



First degree relative of patient who has a diagnosis of AATD

Age > 16 years?

No

Yes

AATD is adult on sent. Healthy children do not require testing. Reconsider when can give own consent (16 years+)

Request genotyping from regional genetics service  
[https://www.newcastlelaboratories.com/lab\\_service/clinical-services/](https://www.newcastlelaboratories.com/lab_service/clinical-services/)

Patient with suspected AATD

Following tests to be initiated:  
- Genotyping  
- Serum AAT levels (clotted to Biochemistry at RVI)

No

Yes

Result: MM genotype – patient is not a carrier or affected

Result: MS or MZ genotype – patient is a carrier

Result: ZZ, SZ, SS genotype – patient affected

- Healthy lifestyle advice to avoid smoking and moderate alcohol intake
- If symptomatic consider referral to Gastroenterology and/or Chest Physician

- Refer to Gastroenterology and Chest Physician
- Healthy lifestyle advice to avoid smoking and moderate alcohol intake
- Recommend carrier testing for first degree relatives