

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
 - Dosing of AAT therapy
 - Aerosolized AAT therapy
 - 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