Oxford Alpha-1 antitrypsin deficiency care pathway for routine referrals

Referral of:
Symptomatic proband with low a1AT plasma concentration suggesting Pi type ZZ or ZS

- Letter (1A) to Referrer requesting phenotype confirmation
- Letter (1C) to Patient (Enclose information & management guidelines)

Any other phenotype

- Standard letter to Referrer (1B)

ZZ/ZS phenotype or homozygosity/compound heterozygosity for variant alleles that include rare but likely significant alleles e.g. null allele or discrepancy between a1AT level and phenotype

- Genetic counsellor appointment (See GP for referral to specialist)
- Standard letters: Patient (1D/1D(1))
- Relatives
- GP and/or referrer (1F)

Referral of:
- Probands with previously identified ZZ or ZS phenotype or rare significant alleles
- Couples where both partners are already known carriers

- Standard letter to GP and/or Referrer (1E)
- (Enclose information & management guidelines)

Further information re: family history/neonatal jaundice and/or early lung or liver disease

- Genetic counsellor telephone to assess for appointment (See GP for referral to specialist)
- Standard letters: Patient (1D/1D(1))
- Relatives
- GP and/or referrer (1F)

Referral for family history of a1AT deficiency or family history of a known carrier

Referral for known carrier of a1AT deficiency

Referral for high anxiety or pregnancy

KEY
- Clinical genetics Appointment
- Advice by letter