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February 2026

Rare Disease Day: Shining a Light on Alpha-1 Antitrypsin Deficiency

Each year on **February 28**, Rare Disease Day brings attention to the millions of people worldwide living with conditions that are often overlooked or misunderstood. In the infusion center, we have the unique opportunity to support individuals affected by rare diseases every day. One such condition is **Alpha-1 Antitrypsin Deficiency (Alpha-1)**.

Alpha-1 Antitrypsin Deficiency is a **genetic condition** that affects the body's ability to protect the lungs and, in some cases, the liver. People with Alpha-1 have low levels or abnormal forms of a protein called **alpha-1 antitrypsin (AAT)**. This protein is produced in the liver and released into the bloodstream, where it plays a key role in protecting lung tissue from inflammation and damage.

When AAT levels are too low, the lungs are more vulnerable to injury over time. This can lead to early-onset lung diseases such as **chronic obstructive pulmonary disease (COPD)**, emphysema, or chronic bronchitis. Some individuals with Alpha-1 may also develop liver disease due to abnormal AAT buildup in the liver. Because symptoms often resemble more common respiratory conditions, Alpha-1 is frequently **underdiagnosed or diagnosed late**.

Alpha-1 is considered a rare disease, affecting approximately **1 in 2,500 individuals**, yet awareness remains limited. Rare Disease Day is an important reminder of the value of early testing and diagnosis. A simple blood test can measure AAT levels, and genetic testing can confirm the condition. Early identification allows patients to take steps to protect their health, including avoiding smoking, reducing exposure to lung irritants, and staying current on recommended vaccinations.

For patients with Alpha-1–related lung disease, **augmentation therapy** is an important treatment option.

RARE DISEASE DAY®

This therapy involves regular infusions of alpha-1 antitrypsin protein to help raise AAT levels in the blood and protect the lungs from further damage. While augmentation therapy does not cure Alpha-1 or reverse existing lung damage, it can help **slow disease progression** when used appropriately. Infusion therapy is often combined with pulmonary rehabilitation, oxygen therapy, and ongoing care from a lung specialist.

Infusion centers play a vital role in the care of patients with Alpha-1 by providing safe, supportive treatment environments and ongoing education. Beyond the medical benefits, infusion visits also offer opportunities for patients to ask questions, share concerns, and feel supported by a knowledgeable care team.

On Rare Disease Day, we recognize the strength and resilience of individuals living with Alpha-1 Antitrypsin Deficiency and reaffirm our commitment to providing compassionate, high-quality infusion care. By raising awareness, encouraging early diagnosis, and supporting ongoing treatment, we help ensure that patients with rare diseases receive the attention and care they deserve—today and every day.