alpha-1
ORGANISATION AUSTRALIA INC

Alpha Times

Newsletter of Alpha-1 Organisation Australia inc

Issue 3 Autumn 2021

From the President's Pen

Happy New Year everyone. Hopefully 2021 will be a safer time for all with alpha-1 and other chronic illness due to the rollout of COVID19 vaccines. I have been monitoring vaccination experiences/ feedback on several Alpha-1 Facebook pages and it seems that most people have had no or only mild side effects. The decision to act early or to wait (as the Australian COVID vaccination implementation rollout occurs) is of course very personal but the good news is that adverse events are very rare and with high vaccination levels, Alphas should feel much safer venturing out.

This year is shaping up to be a good one for our charity with new initiatives and projects. We have applied and been accepted to become a partner organisation with Rare Voices Australia, which offers many benefits. See www.rarevoices.org.au. These include policy support, strategy mentorship, invitations to rare disease events and greater connection into their rare disease national advocacy activity. We now have a strategic relationship with the National Asthma Council and have been invited to comment on the Australian Asthma Handbook. This is important as many alphas are misdiagnosed with asthma. Our aim is to have a "think alpha-1" inclusion in the diagnosis section of the handbook to assist with a timely diagnosis of alpha-1.

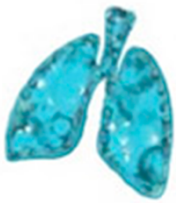
Hornsby District Rotary Club now has alpha-1 antitrypsin deficiency as an "area of focus", helping raise awareness.

All the best
Gaynor
--
Gaynor Heading PhD
President
Alpha-1 Organisation Australia inc

SPOTLIGHT ON RARE DISEASES DAY
28 FEBRUARY

Help raise awareness of this condition that affects many adults and children.

Alpha-1 Antitrypsin Deficiency
A genetic condition passed from parent to child through genes



Covid 19 been getting you down?

Are you feeling isolated or cut off from your normal activities?

Here are some tips for surviving shutdowns and the "new normal".

1. Start a hobby that you can do at home, e.g., sewing, drawing, woodwork, gardening, reading, puzzles, walking.
2. Join Zoom and have regular virtual meetings with family, friends, book club, exercise groups, faith groups.
3. If you are feeling anxious, try some mindfulness and relaxation techniques, e.g., breathing exercises, muscle relaxation exercises, meditation. Exercise is good for relieving stress. Binge-watch your favourite series.
4. If you are feeling stressed about catching Covid 19, be sure to follow health guidelines, and practise good personal hygiene, including always wearing a mask when in a crowd. Limit the amount of time you spend catching up on Covid 19 updates. Exercise or do a repetitive, soothing activity such as weeding or chopping vegetables.
5. Start a positive diary. Write down 3 positive things that happened each day.

Today was a good day. I picked some flowers from my garden, I went for a walk with my dog and I did some shopping for the old lady across the road.

Newly Diagnosed? Part 2.

In Part 1 of this series, we looked at a few things to do as a priority when you're first diagnosed with Alpha-1 antitrypsin deficiency (A1AD). In Part 2 we look at what this might mean for your relatives.

As A1AD is genetic (hereditary), you have inherited the condition from your parents. You get one gene from each parent, so if you have one abnormal (i.e. not M) gene, one or both of your parents may also have one or two abnormal genes, whereas if you have two abnormal genes (e.g. ZZ or SZ), then both of your parents have at least one abnormal gene. To illustrate this, in my case, being ZZ, I know both my parents were either MZ or ZZ. As both lived to a normal life expectancy and were asymptomatic it is likely that both were MZ.

Knowing that one or both of your parents had at least one abnormal gene, it follows that your siblings (if any), together with any uncles/aunts/nephews or nieces may also have an abnormal gene and you should advise all your blood relations of your condition so that they can choose to get tested. Because most of them will not have heard of A1AD it is worth pointing them to some introductory information such as the brochure 'What is Alpha-1 Antitrypsin Deficiency' which can be found on our website (Alpha-1 Organisation Australia | Resources (a1oa.org.au)).

If you have children, they may have inherited an abnormal gene from you – clearly if you have two abnormal genes they definitely will have, whereas if you have one abnormal gene, they may have inherited a normal or abnormal gene from you. They inherit their second gene from their other parent and therefore if you don't want your children tested at this stage, testing their other parent may help – again, in my case, my son's other parent has two normal genes (MM) so we know that he is MZ, and will have his liver and lung health closely monitored for any symptoms. Drawing a 'family tree' with known gene status of each family member can help as you communicate with the rest of your family.

In Part 3 of this series, we'll start looking in more detail at some of the typical symptoms of A1AD, starting with neonatal liver issues.

Update on application to MSAC for Genotyping of rare Alpha-1-Antitrypsin variants

There is an application to MSAC from the Royal College of Pathologists seeking more comprehensive alpha-1 antitrypsin genotype testing to include the 14 variants of alpha-1 that are most associated with disease, where the patient has low alpha-1 antitrypsin (AAT) levels and is a non-smoker, has emphysema at a young age, or panniculitis, or cirrhosis of the liver or abnormal liver function (including neonatal hepatitis) without other risk factors, or anti-proteinase 3-positive vasculitis, or family history of AAT deficiency.

Applications proceed through the Advisory sub-committee (PASC) and Evaluation sub-committee (ESC). The public can provide feedback during the process. The application has been considered by PASC and a scientific assessment of the proposed service will be conducted.

The final outcome is expected mid-year.

What is MSAC?

The Medical Services Advisory Committee (MSAC) is an independent non-statutory committee established by the Australian Government Minister for Health in 1998. MSAC appraises new medical services proposed for public funding and provides advice to Government on whether a new medical service should be publicly funded (and if so, its circumstances) on an assessment of its comparative safety, clinical effectiveness, cost-effectiveness and total cost, using the best available evidence. Amendments and reviews of existing services funded on the Medicare Benefits Schedule (MBS) or other programmes (for example, blood products or screening programmes) are also considered by MSAC.
The MSAC also advises the Australian Health Ministers' Advisory Council (AHMAC) on health technology assessments referred under AHMAC arrangements.

News Flash

Alpha-1 Organisation Australia inc has been invited to give evidence at the Parliamentary Inquiry into approval processes for new drugs and novel medical technologies in Australia, following our submission.
https://www.aph.gov.au/Parliamentary_Business/Committees/House/Health_Aged_Care_and_Sport/Newdrugs

Aunty Alpha

Dear Aunty Alpha

I have had an irritated throat and husky voice for a while with difficulty speaking sometimes. My pulmonologist says I have silent reflux. What does this mean? Is this part of alpha-1 antitrypsin deficiency? Regards, Emma

Dear Emma,

These can be symptoms of “silent reflux”, or laryngopharyngeal reflux (LPR). Unlike gastro-oesophageal reflux disease (GERD), silent reflux causes few symptoms. Look for a bitter taste in your mouth, sore throat or burning sensation, difficulty swallowing, hoarseness, frequent throat clearing, chronic post-nasal drip. It is unclear if reflux causes breathing problems, or if breathing problems cause reflux, though lungs tend to become enlarged over years of heavy breathing causing them to press on the stomach enabling escape of acid into the oesophagus.

Evidence is emerging that there is a high incidence of LPR and GERD with asthma, chronic obstructive pulmonary disorder (COPD) and alpha-1 antitrypsin deficiency.

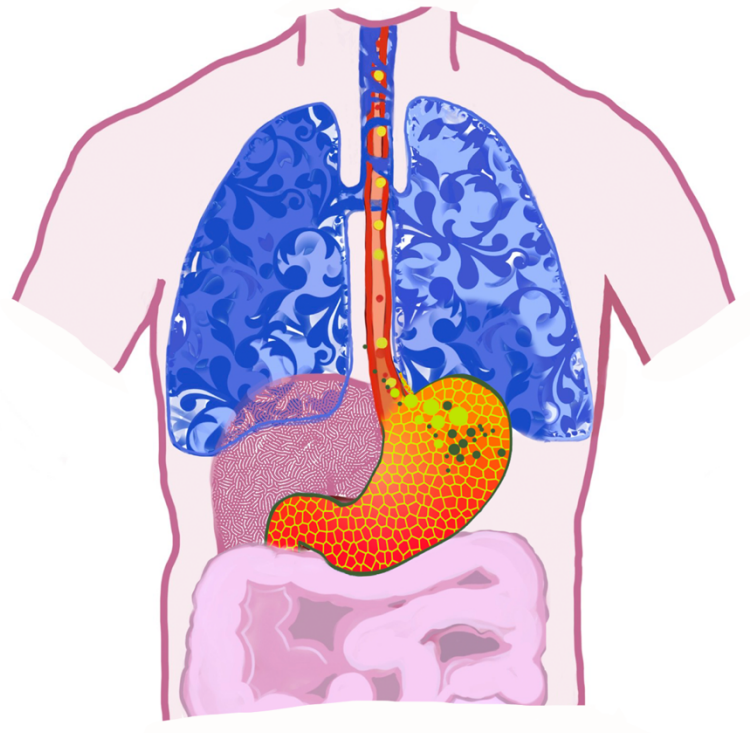
This can be dangerous as if left undiagnosed and untreated it can lead to oesophageal cancer. Your doctor may prescribe a proton pump inhibitor, PPI, to reduce the amount of acid produced in your stomach when you eat, and/or an H2 blocker, which also decreases the amount of acid in your stomach.

Lifestyle changes can help. Stop eating 3 hours before lying down. Elevate head and shoulders when you sleep. Lose weight. Quit smoking. Sleep on you left side. Reduce dietary acid; eliminate or limit carbonated drinks, alcohol, vinegar, fatty meals, citrus, tomato, spicy meals, chocolate, coffee, processed food (look for acidity regulator- this is hidden acid). A good resource to help with this is *The Acid Watcher Diet* by Dr Jonathan Aviv. Over time you may be able to cautiously reintroduce some of these foods.

Have a question about Alpha-1 Antitrypsin deficiency?

Write to Aunty Alpha at contactus.a1oa@gmail.com

Conditions such as alpha-1 antitrypsin deficiency that can lead to enlarged lungs, which in turn press on the stomach, can make sufferers more prone to silent reflux.



Antitrypsin and its Role in COVID-19

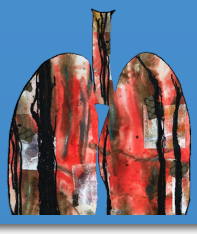
It is clear from the media and research that COVID-19 affects individuals differently. Most people develop mild to moderate illness and recover without going to hospital. Many though have not been so lucky.

Research published in *Genetics and Evolution* assessed the spread of one variant of the virus, D614G, which spread much faster in Caucasian populations than it did in Asia. One explanation for individual differences is linked to the protective protein *alpha-1 antitrypsin* and suggested that the slower spread of COVID-19 in Asia is due to fewer individuals with alpha-1 antitrypsin deficiency in Asian countries.

At a cellular level, the virus appears to use neutrophil elastase (the protein usually used to clear up bacterial infections in the lungs) to enter cells. Without adequate antitrypsin levels, neutrophil elastase levels are higher making it easier for cells to quickly take the virus into the body, enabling greater transmission in vulnerable populations.

With new strains spreading, Alphas need to be extra cautious as some strains, e.g., the UK mutation, are claimed to be deadlier. Such concerns support the argument that most people should be vaccinated against COVID-19 despite any temporary and unpleasant short-term side effects (e.g., headaches, fevers, flu-like symptoms). The research indicates that vaccines are safe and effective and although there have been reports of rare allergic reactions, the risk of a severe reaction is outweighed by the protection it offers to extremely vulnerable individuals.

The A1OA is thankful that the Australian Government has offered free vaccination to all Australians. In February, we wrote to the Minister for Health - the Hon Greg Hunt - outlining our concerns about vulnerable Alphas and asking that Alphas be considered a priority group for vaccination. We included in our letter that this pandemic supports the argument for subsidized augmentation therapy for individuals with severe alpha-1 antitrypsin deficiency. We await a reply.



An Alpha Story

Leonie Robison

As a child I was severely affected by asthma, being hospitalized and on oxygen several times. On one occasion, I was suffering from such a severe attack that when the family GP arrived, he was worried that if we waited for an ambulance I would not survive, so he drove me to hospital himself. I eventually "grew out of" my asthma and was symptom free from the age of 18 to 30. At that time, I started developing mild breathlessness, which necessitated starting on cortisone puffers. Over time my wheezing gradually worsened, leading to an ever-increasing dosage of cortisone. Not once in the following 32 years did any doctor ever mention alpha-1 antitrypsin deficiency and I assumed I had some unusual form of asthma. During this time, I also had a number of other symptoms, generally mild, which were hard to pin down to anything in particular. Most worrying of these was a persistent cough and irritated throat, which I put down to a combination of allergies and puffers. I started trying to find out why this was happening as I had reached the stage of barely being able to talk, and a lung doctor suggested, to my surprise, that I had reflux. I started medication for this, and it is now reasonably well under control although I still have problems with a husky voice and talking on the phone. I now know that I am a ZZ alpha and am managing reasonably well.