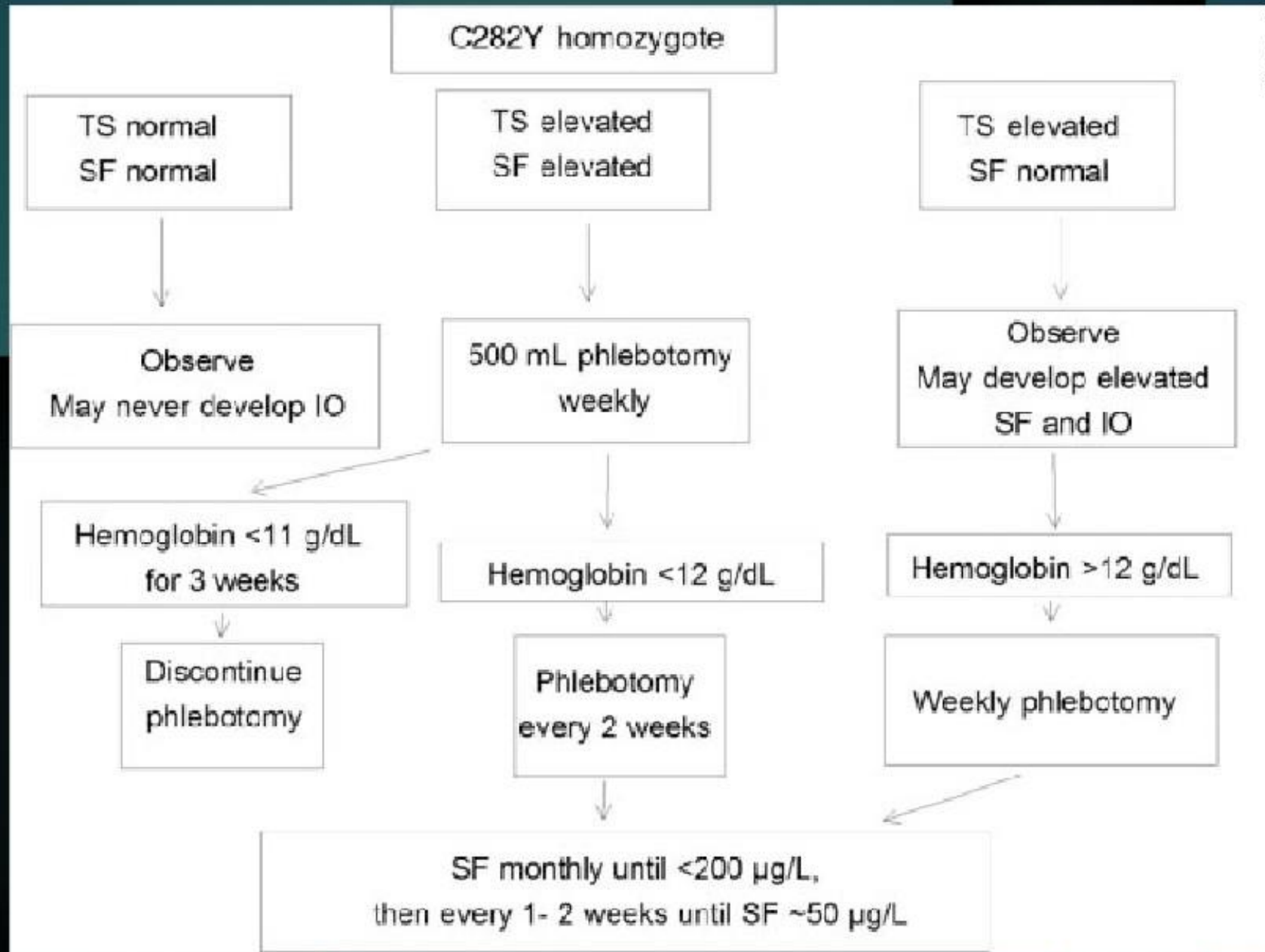


The background features a dark teal color with several geometric shapes: a large black rectangle on the left, a large teal circle on the right, a black square in the top right, a red vertical rectangle in the top right, and a black square in the bottom right.

# HAEMOCHROMATOSIS

- BY HARI SHANKAR PANDEY



## Hemochromatosis

Transferrin saturation >45% for female, >50% for males x2

HFE gene C282Y homozygosity = HFE hemochromatosis  
(C282Y/H63D compound heterozygotes or H63D homozygotes with increased ferritin or transferrin saturation-investigate for other causes of hyperferritinemia.  
Iron overload development is rare)

Serum ferritin - normal  
1 y follow up

Ferritin <1000 µg/L

Ferritin >1000 µg/L

Hemoglobin >12 g/dl  
weekly phlebotomy

500 ml phlebotomy  
weekly

Assess for cirrhosis  
with liver biopsy

Hemoglobin <12 g/dl  
phlebotomy every 2 wk.

Hemoglobin <11 g/dl for 3 wk  
discontinue phlebotomy

If cirrhosis: AFP  
and ultrasound  
every 6 mo to  
assess for HCC

Serum ferritin monthly until <200 mg/L then every 1-2 wk until ferritin ≤ 50 µg/L

### Lifestyle modifications

- Avoid iron supplements
- Limit vitamin C intake to 500 mg po qd
- Consume ethanol and red meat in moderation
- Do not consume raw shellfish

Immunize against  
hepatitis A and B

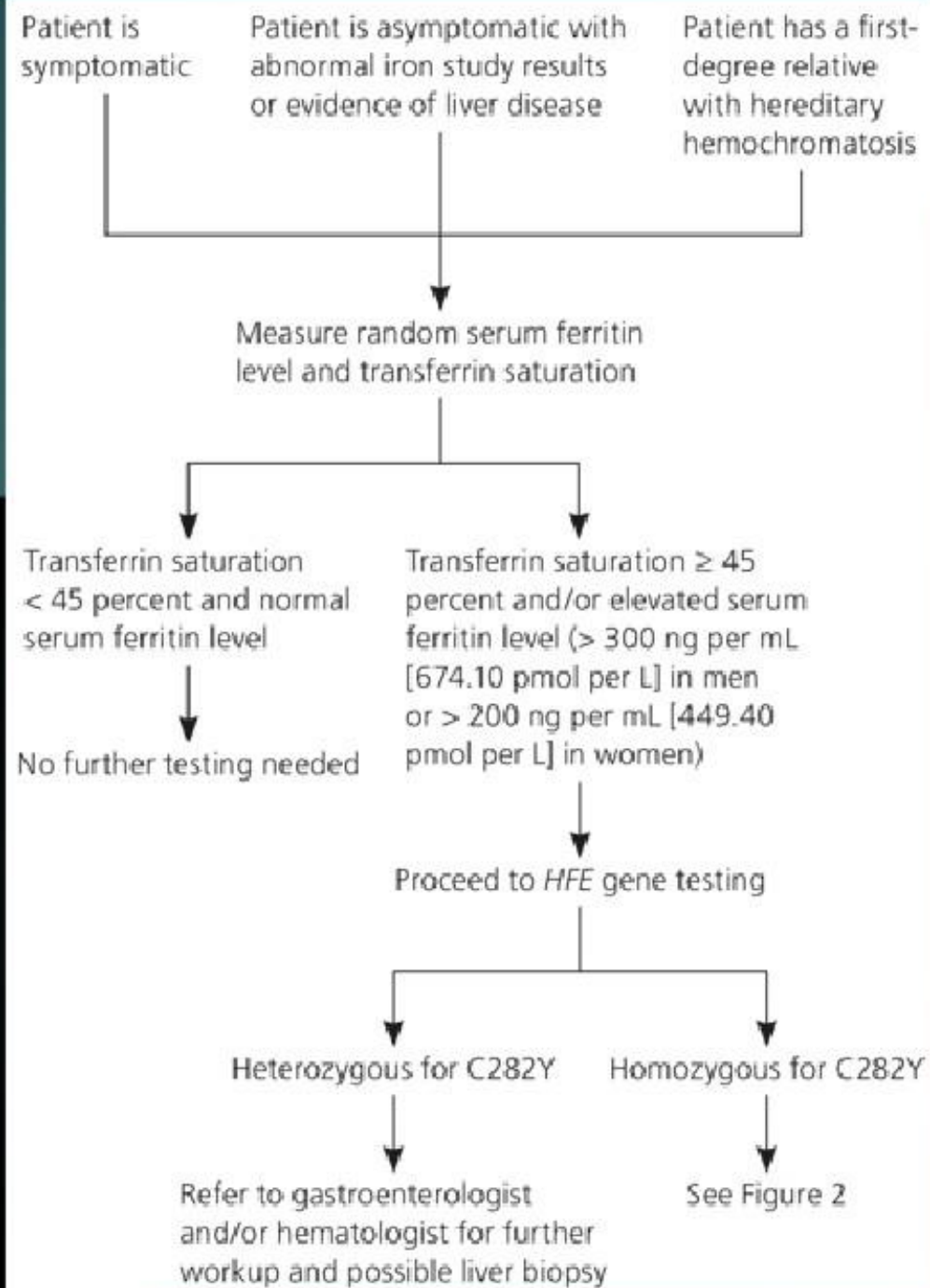
### Assess for complications

- Diabetes
- Joint disease
- Hypothyroidism,
- Cardiac disease
- Porphyria cutanea tarda
- Osteoporosis

Patients intolerant to phlebotomy due to anemia - consider iron chelation therapy although still experimental at this stage

- Deferoxamine\* - parentally administered iron chelator
- Deferasirox\* - oral iron chelator

\*not FDA approved for this indication



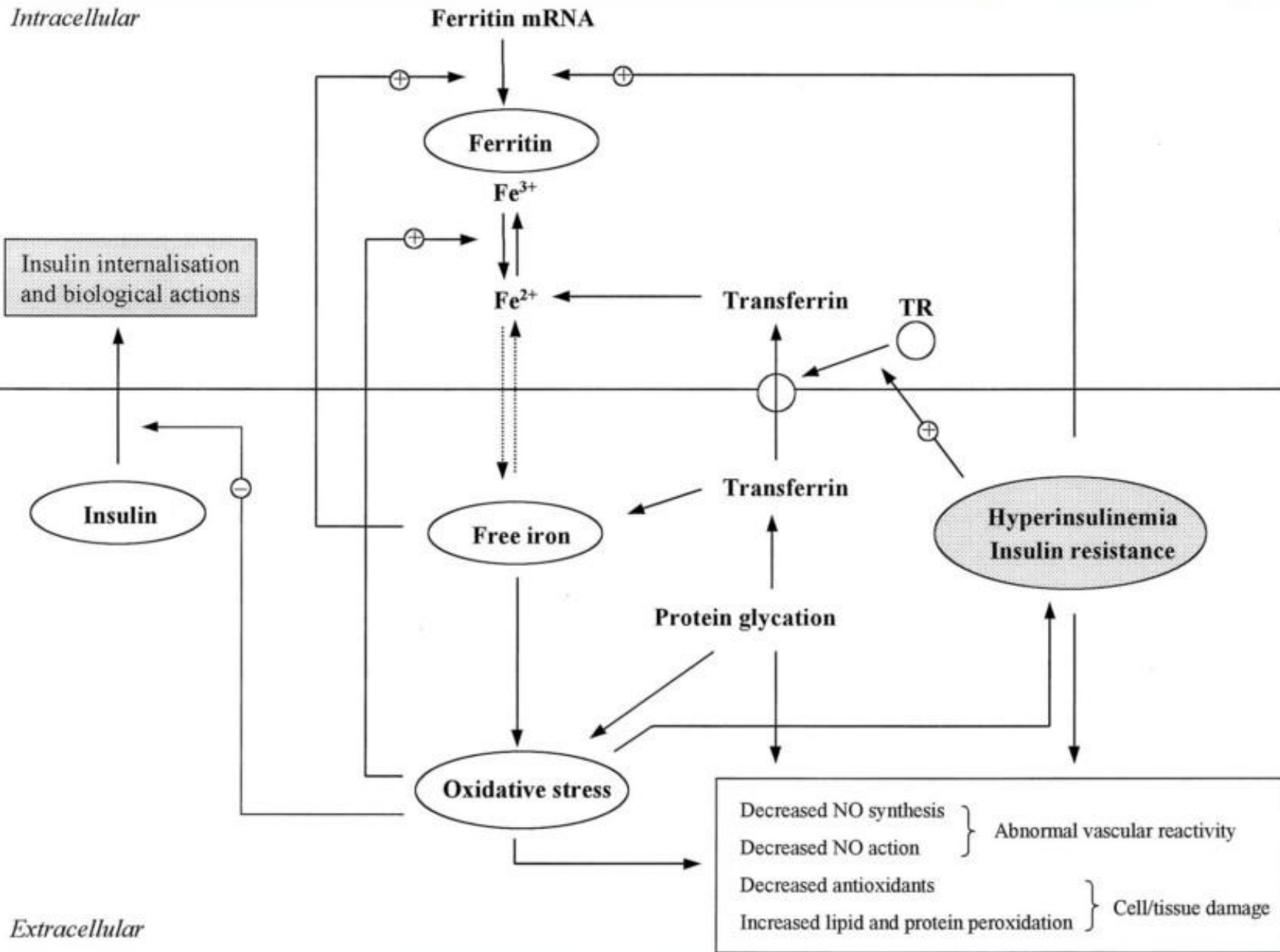
# Hemochromatosis Overview

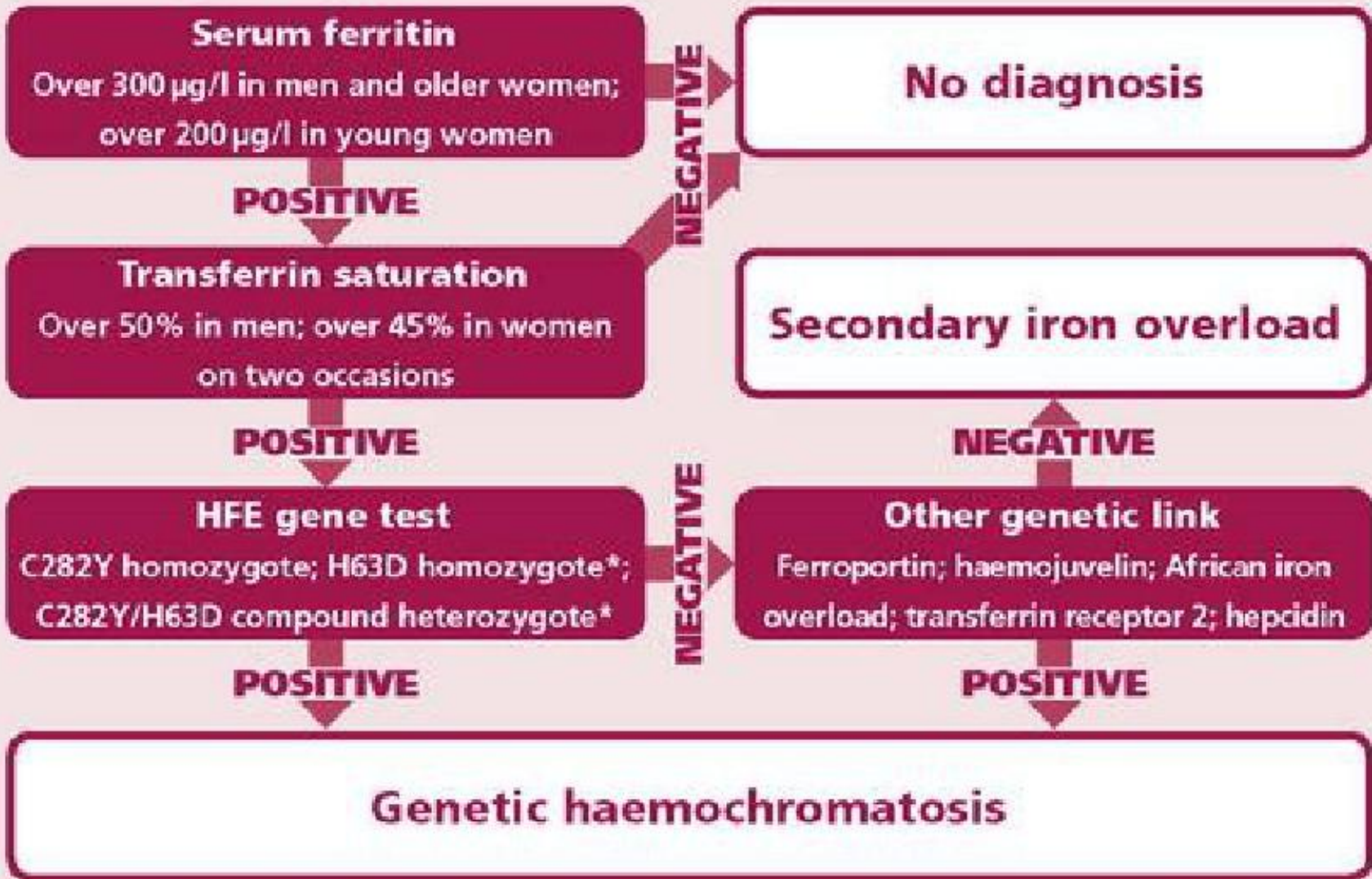
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## ■ History

- Classic triad described in the 1865 by Trousseau
    - Diabetes, bronze skin, cirrhosis
  - Named “Hemochromatosis” in 1889 by Von Recklinghausen
    - Iron storage and widespread tissue injury
  - Inheritance described in 1935
  - HLA linkage to chromosome 6 identified 1976
-

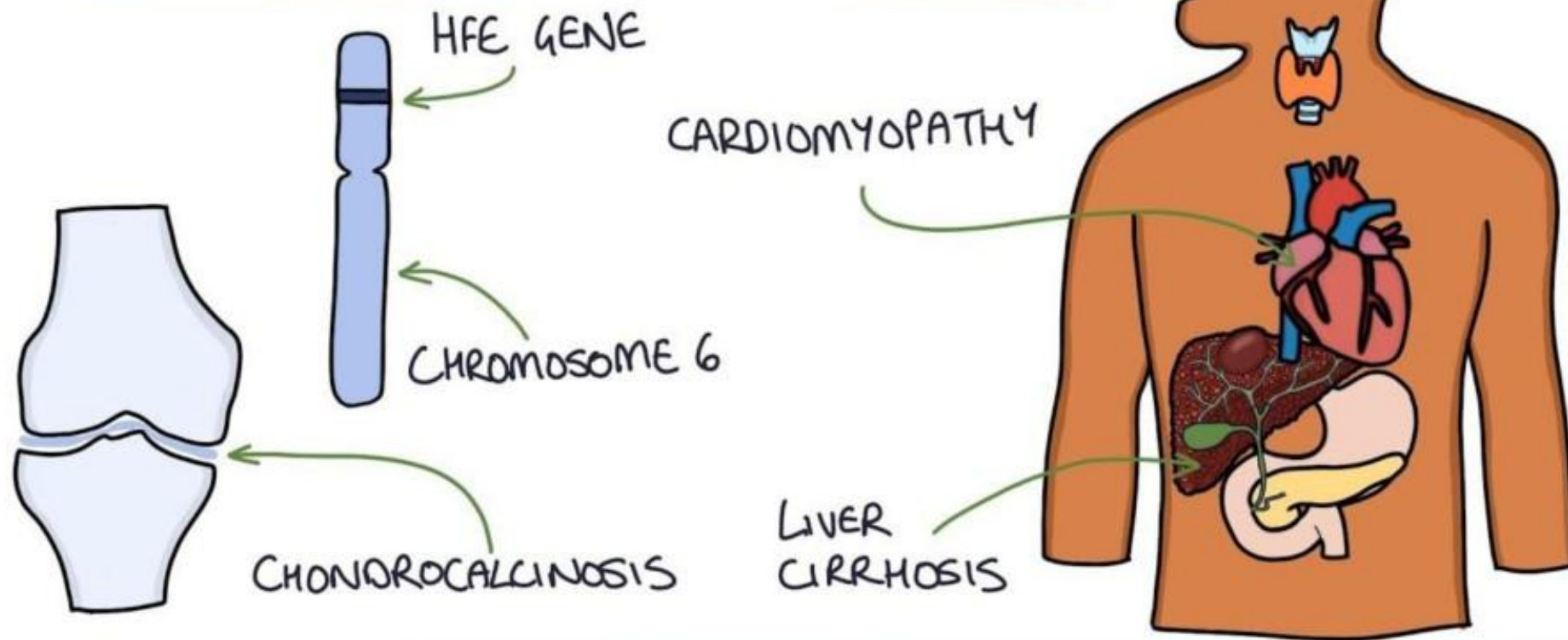
Intracellular





\*Rule out causes of secondary iron overload before confirming the cause as H63D homozygosity or C282Y/H63D compound heterozygosity

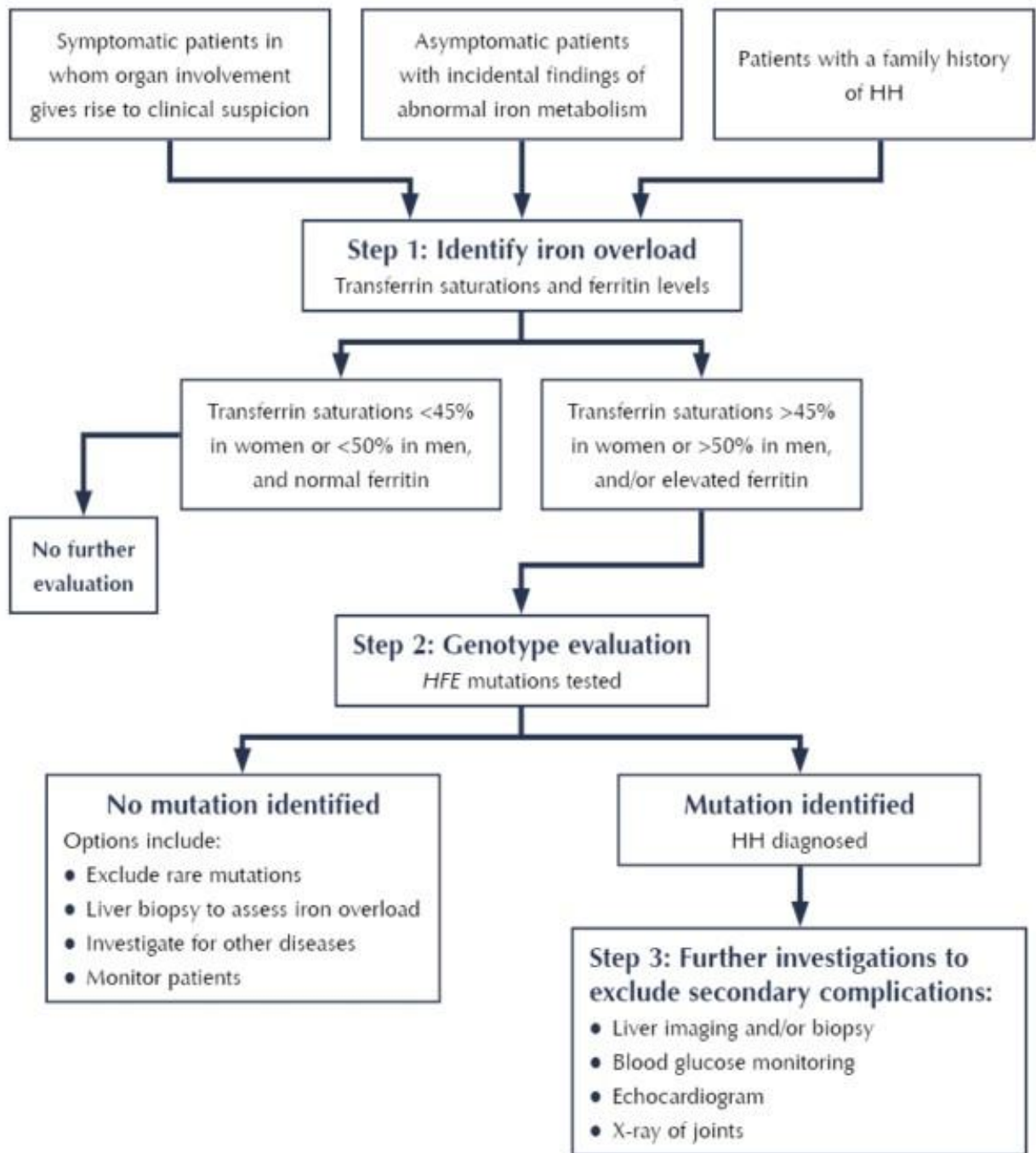
# HAEMOCHROMATOSIS





# Iron Overload or Hemochromatosis treatment

- The most common way to treat iron overload is to reduce the amount of iron in the body. This can be done through diet by eating **foods** low in iron or through the withdrawal of blood. The preferred treatment for reducing iron levels in hemochromatosis patients is called **therapeutic phlebotomy** (**repeated venesection**).



Factors unrelated to AβE that affect phenotypic expression



Pathogenic steps and principal biochemical effects



Proportion of C827P homozygotes expressing the indicated abnormality



## HFE hemochromatosis

- Caucasian, male, 40–50 years old
- Fatigue, dark skin, arthralgia and/or hepatomegaly
- Elevated TS and SF

## TFR2 hemochromatosis

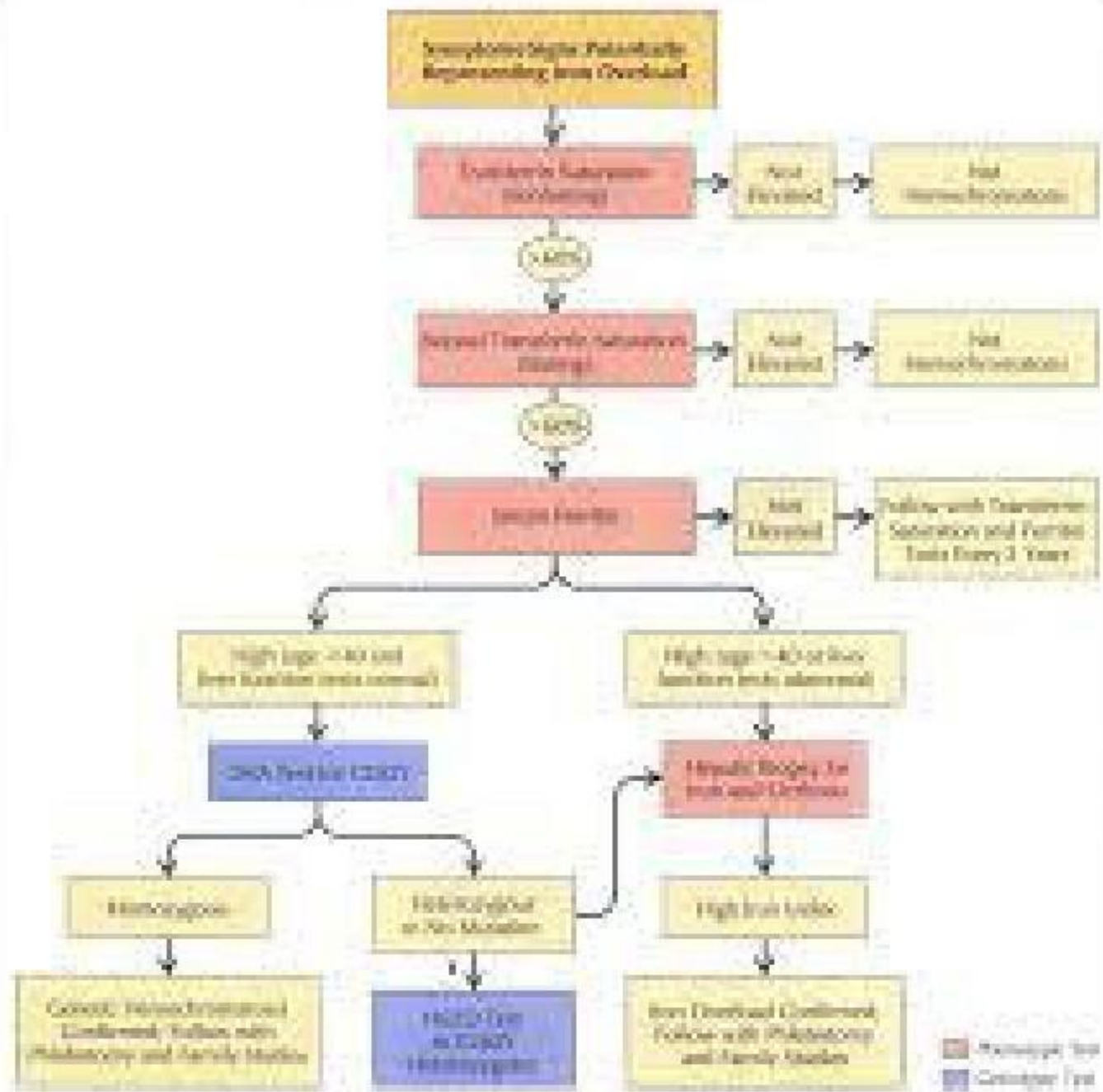
- Caucasian or non-Caucasian, male or female, 30–40 years old
- Cardiomyopathy, endocrinopathy, liver disease
- Elevated TS and SF

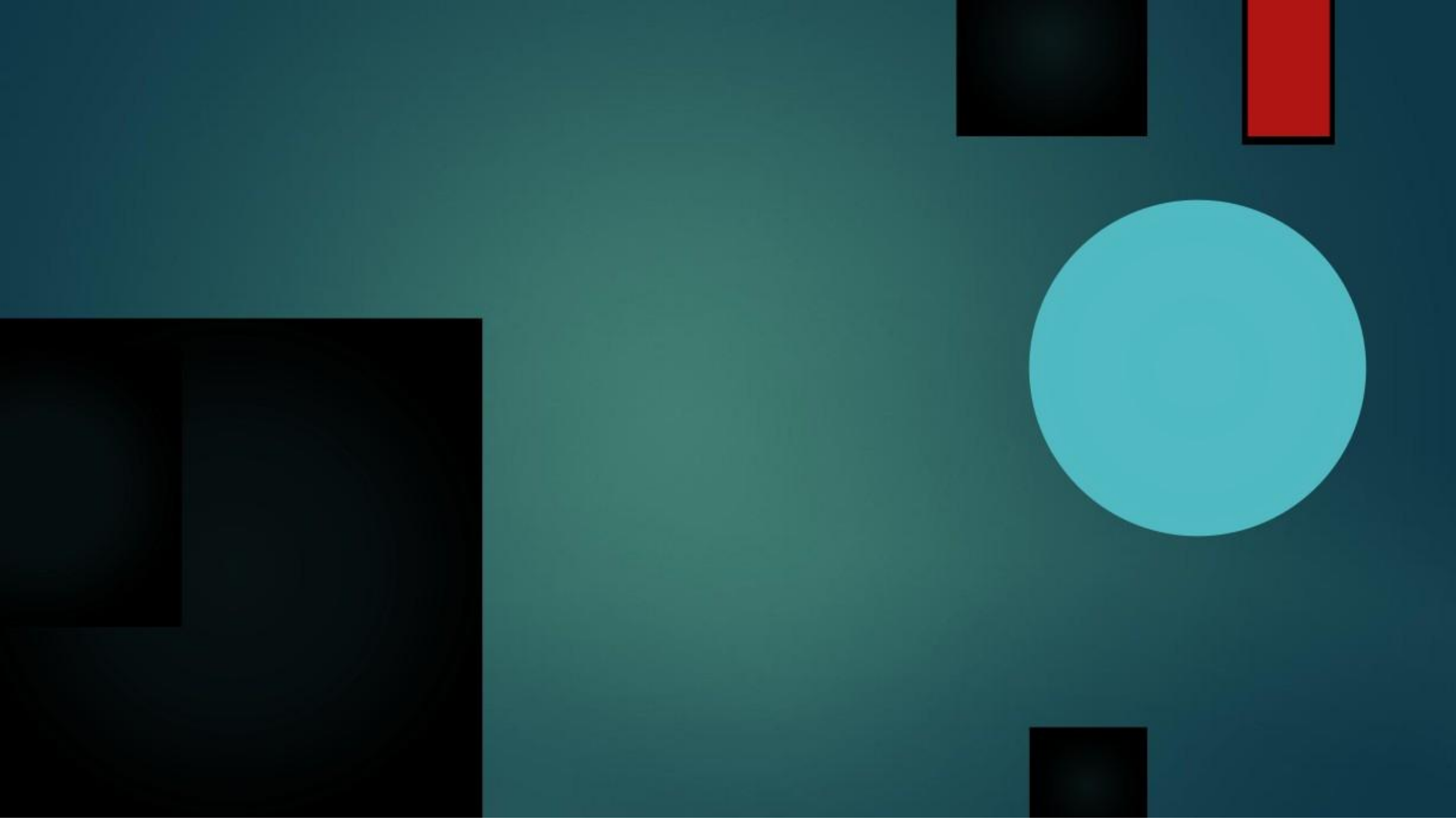
## HJV or HAMP hemochromatosis

- Caucasian or non-Caucasian, male or female, 15–20 years old
- Impotence/amenorrhea and/or cardiomyopathy
- High TS and SF

## Ferroportin disease

- Caucasian or non-Caucasian, male or female, 10–80 years old
- One parent with unexplained hyperferritinemia
- Unexplained elevation of SF and normal TS





HOW WAS HEMOCHROMATOSIS

DISCOVERED ?

HOW DOES

HEMOCHROMATOSIS AFFECT

DAILY LIFE ?

HOW MANY TYPE OF

HEMOCHROMATOSIS ARE THERE ?

CAN HEMOCHROMATOSIS CAUSE

MENTAL PROBLEM

?