Appendix 1 (as provided by the authors): Applying the results of this review in clinical practice (fictional case)

Claire is a 40-year-old woman who was diagnosed with asthma nine years ago by her family physician. Her diagnosis was made based on symptoms of dyspnea on exertion, episodic wheeze, and cough; she has never undergone spirometry. She has been treated with inhaled budesonide and formoterol twice daily, plus salbutamol as needed. She smoked a ½ pack of cigarettes per day from age 18 to age 32. Claire's asthma symptoms were initially mild, but have worsened gradually. She can no longer keep up with her husband during their morning 'power-walks', and she has made two trips to the emergency room this past year for exacerbations of her asthma that were slow to resolve despite prednisone therapy. Her family physician referred her to a respirologist for further evaluation.
At the respirologist's office, she undergoes spirometry, which reveals moderate airway obstruction with a significant bronchodilator response, but her postbronchodilator FEV_1 (forced expiratory volume in 1 second) is only 56% of predicted. Given that she has substantial residual airflow limitation after bronchodilator, more complete pulmonary function testing is requested and a serum alpha$_1$ antitrypsin level is ordered.

Claire's alpha$_1$ antitrypsin level returns at 26 mg/dL (lower limit of normal 90 mg/dL). She undergoes alpha$_1$ antitrypsin genotyping which shows that she is homozygous for the Z allele. Full pulmonary function tests shows moderate airways obstruction with gas trapping and a reduced diffusing capacity, consistent with emphysema. Her chest x-ray is normal, as are her liver enzymes and function tests. She is advised to discuss alpha$_1$ antitrypsin deficiency screening with her family members, and the importance of maintaining abstinence from cigarette smoking and avoidance of detrimental respiratory exposures is discussed. She receives the influenza and pneumococcal vaccines. Given her persistent dyspnea, a long-acting anticholinergic is added to her current medical therapy. Pulmonary rehabilitation is considered, but she does not have significant exercise limitation to warrant it; it may be offered in the future. She is referred to a respirologist with experience managing alpha$_1$ antitrypsin deficiency, for consideration of augmentation therapy.

Appendix to: Brode SK, Ling SC, Chapman KR. Alpha-1 antitrypsin deficiency: a commonly overlooked cause of lung disease. *CMAJ* 2012. DOI:10.1503/cmaj.111749.