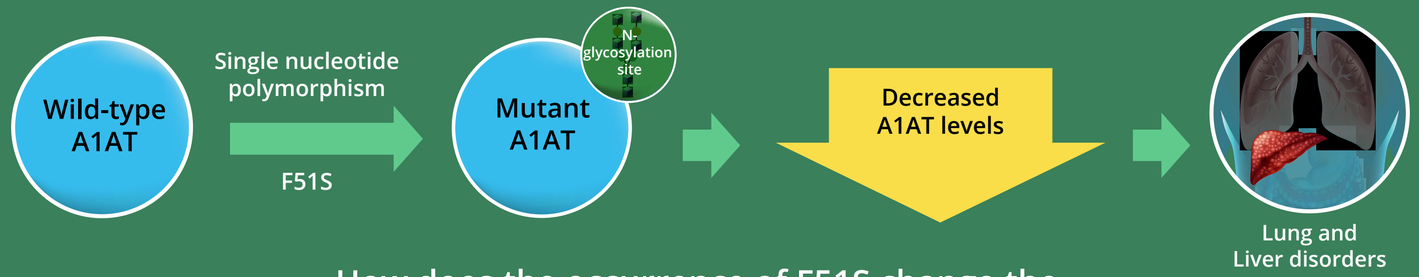
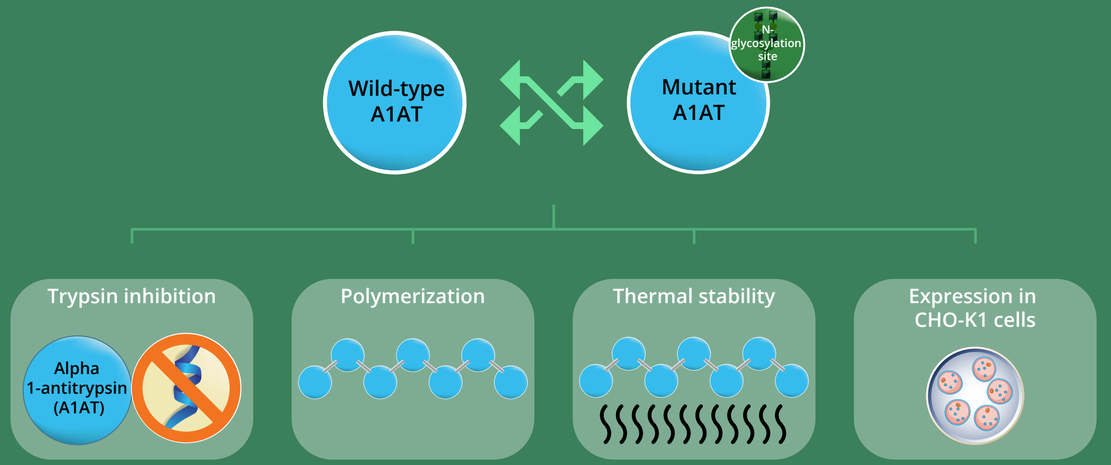
Characterizing the rare F51S mutation of alpha 1-antitrypsin

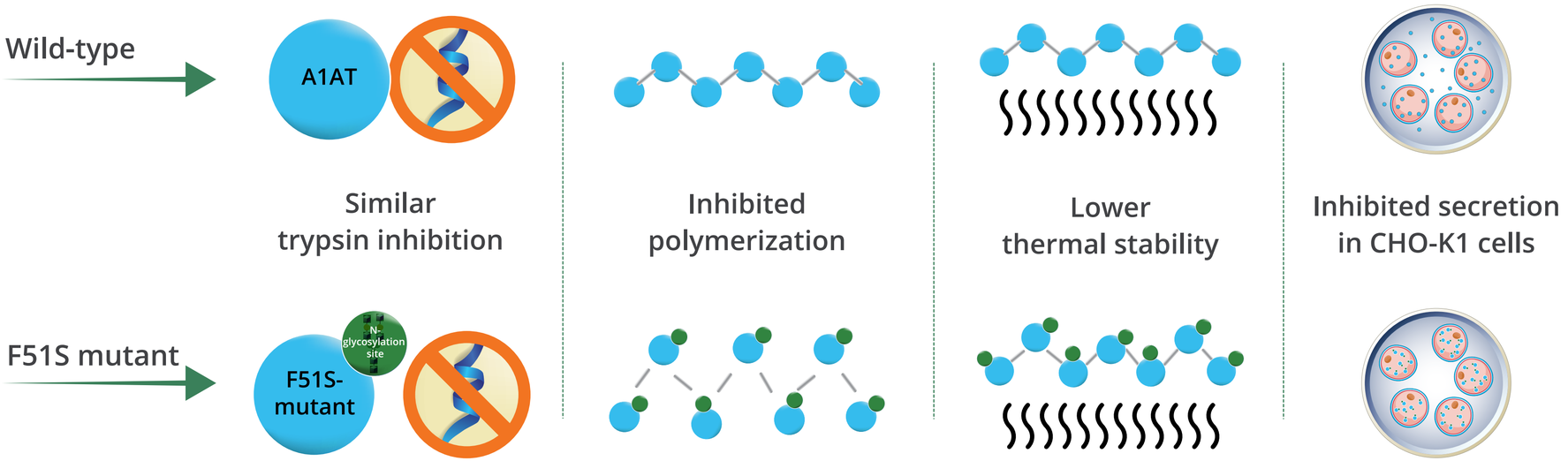
Alpha 1-antitrypsin (A1AT) deficiency caused by gene mutations is associated with lung and liver diseases



How does the occurrence of F51S change the conformation and stability of the A1AT?



Compared to the wild-type A1AT, mutant A1AT had...



F51S mutation alters A1AT conformation and secretion, implying that it plays a role in A1AT deficiency, and can aid researchers in finding cures for lung and liver diseases caused by A1AT deficiency.

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FUNCTIONAL CHARACTERIZATION OF A SNP (F51S) FOUND IN HUMAN ALPHA 1-ANTITRYPSIN

TRINH *et al.* (2019) | DOI: 10.1002/MGG3.819

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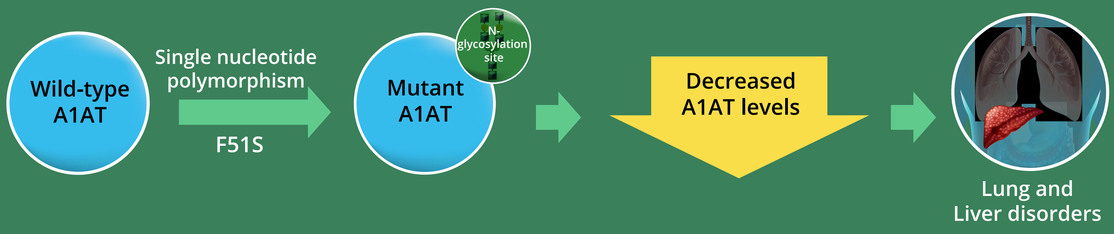

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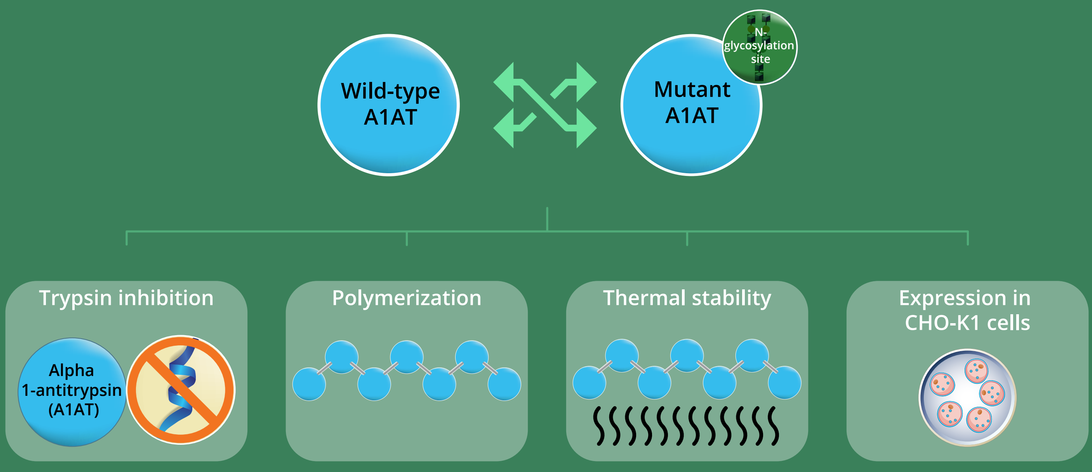

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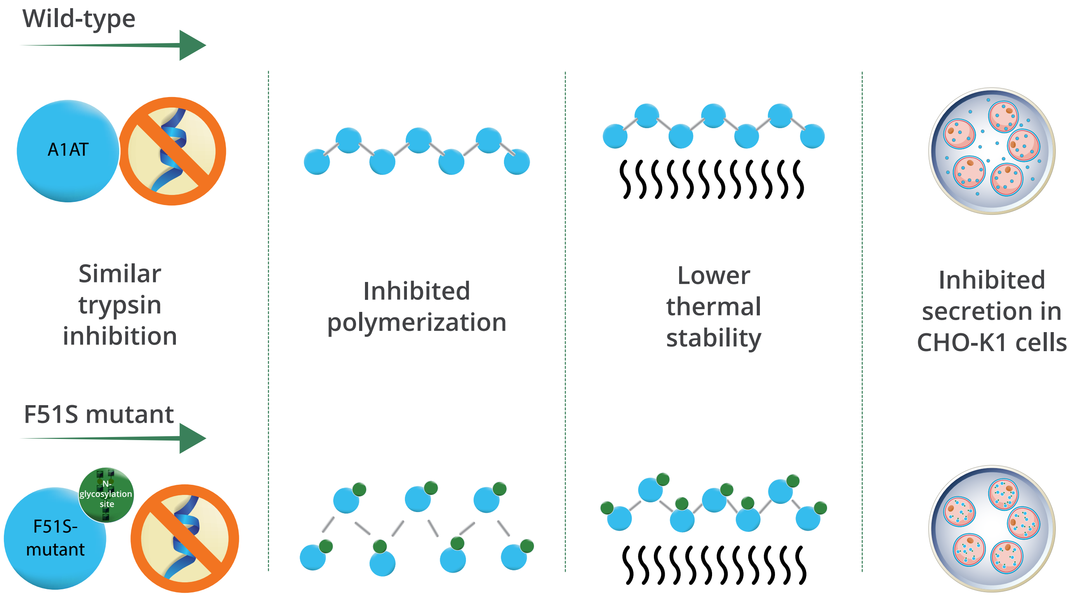

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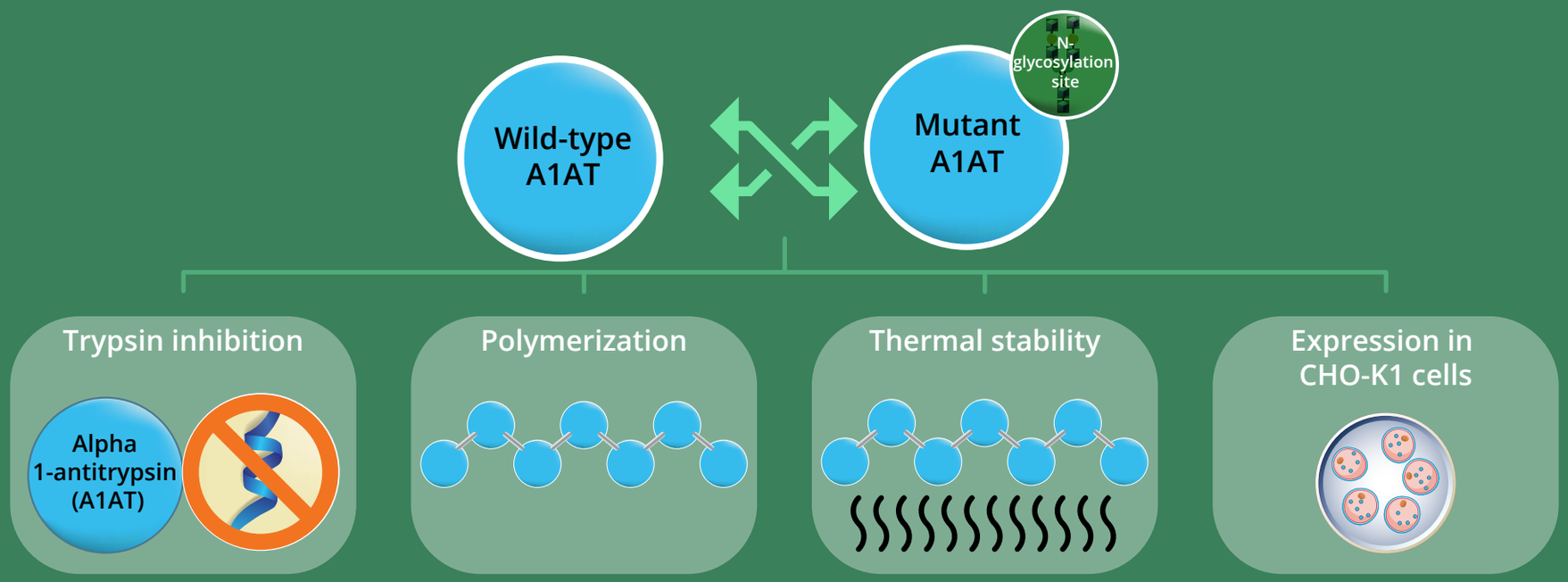

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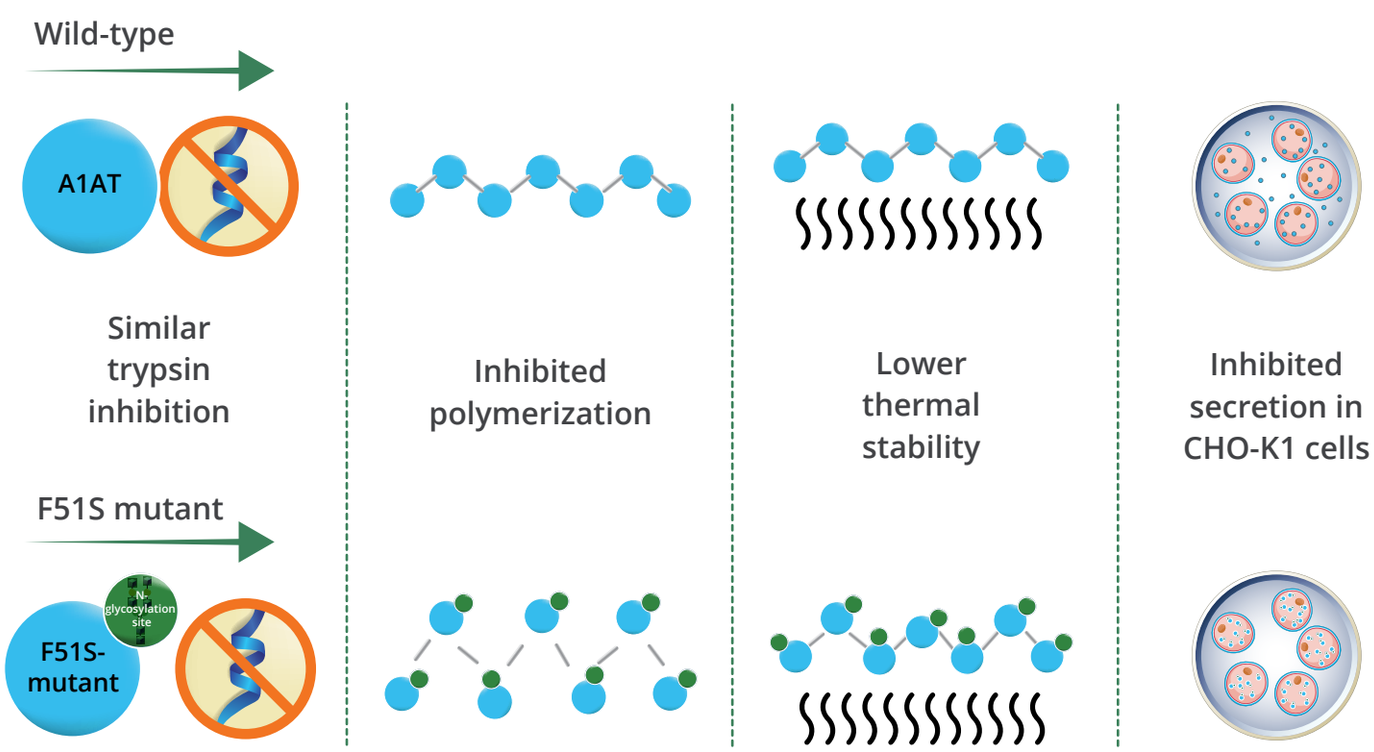

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