

CLINICAL FEATURES OF HH AND RESPONSE TO THERAPY

CLINICAL FEATURE	FREQUENCY	RESPONSE TO THERAPY
Lethargy, weakness and fatigue	Very common nonspecific initial symptom	Usually resolves
Elevated liver transaminases	Seen in up to 75% of clinical cases but often normal in asymptomatic cases	Usually resolve /marked improvement
Hypogonadotropic hypogonadism	Seen in 25-45%	If pituitary injury limited may resolve
Diabetes Mellitus	Uncommon	Often temporary improvement
Hyper or hypothyroidism	Rare	Resolution is rare
Cardiac effects (cardiac failure, conduction problems)	Rare	Unpredictable response
Hyperferritinemia	In all clinical cases	Responds to treatment
Hyperpigmentation	Seen in up to 70%	Usually resolves
Arthritis/arthralgia	Seen in up to 40%	Arthralgia may improve but arthritis usually does not respond
Hepatomegaly	Variable	Resolution often occurs provided cirrhosis absent
Hepatic cirrhosis/risk for Hepatocellular cancer	< 5%	Usually no change. Regression of fibrosis does occur but not cirrhosis
Right upper quadrant pain	Uncommon	May resolve if due to iron overload alone