Survey of our Alpha1 community – patients and clinicians

- 100% of respondents supported streamlining pathology testing for Alpha1 Antitrypsin. That is, reducing the amount of testing from 4 possible tests to 3 and potentially 2 tests.
- 100% of respondents agreed that people with respiratory symptoms indicative of AAT deficiency and abnormally low AAT level, or people with a demonstrated family history of AAT deficiency, should be eligible for and recommend the new test.
- 92% of respondents said that the test should be recommended for people with adult onset asthma and all persons diagnosed with COPD.
- 100% of respondents believe that patients undergoing genetic testing should have access to genetic and psychological counselling while waiting for results and after receiving results.
- 96% of respondents believe that understanding the genetic variation of AAT deficiency would provide useful knowledge to help people make informed decisions about lifestyle and work factors, such as diet, smoking, liquor consumption and exposure to environmental pollutants.
- 100% of respondents believed that early understanding of the genetic variation of AAT deficiency would help in the management of diseases, such as liver disease, COPD and chronic bronchitis, caused by low AAT levels.
- 92% of respondents saw no disadvantage in having this test list on the Medicare Benefits Schedule.

One respondent nominated that: "there are a several potential disadavantages, including the potential of over testing and increasing cost. The implications of a genetic diagnosis on increased insurance premiums as well as no readily available treatment options."

Individual written responses:

- After my diagnosis I had a bilateral lung transplant 12 years later. I was then able to finally get a diagnosis for my niece and my daughter took three tests before getting a positive. She will not have children now which is tragic. Genetic counselling is imperative as we are unaware who else in our family has inherited this very debilitating syndrome.
- I would hope that such testing would not have people being misdiagnosed or told they would be dead in 2 years as I was There was no known history in the family.
- I have A1ATD, SZ genotype. My son has recently had himself tested. He paid for the tests himself. He was fortunate that he is in the financial situation where he could afford to do this. Very many others would not be so fortunate and could be stressed not knowing.
- This test could save the tax-payer in the long run, giving people informed decisions about their health and making better choices based on their medical condition. Some people don't even know they have this condition.

- Can't happen quickly enough ALPHA1 is a neglected disease all-round in identifying education and treatment of patients - I have had it for 20 years
- Any assistance to have the disease diagnosed early is an advantage to all sufferers.
- Early testing: I am all for it. Mine was not discovered until too late now I have been told I am too old for any help.
- My experience is having had two phenotype tests. I am a relatively healthy alphaone zz 67 yr old male. My three sisters are also zz alphas, one of whom progressed to a thankfully successful double lung transplant. Genotyping has the potential to help clarify genetic variations that lead to severe illness; at the moment we have limited information to assist with diagnosis and treatment selection.
- Will this test be available to AAT children too?
- By having the relevant information about the deficiency easily diagnosed, effective treatments could start much earlier and without unnecessary delay. This easier testing could also be an effective way of providing knowledge to the general public about the ramifications of this deficiency.