



Rare and Genetic Disease Network

CME / CE

Rare Disease Clinical Trials

Module: Rare Pulmonology Clinical Trials

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Rare Pulmonology Conditions

What is a rare disease?

- A rare disease is any disease affecting less than 200,000 people in the United States

Why is a Rare Disease Designation Important?

- Orphan Drug Act
- No one left behind

Common vs Rare

- Asthma vs Eosinophilic Asthma
- COPD vs Alpha-1 Antitrypsin Deficiency
- Bronchitis vs Cystic Fibrosis



A Few Examples

Alpha-1 antitrypsin deficiency

- Inherited conditions that can lead to lung disease in young adults
- *15 clinical trials currently recruiting in U.S.*

Cystic fibrosis

- Chronic lung condition due to genetic mutation that leads to excessive mucus build up
- *91 clinical trials recruiting*

Idiopathic pulmonary fibrosis

- Progressive loss of lung function due to scarring of lungs. Cause is unknown
- *40 clinical trials recruiting*

Pulmonary arterial hypertension

- High blood pressure in lungs that can lead to cardiac failure
- *98 clinical trials recruiting*

Rare Pulmonology Research is Common

Number of Clinical Trials on Pulmonary Diseases

	1990 - 1999	2000 – 2009	2010 – April 2017
Lymphangioleiomyomatosis (LAM)	2	2	17
Idiopathic pulmonary hypertension	33	60	116
Pulmonary hypertension	464	685	660
Sarcoidosis	96	80	71
Non-cystic fibrosis bronchiectasis	1	10	20

Why Join a Clinical Trial?

Current treatment is...

- Not safe
- Not available
- Not effective

Other reasons

- Free access to new treatments
- Advance science

Are clinical trials safe?

- Clinical trials are designed by experts to succeed, not fail
- Clinical trials have highly regulated and intense safety monitoring.
- Monitoring of side effects more common in clinical trials than in standard care

Discussing Clinical Trials With Your Patient

Prior the meeting

- Is there a clinical trial?
- Is the patient eligible?
- What type of clinical trial is it?

Setting up the meeting

- Quiet room with no interruptions
- Allow caregiver/friend/relative to attend
- Pen, paper, laptop to take notes

Things to do during meeting

- Explain the trial is voluntary
- Explain the disease, current treatment options (and then the trial)
- Use simple terminology
- Listen to the patient (they are in charge)

After the meeting

- Reinforce the notion that the patient has rights
- Schedule a call/meeting with PI
- Determine your role in helping the patient

Common vs Rare Clinical Trials

	Common Conditions	Rare Conditions
Phase 1	10-100 patients	5-20 patients
Phase 2	100-500	5-40
Phase 3	500-5000	10-500
Inclusion Criteria	Very strict	Strict
Exclusion Criteria	Very strict	Strict
Number of locations	Numerous	Few
Recruiting patients	Easier	More Difficult

How to Research Clinical Trials

 U.S. National Library of Medicine

ClinicalTrials.gov

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Condition or disease ⓘ

Alpha 1-Antitrypsin Deficiency

x

Other terms ⓘ

x

Country ⓘ

United States

▾ x

State ⓘ

▾ x

City ⓘ

x

Distance ⓘ

▾

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15 Studies found for: **Recruiting Studies | Alpha 1-Antitrypsin Deficiency | United States**Also searched for **ANTITRYPSIN** and **AATD**. [See Search Details](#)Applied Filters:
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Condition or disease ⓘ **Other terms** ⓘ

Alpha 1-Antitrypsin Deficiency x

Country ⓘ **State** ⓘ **City** ⓘ **Distance** ⓘ

United States x Pennsylvania x Hershey x

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Zero Studies Currently Recruiting in PA

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Condition or disease  **Other terms** 

Alpha 1-Antitrypsin Deficiency x x

Country  **State**  **City**  **Distance** 

United States  x Pennsylvania  x x 

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5 Studies found for: **Recruiting Studies | Alpha 1-Antitrypsin Deficiency | Pennsylvania, United States**

Five Studies Currently Recruiting in PA

Search Results (5 trials recruiting in PA)

Study Title	Condition	Intervention	Locations
Efficacy and safety of Alpha1-protease inhibitor in people with PE due to AATD	PE due to AATD	Drug: Alpha-1 MP	47 locations
Carbamazepine in sever liver disease due to AATD	AATD Liver cirrhosis	Drug: Carbamazepine	3 locations
Alvelestat for the treatment of AATD	AATD	Drug: Alvelestat	9 locations
Glassia safety, immunogenicity and bronchoalveolar lavage study	AATD	Biological: Glassia	20 locations
Evaluating the genetic causes and progression of cholestatic liver diseases	AATD Liver cirrhosis Alagile syndrome		17 locations

PE = pulmonary emphysema; AATD = alpha-1 antitrypsin deficiency

Pros and Cons

Pros

- Focused Care
- Advancing Science
- Free Medicine
- Possible Treatment

Cons

- Travel expenses
- Time commitment
- Placebo?
- Safety and efficacy?



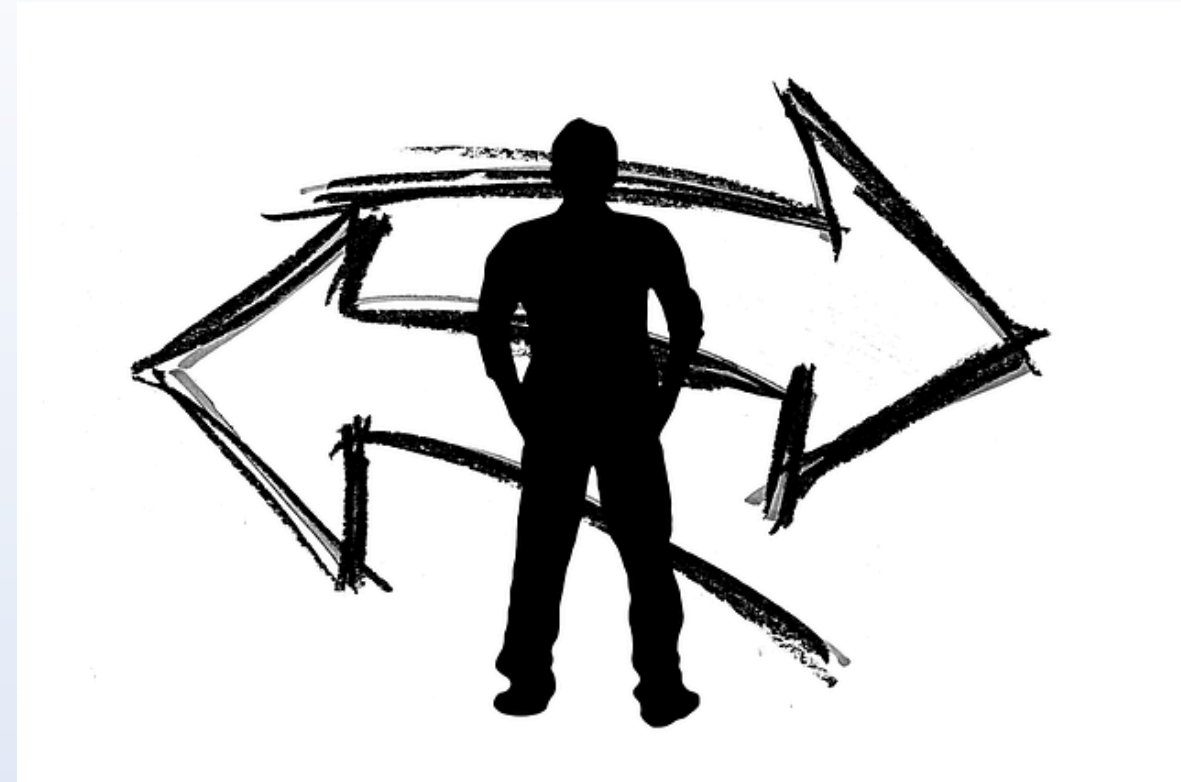
Case Study

Patient

- 47 yr old male
- Pulmonary emphysema due to alpha-1 antitrypsin deficiency
- Current therapy options not effective

Concerns

- Patient wants to be enter clinical trial but has some misgivings about:
 - Placebo
 - The impact the trial will have on family
 - Being treated by a different practice



Summary

- Rare diseases affect < 200,000 people in the United States
- Many rare diseases in great need for better treatments
- Joining a clinical trial is one option
- Discussing a clinical trial with your patient is a sensitive discussion
 - Do your homework
 - Listen to what the patient wants
- Clinicaltrial.gov is an excellent source to find clinical trials