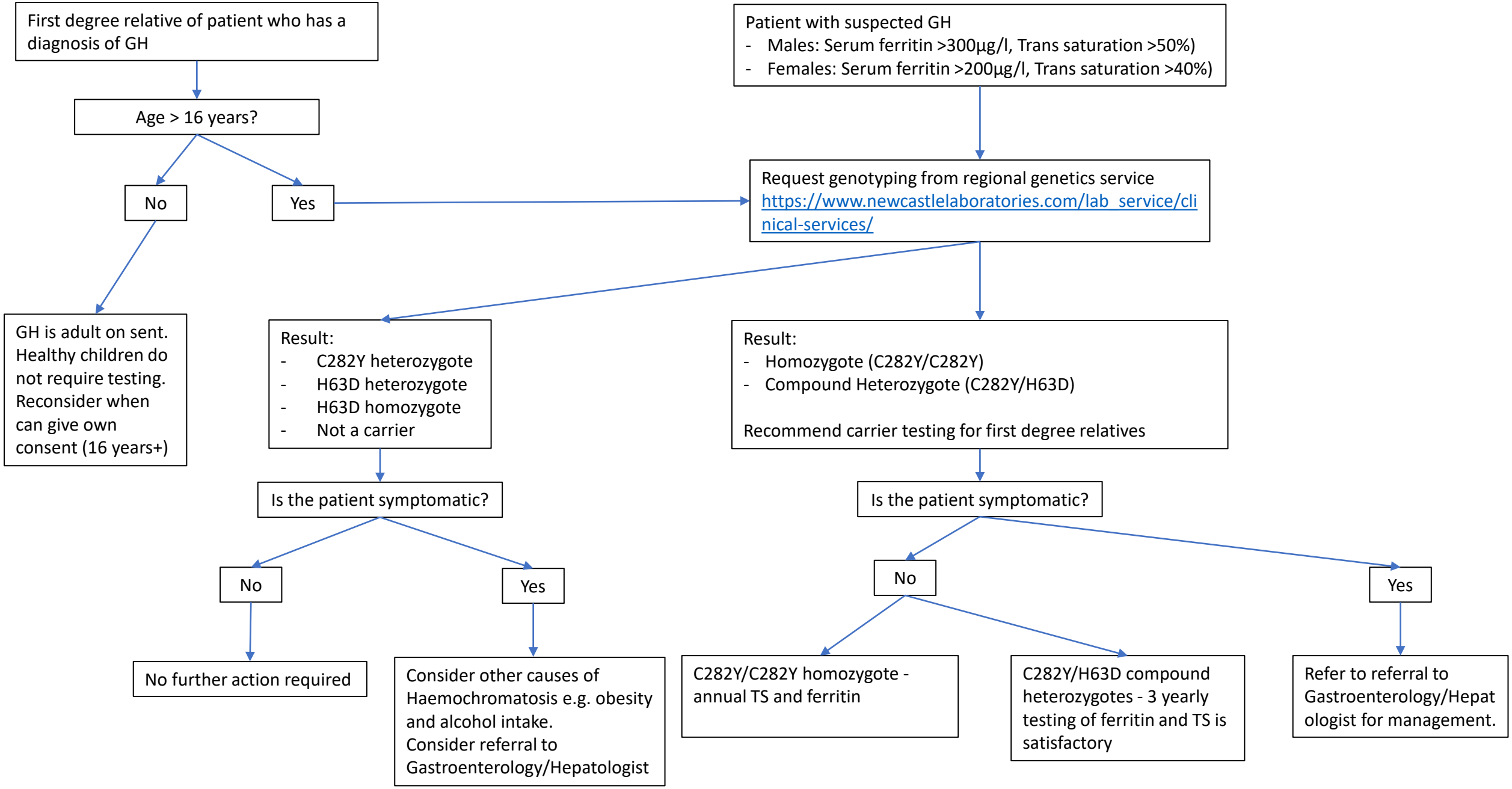
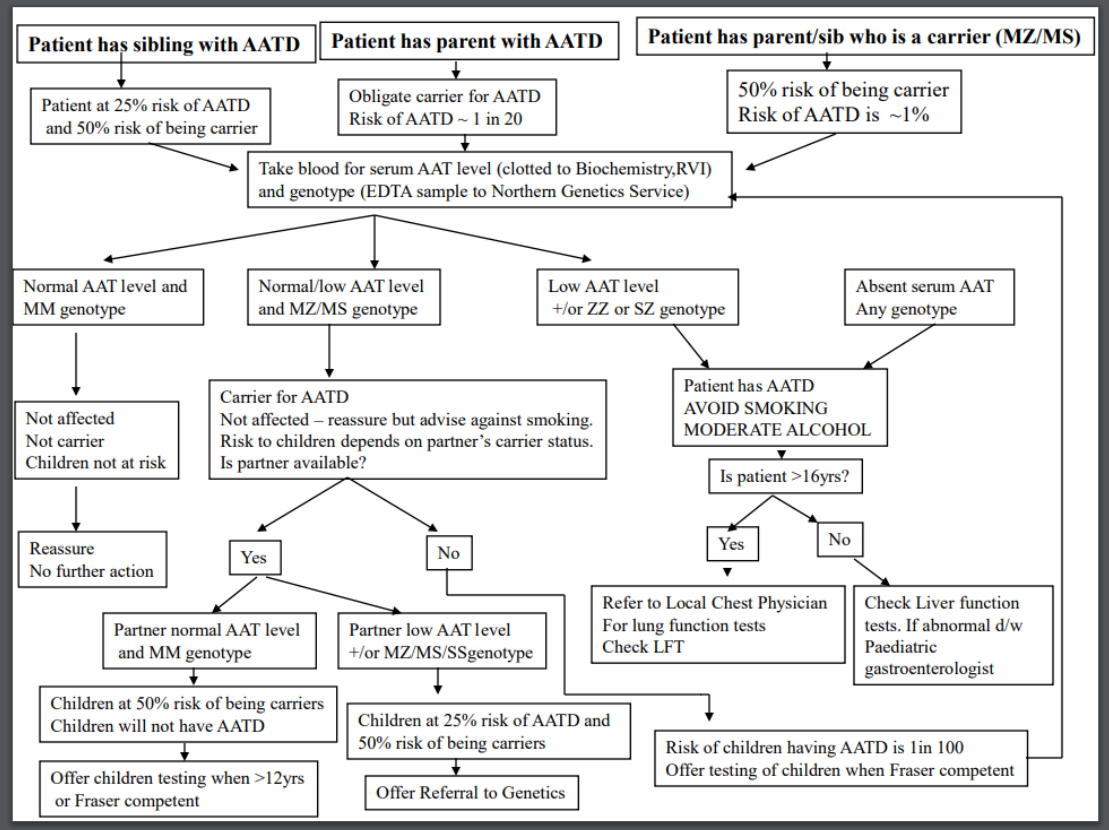

First degree relative of patient who has a diagnosis of GH
Age > 16 years?
No
Yes
GH is adult on sent. Healthy children do not require testing. Reconsider when can give own consent (16 years+)
Patient with suspected GH
- Males: Serum ferritin >300µg/l, Trans saturation >50%)
- Females: Serum ferritin >200µg/l, Trans saturation >40%)
Request genotyping from regional genetics service https://www.newcastlelaboratories.com/lab_service/clinical-services/
Result:
- C282Y heterozygote
- H63D heterozygote
- H63D homozygote
- Not a carrier
Is the patient symptomatic?
No
Yes
No further action required
Consider other causes of Haemochromatosis e.g. obesity and alcohol intake.
Consider referral to Gastroenterology/Hepatologist
Result:
- Homozygote (C282Y/C282Y)
- Compound Heterozygote (C282Y/H63D)
Recommend carrier testing for first degree relatives
Is the patient symptomatic?
No
Yes
C282Y/C282Y homozygote - annual TS and ferritin
C282Y/H63D compound heterozygotes - 3 yearly testing of ferritin and TS is satisfactory
Refer to referral to Gastroenterology/Hepatologist for management.


Patient has sibling with AATD
Patient has parent with AATD
Patient has parent/sib who is a carrier (MZ/MS)
Patient at 25% risk of AATD and 50% risk of being carrier
Obligate carrier for AATD Risk of AATD ~ 1 in 20
50% risk of being carrier Risk of AATD is ~1%
Take blood for serum AAT level (clotted to Biochemistry,RVI) and genotype (EDTA sample to Northern Genetics Service)
Normal AAT level and MM genotype
Normal/low AAT level and MZ/MS genotype
Low AAT level +/or ZZ or SZ genotype
Absent serum AAT Any genotype
Not affected Not carrier Children not at risk
Carrier for AATD Not affected – reassure but advise against smoking. Risk to children depends on partner's carrier status. Is partner available?
Patient has AATD AVOID SMOKING MODERATE ALCOHOL
Is patient >16yrs?
Reassure No further action
Yes
No
Yes
No
Refer to Local Chest Physician For lung function tests Check LFT
Check Liver function tests. If abnormal d/w Paediatric gastroenterologist
Partner normal AAT level and MM genotype
Partner low AAT level +/or MZ/MS/SSgenotype
Children at 50% risk of being carriers Children will not have AATD
Children at 25% risk of AATD and 50% risk of being carriers
Risk of children having AATD is 1in 100 Offer testing of children when Fraser competent
Offer children testing when >12yrs or Fraser competent
Offer Referral to Genetics