Research Agenda of the Alpha-1 Foundation
(Lay version)
Revised by MASAC 10/14/2016

BASIC RESEARCH: IDENTIFYING TARGETS FOR NEW THERAPIES
• Mechanisms of Z alpha-1 antitrypsin synthesis, folding and secretion in the liver
• Mechanisms of lung tissue destruction
• Determinants of liver tissue regeneration
• Determinants of lung tissue regeneration
• Animal Model Development of alpha-1 antitrypsin deficiency and associated liver and lung disease
• Animal model evaluation of new therapies
• Liver cell transplantation
• Gene therapy
• Small molecule, rapid testing of drug candidates
• Stem-cell therapy

CLINICAL RESEARCH: EPIDEMIOLOGY & NATURAL HISTORY OF ALPHA-1 ANTITRYPSIN DEFICIENCY
(focus on ZZ and MZ genes)
• Targeted detection and newborn screening
• Predicting course and outcomes
• Natural history following lung and liver transplantation
• New diagnostic technologies
• Markers of disease severity and progression, and response to therapy
• Quality of life, patient reported outcomes, healthcare costs, and symptom management
• Evaluation and treatment of comorbidities in alpha-1 antitrypsin deficiency

EVALUATING NOVEL THERAPEUTIC APPROACHES
• Alpha-1 antitrypsin augmentation therapy
  o Dosing of AAT therapy
  o Aerosolized AAT therapy
  o Recombinant AAT
• Augmentation therapy in lung transplant recipients
• Anti-inflammatory therapy for lung disease
• Substitutes for alpha-1 antitrypsin in lung disease treatment
• Stem cell therapies
• Gene-directed therapies
• Therapies for liver disease

ETHICAL, SOCIAL & LEGAL ISSUES RESEARCH
• Social dimensions of living with alpha-1 antitrypsin deficiency
• Equitable access to, and distribution of medical therapies
• Impact of genomic information and precision medicine on alpha-1 patients and their families
• Burdens of illness on patients and caregivers
• Disease risk and lifestyle choices
• Patient-centered research
DEVELOPING AND RENEWING THE ALPHA-1 RESEARCH WORKFORCE

- Clinical research training in AAT deficiency
- Basic research training in AAT deficiency
- Alpha-1 career development