

# Hemochromatosis

## What

- ① Genetic Iron Overload
- ② C282Y most common
- ③ Incomplete Penetrance 80%.  
20%.

## Who

Middle Age

F present later owing  
to menstrual blood loss

## Clinical Features

### MSK

2° OA  
2<sup>nd</sup>/3<sup>rd</sup> MCP  
Any joint

### Endo

Diabetes  
Hypogonadism  
Hypothyroidism  
Hypopituitarism

### Cardiac

CHF

### Liver

Chronic Liver  
Dz

### Skin

Hyperpigmentation

## Dx

### ① Iron Overload

Serum Tissue  
% sat > 40% ↑ Ferritin

Other { Cardiac / Liver MRI  
• Needed { Liver Biopsy

### ② Assess for alternate causes

See Iron Overload Schema

Alternate cause may exacerbate Hemochromatosis

### ③ Genetic Testing

### ④ Response to Rx

Anemia w/ Rx → Genotype was innocent bystander

## Rx

### ① Phlebotomy

### ② Iron Chelator