

Alpha 1 antitrypsin

Refer for possible replacement therapy: SZ, ZZ, ZF, null

1:13 Americans have a good and bad gene.

MS – no liver disease, no increased lung disease with smoking. Usually normal level. Children/spouse need testing. Give carrier brochure.

Everyone in the family can get free testing at www.alphaoneregistry.org

They will send a questionnaire and kit (finger stick)

No therapy

MZ=MF -90% risk with smoking/5 fold increase in emphysema/bronchiectasis and airways disease; occasion liver disease. Give brochure for liver disease, carrier, lung disease.

MI is similar but no liver dx.

SZ-if smoke/job (farmer, dust fumes, welders)/second hand smoke/, get lung disease. Can get augmentation therapy.

ZZ- liver dx the worst, especially young adults. CBC with low plt. Follow PFTs and DLCO. Walk test. U/S of liver, LFTs and alphetoprotein.

Z- A single Z allele may lead to liver disease, especially if concomitant hemochromatosis gene mutations or Hep B infection.

S null

F gene – normal levels but bad function

SS- no risk liver dx, lung no increased risk and normal levels

In patients with bronchiectasis and emphysema, also test for CF mutation and NTMB infections.