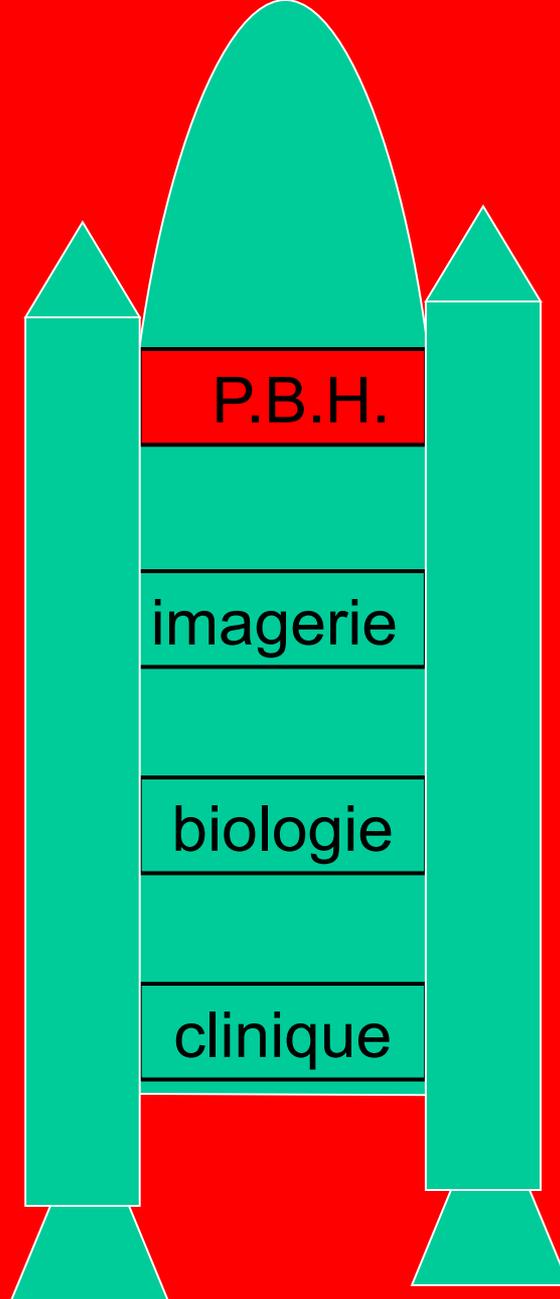
clinique

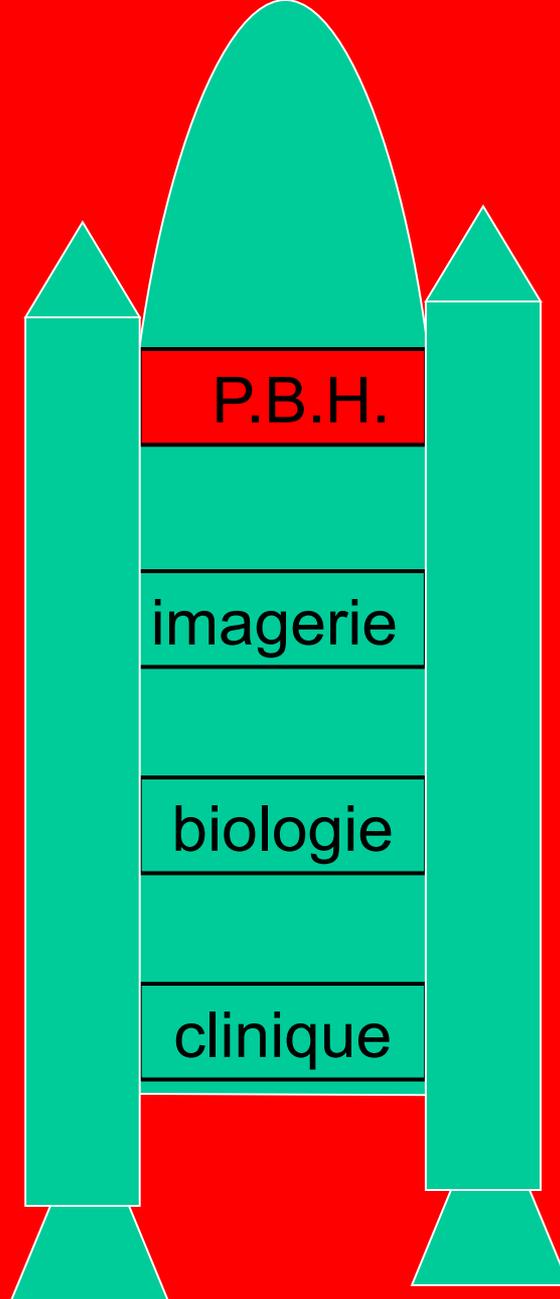


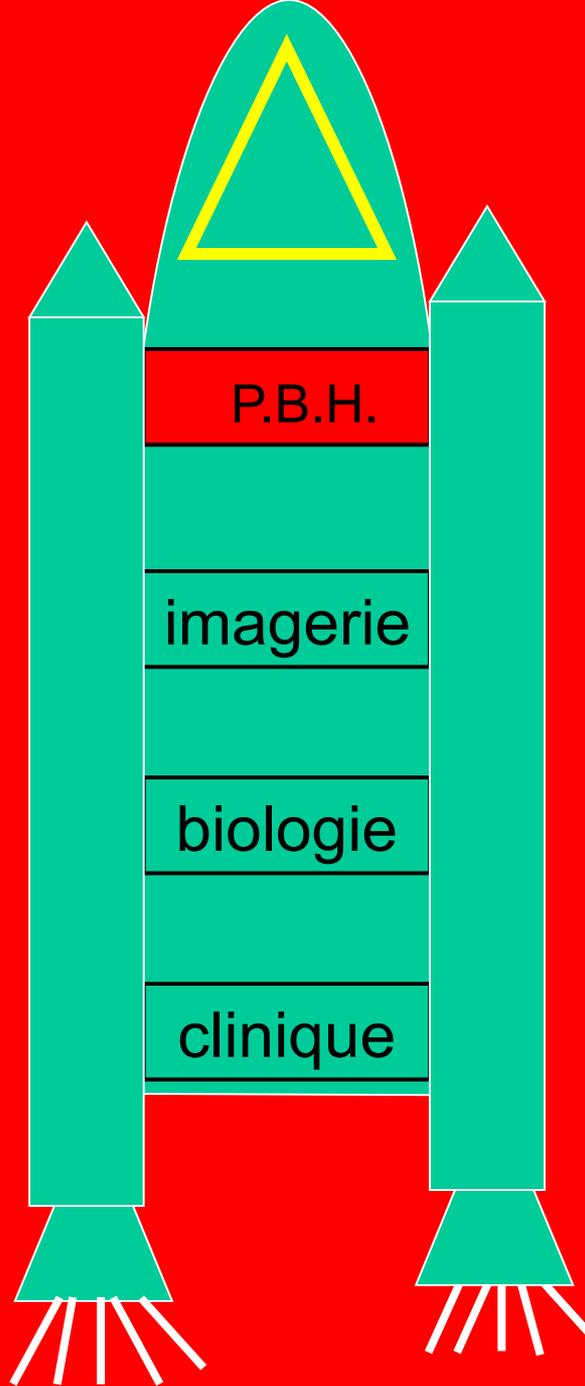
biologie

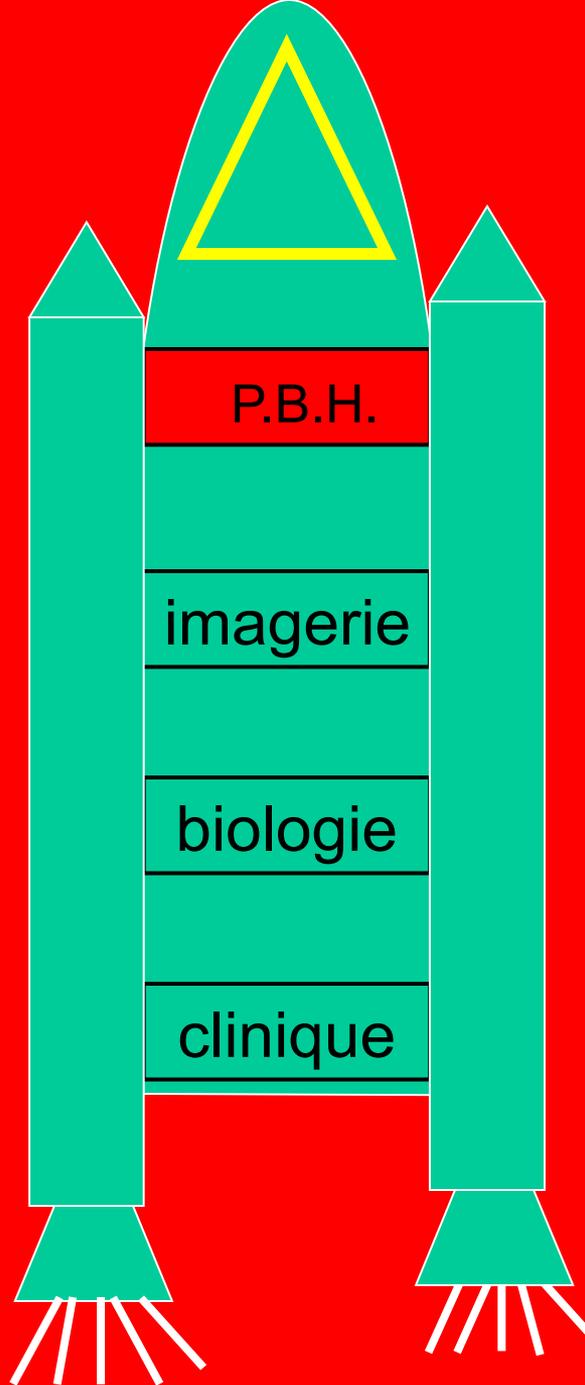
clinique

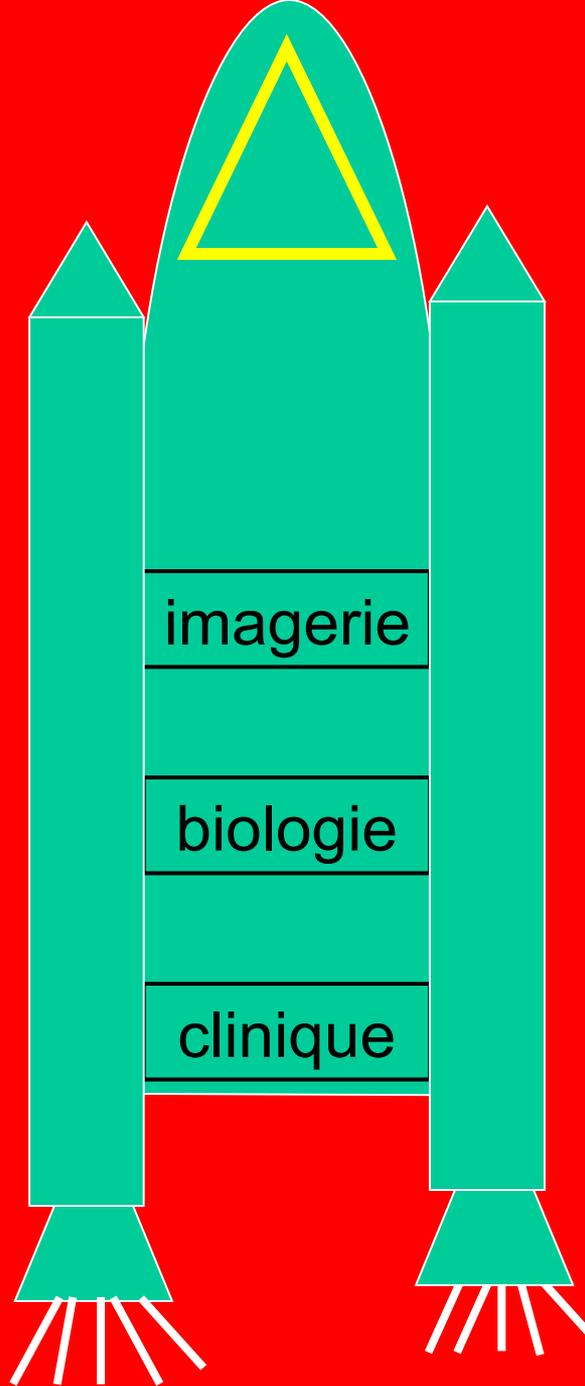


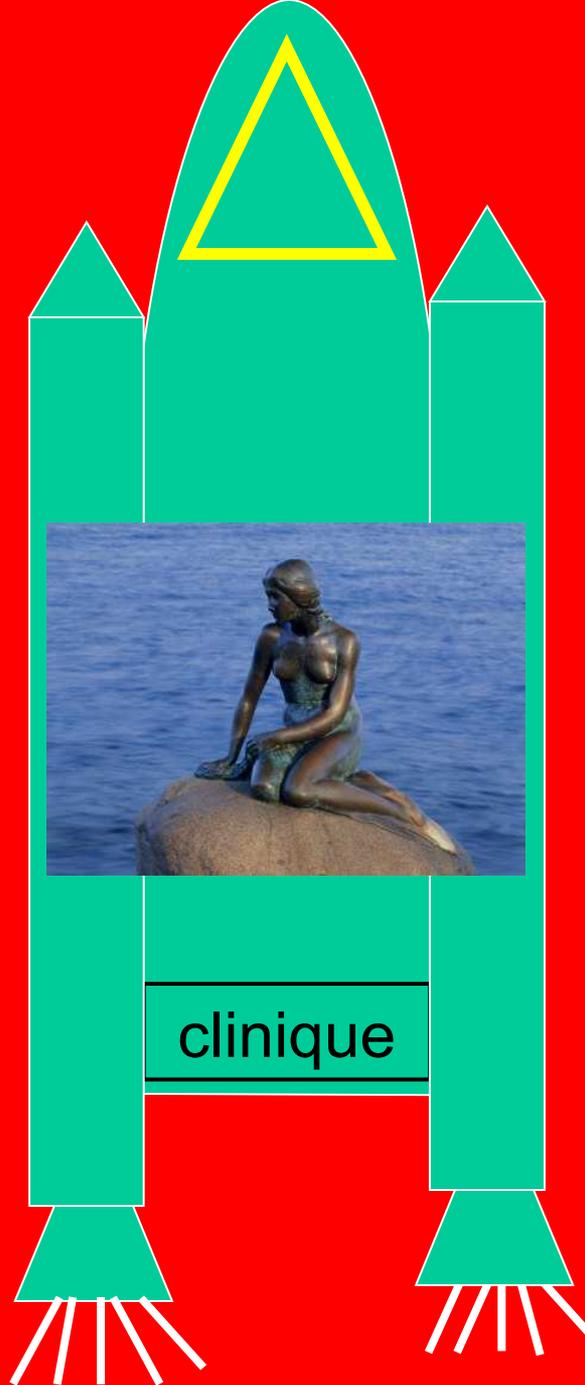




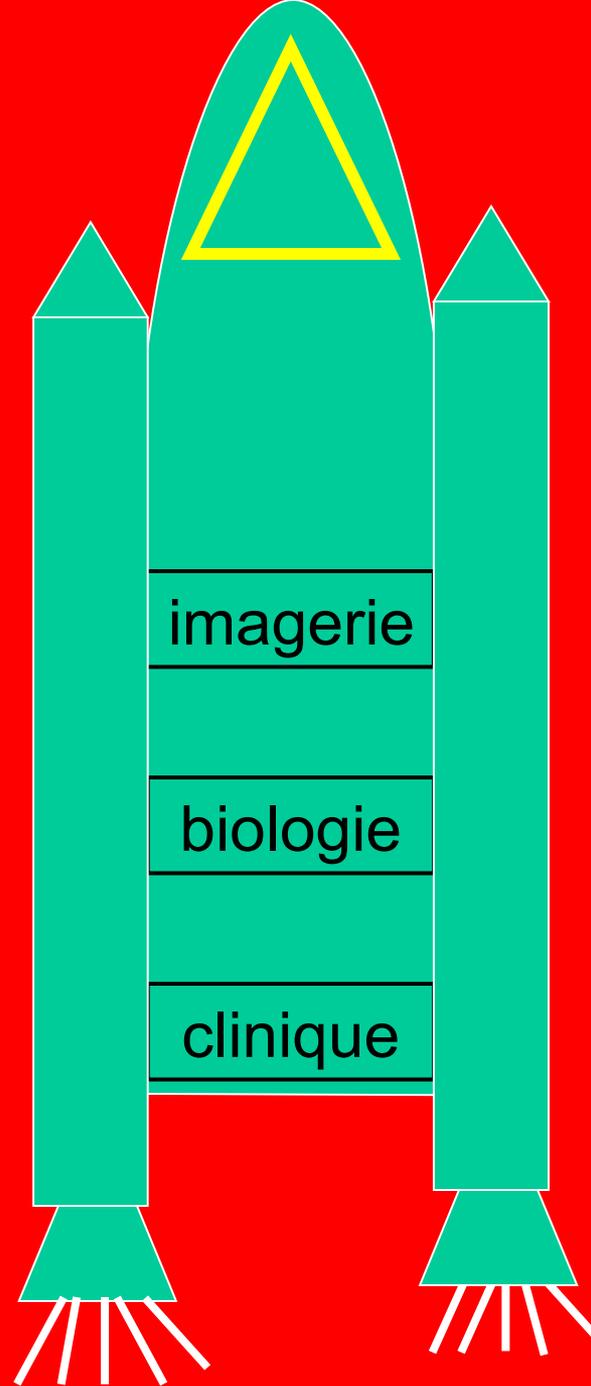








clinique

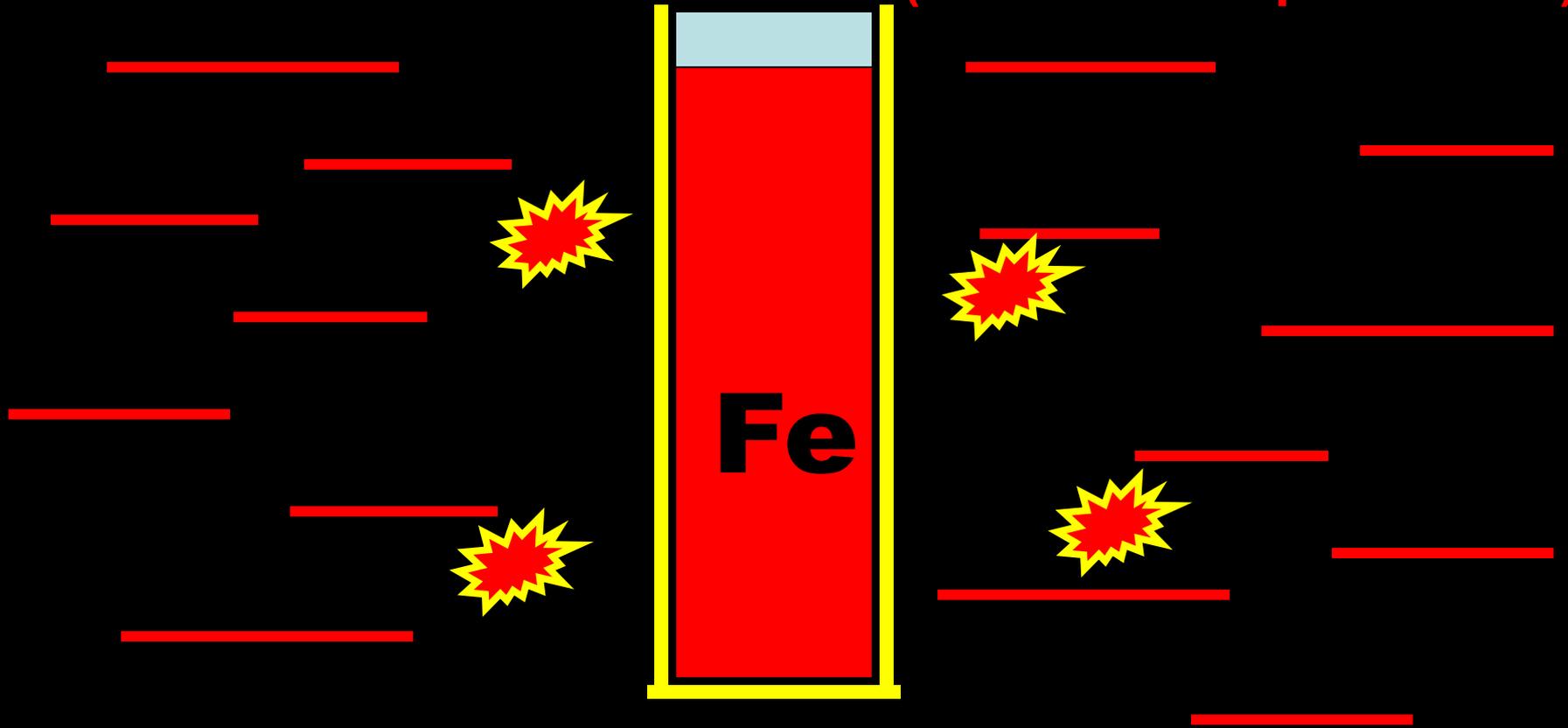


imagerie

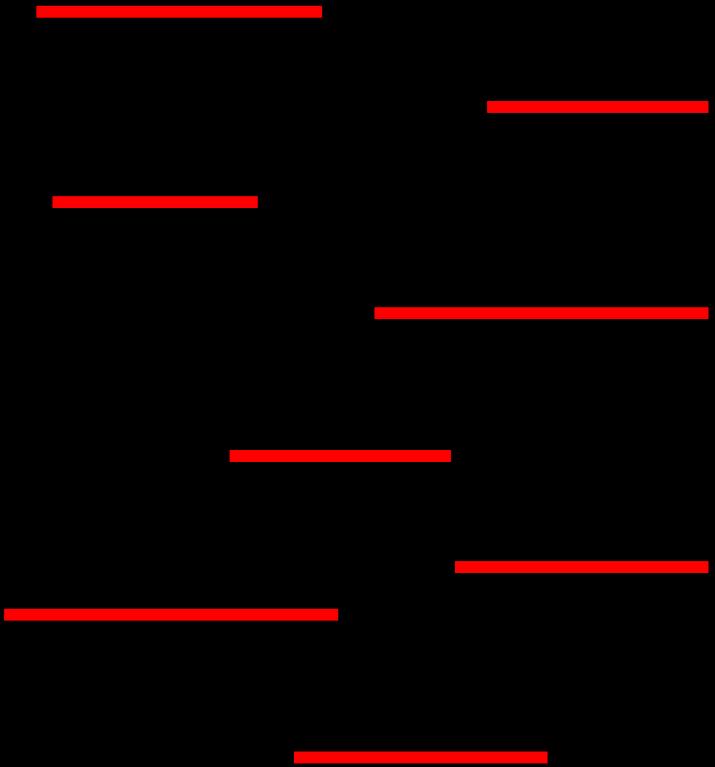
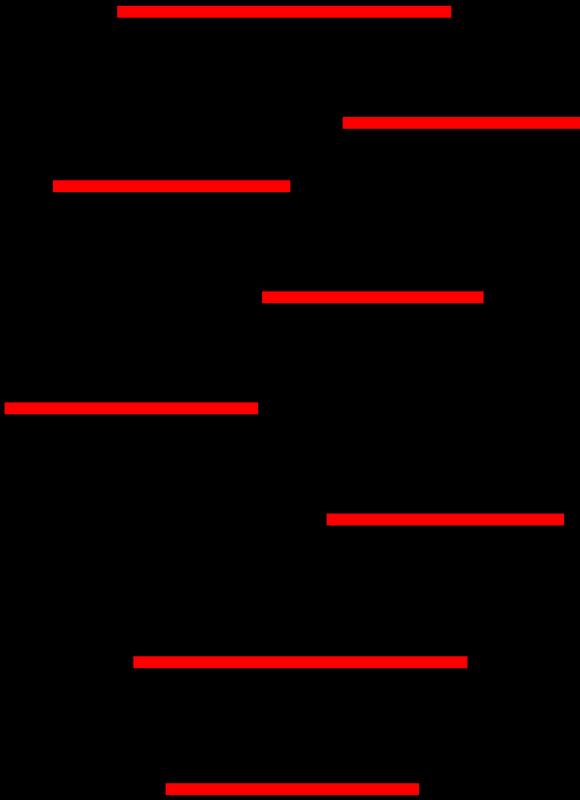
biologie

clinique

FPR (Fer Plasmatique Réactif)



Saturation Transferrine > 75%

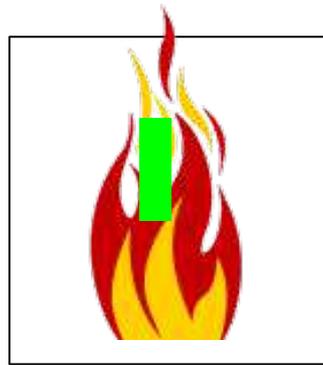
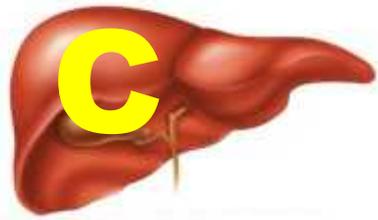




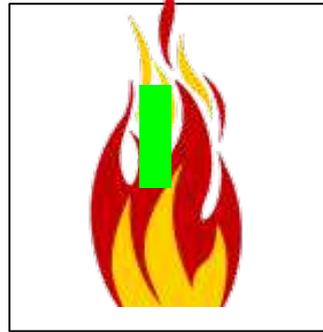
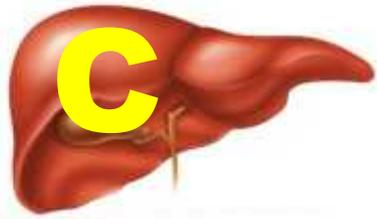
Ferritine



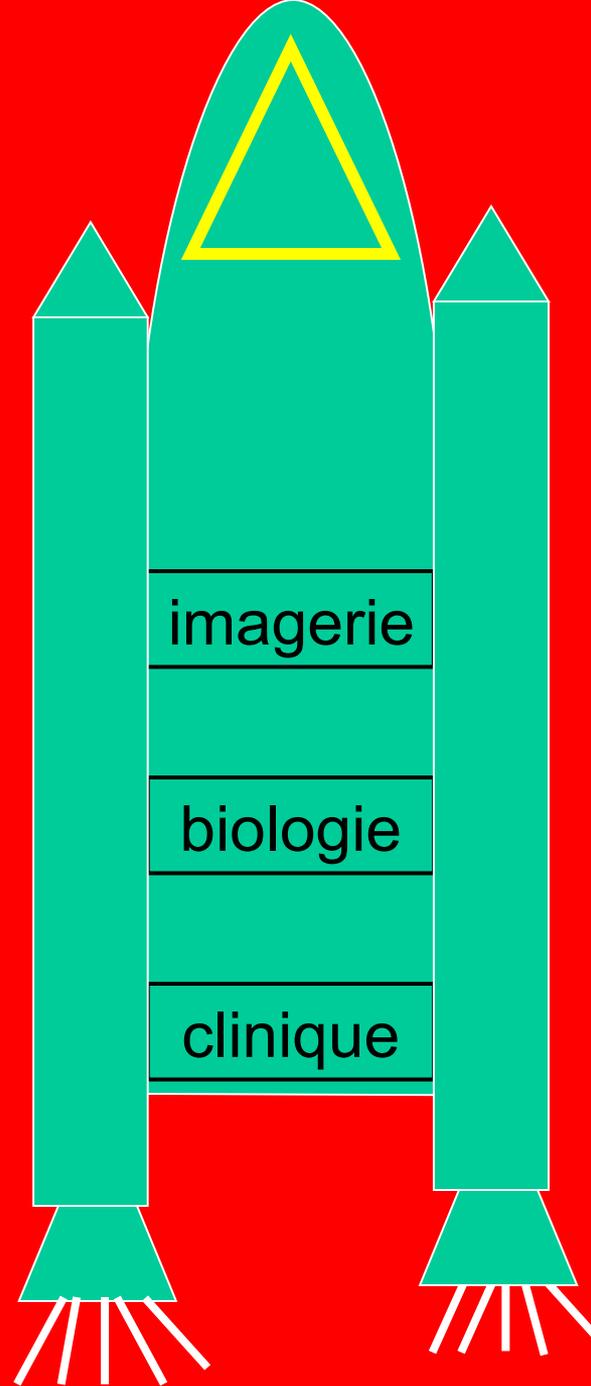
Ferritine



Ferritine



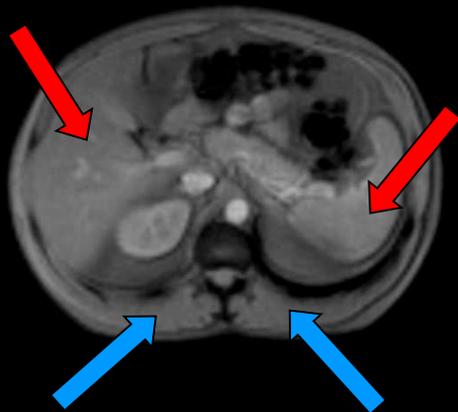
Ferritine



imagerie

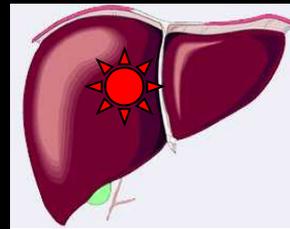
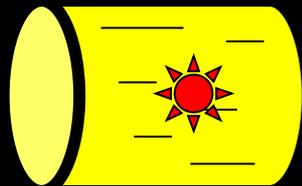
biologie

clinique



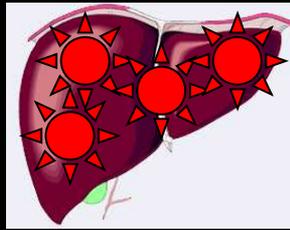
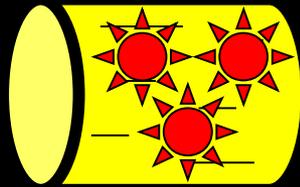
(www.radio.univ-rennes1.fr)

dosages
sanguins

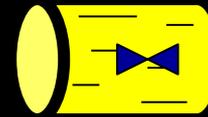


IRM





**Hémochromatose avec
déficience en
hepcidine**



**Hémochromatose HFE
(C282Y/C282Y)**

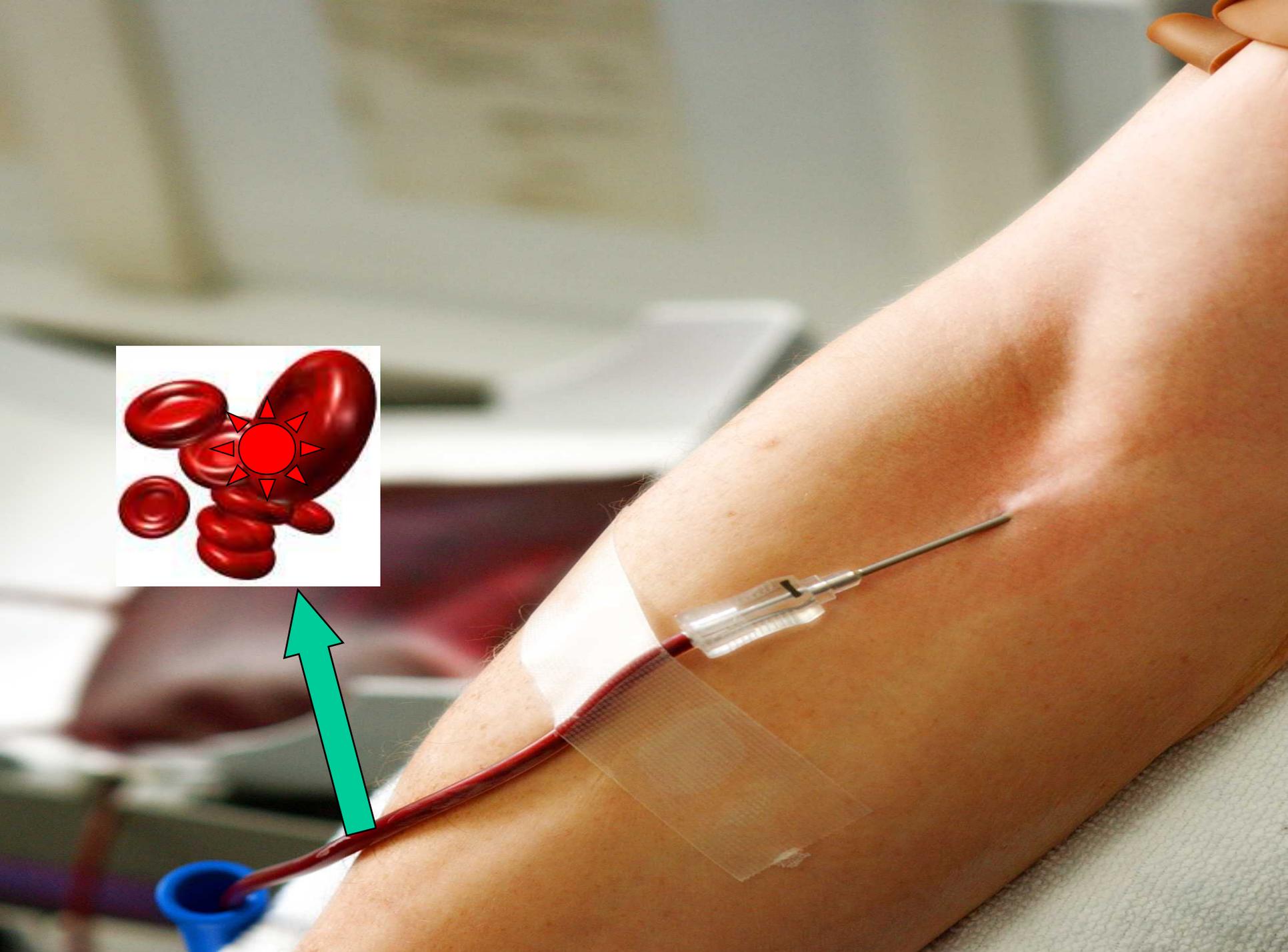
**Hémochromatose HFE
(C282Y/mutation rare)**

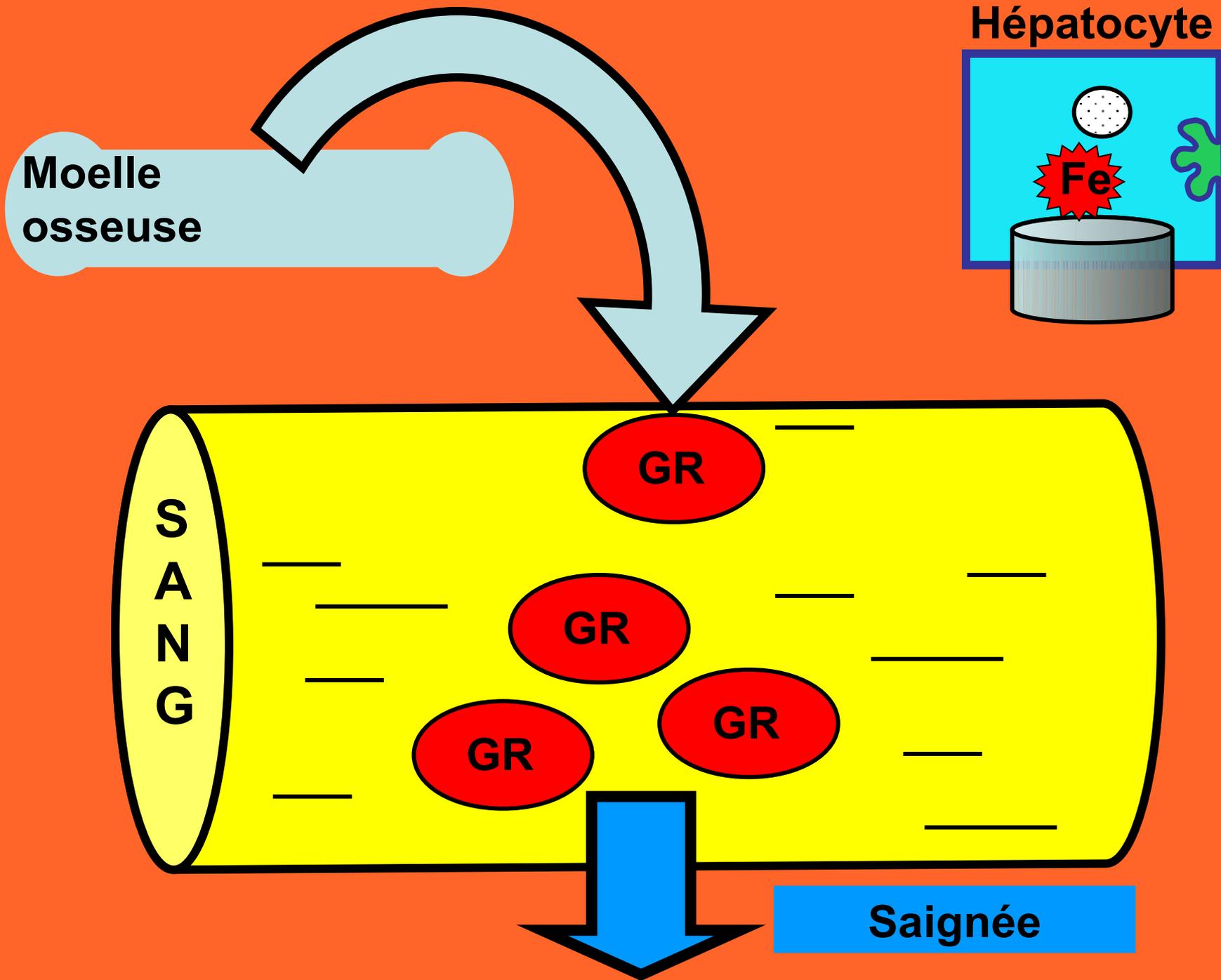
**Hémochromatose non HFE
(HJV , TFR2,...)**

Physio-pathologie

Diagnostic

Traitement





FUTUR

immédiat

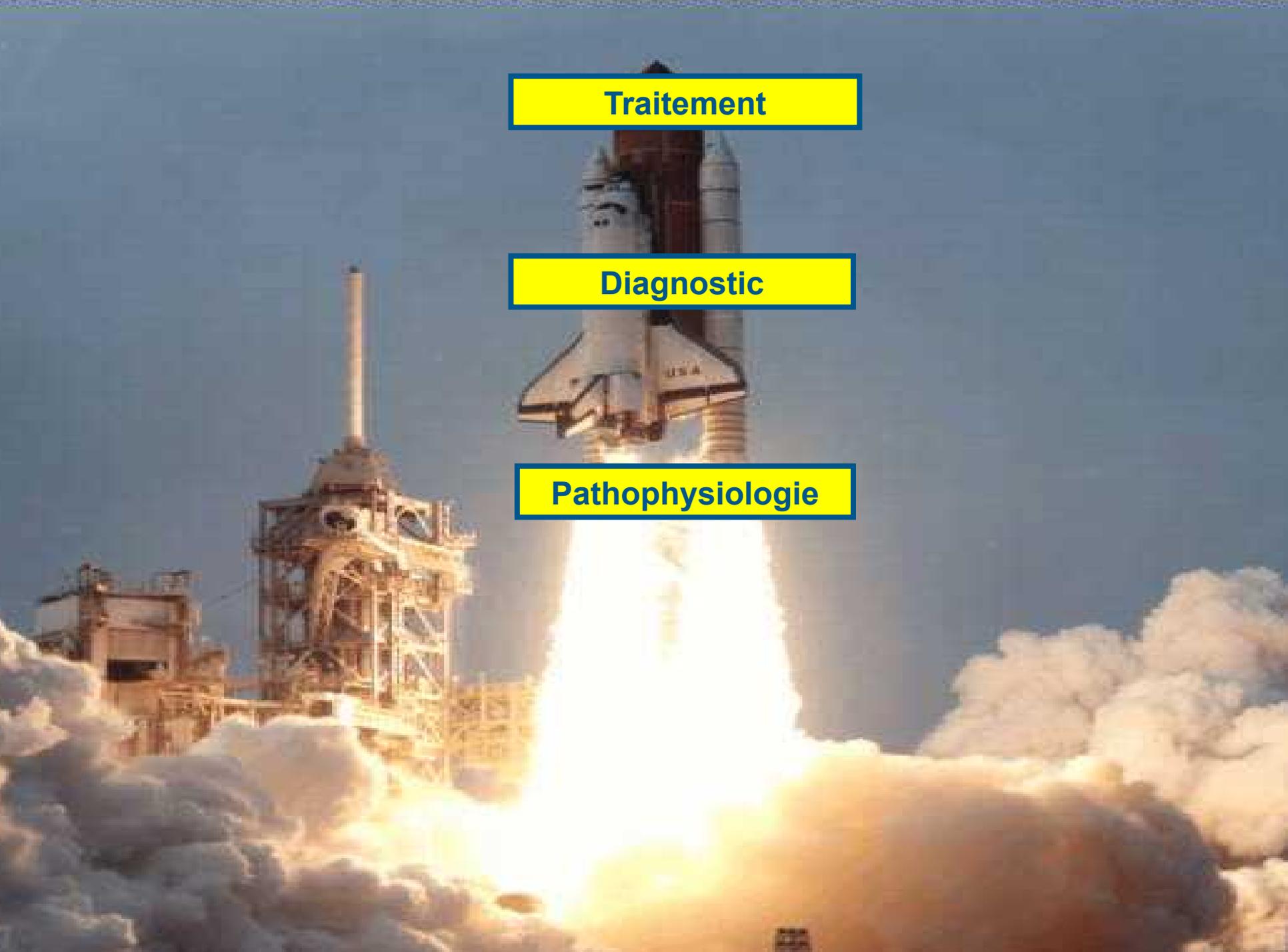
Chélation du fer

Déférasirox



A plus long terme

**Supplémentation en
hepcidine...**

A photograph of the Space Shuttle Columbia during its ascent. The shuttle is positioned vertically, with its external tank and boosters visible. A large, bright plume of fire and white smoke trails behind it, indicating a powerful launch. To the left, the launch pad service structure is partially visible. The sky is a clear, pale blue. Three yellow rectangular boxes with black borders are overlaid on the image, containing the text 'Traitement', 'Diagnostic', and 'Pathophysiologie' from top to bottom.

Traitement

Diagnostic

Pathophysiologie

Hémochromatose C282Y en 2014

«Take-home messages»

1. La surcharge en fer est l'aboutissant de la cascade : mutation C282Y-hypohepcidinémie-hyperhypersidérémie
2. La toxicité du fer est due au «fer non lié à la transferrine» et surtout à sa composante dite «fer plasmatique réactif »
3. L'homozygotie C282Y est nécessaire mais non suffisante au développement de la maladie

4. Le diagnostic est devenu non invasif, basé sur la clinique et la biologie, et... l'I.R.M en cas de facteur de surestimation de la ferritinémie

5. Le traitement reste basé sur la soustraction sanguine mais peut, dans de rares cas, recourir au déférasirox (hors AMM)

6. L'avenir thérapeutique de l'hémochromatose est la supplémentation en hepcidine.