alpha-1 ORGANISATION AUSTRALIA INC

Alpha Times

Newsletter of Alpha-1 Organisation Australia inc *Issue 9 Winter 2022*

From the President's Pen

Dear Friends,

The key issues of trustworthy *information*, *individual support* and *advocacy* are uppermost in my thoughts this month, especially with the recent change of government which brings renewed hope of affordable treatment. At Alpha-1 Organisation Australia we hope that one day soon, Australians with severe antitrypsin deficiency will have access to subsided augmentation treatment (also known as Alpha-1 proteinase treatment). To support this aim, I recently wrote to the new Minister for Health – Hon. Mark Butler – outlining the urgent need for government recognition of the existing evidence, and the benefits associated with treatment access.

During the last few months, I had the pleasure of providing input into the design and development of the Alpha-1 Antitrypsin Deficiency webpage for the Australian ***RARE portal*** – an Australian web portal being developed by Rare Voices Australia (RVA) – which will provide information about many rare diseases, and will include links to other resources e.g. evidence-based charity websites, across Australia.

To support people diagnosed with Alpha-1 antitrypsin deficiency I am currently participating in the *Mental Health First Aid* course, which leads to a Certificate of Accreditation. The knowledge and skills derived from the program further enables our charity to provide support to people who may be experiencing anxiety, depression or other mental health issues linked to their diagnosis, by providing better initial support to someone who is developing a mental health problem or experiencing a mental health crisis. Our partnership with Rare Voices Australia (RVA) is supporting this work.

My advocacy work includes advocating to bring Alpha-1 clinical trials to Australia. During June, I contacted Inhibrx to encourage the company to bring the INBRX-101 trial here. This trial shows great promise using recombinant therapy - monthly dosing - to raise antitrypsin levels to normal protective levels.

During winter I hope that you all manage to keep warm and safe and can avoid lung irritants from indoor heating e.g. noxious gases from gas heaters, or gases and particles from indoor wood burners. Remember, even well maintained, flued heaters emit some lung irritants into the room. Outdoor and indoor cold and dry air can be hard on individuals too as it irritates the airways and can cause sinus problems. I will be wrapping a scarf around my nose and mouth, monitoring weather forecasts, and using saline nasal spray to avoid nose bleeds this winter. If you have tips on staying well during winter, we would love to hear them via contactus.a1oa@gmail.com. Don't forget, you can reach out anytime if you would like to chat to someone who understands what it is like to be diagnosed with or is living with Alpha-1 antitrypsin deficiency. We are here to support you.

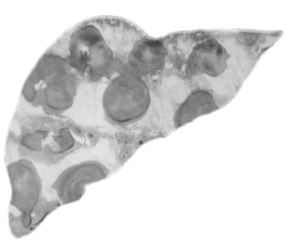
All the best
Gaynor

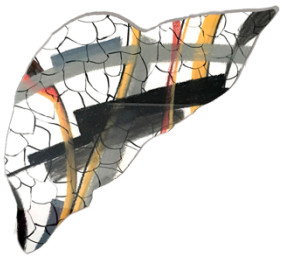
The Alpha-1 Foundation 2022 Hybrid Alpha-1 National Conference took place June 8-11, 2022

Many important topics relating to Alpha-1 were covered. Summaries of these can be found in the coloured boxes below with some extra points.

Some notes on Alpha-1 Antitrypsin Deficiency and Liver

- Alpha-1 antitrypsin (AAT) is made in the liver.
- AAT stops neutrophil elastase from destroying lung tissue. Some individuals with Alpha-1 won't develop emphysema, possibly through the inheritance of other protective genes.
- People with Alpha-1 Antitrypsin Deficiency (Alphas) have a risk of liver cirrhosis, which can shorten their lifespan, although many Alphas never get cirrhosis.
- ZZ genotype has a high risk of liver disease and MZs possibly also.
- Liver disease (fibrosis and cirrhosis) can be diagnosed by
    - Initially by checking liver enzyme levels
    - Liver function blood tests
    - Liver ultrasound, fibroscan or CT scan
    - Liver biopsy
- Alphas can have cirrhosis with no symptoms.
- Alphas can have increased risk of portal hypertension, liver cancer and liver failure (rare).
- Some doctors suggest ZZ Alphas have annual liver function tests and liver ultrasounds after age 50.
- If you have liver disease, the chance of developing portal hypertension is high. There can be an enlarged spleen, a low platelet count, ascites (fluid in the abdomen), and lack of mental clarity due to toxins not being cleared by the liver.
- Oesophageal varices are another hepatic hypertension complication. Scar tissue can interfere with blood flow through the liver so blood flow seeks easier paths, e.g., stomach veins, which can cause swellings in the oesophagus lining, which in turn can bleed.
- Alphas should try to minimize liver damaging lifestyles or infections, e.g., limit alcohol, maintain a healthy weight, try to avoid developing fatty liver by eating a healthy diet, be careful with herbal supplements and medicines that may harm.
- Screen and vaccinate for liver damaging viruses, e.g., Hepatitis A and B.





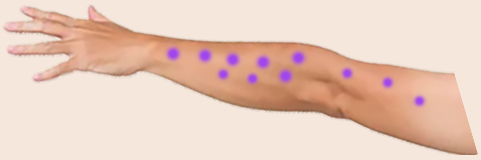
Tips for Everyday Living as an Alpha

- Join local support groups or Join Alpha-1 Organisation Australia www.a1oa.org.au.
- Sign up to educational events, online webinars offered by various organisations.
- Find hobbies, entertainment and jobs that will not cause lung irritation, that you can do as an Alpha, depending on the severity of your condition.
- Move to a lower altitude, if possible, for easier breathing.
- Avoid smoking cigarettes, marijuana, vaping.
- Exercise, strength train, attend lung rehab.
- Try to avoid exacerbations and lung infections. Have rescue antibiotics ready.
- Keep items within easy reach if you severely affected to make everyday tasks easier.
- Have oxygen where you are most likely to need it, use as prescribed.
- Limit alcohol intake.
- Get a good night's sleep.
- Frequent, small meals to reduce pressure on diaphragm if lung affected, and for less oxygen to be diverted to digestion. Alphas have a tendency to reflux. If liver affected, eat small meals every 2-3 hours to limit muscle protein breakdown.

- Speech therapy can help with e.g., paradoxical vocal cord movements, and swallowing.

Necrotising Panniculitis

- Inflammation of the fatty layer under the skin, leading to death of cells there.
- Rare complication of Alpha-1, occurs in about 1 in 1000.
- Painful lesions with bumps under the skin that can burst open and leak oily fluid.
- Occur mainly on the arms and legs but can also occur on the abdomen and back.
- A punch biopsy is needed to confirm diagnosis.
- Exact cause unknown but thought to be inflammation and blockage of blood vessels or deposits of polymers.
- There are many treatments, e.g. antibiotics, corticosteroids, NSAIDs, but the best is augmentation therapy to restore the patients Alpha-1 Antitrypsin levels.



Alpha-1 Testing

Who should get tested?

- Anyone with adult-onset asthma.
- Anyone who has never smoked but has COPD or emphysema.

What test do I need?

- Firstly, you will have a blood test to determine your alpha-1 antitrypsin level. If this is found to be low, then
- Secondly, you will have another blood test to find out your genotype (sometimes referred to as phenotype test).

I have been diagnosed with Alpha-1 Antitrypsin Deficiency. Should my family get tested?

- Siblings and children should be tested.

Are all genotypes tested for?

- Only the most common alpha genes are tested for. Rarer genes may need further testing.

Vaccination

- Vaccination for Hepatitis A and B for people on augmentation therapy is no longer recommended as no cases have been recorded in these individuals.
- Annual Influenza vaccination is recommended.
- Pneumococcal vaccine is recommended.
- Shingles vaccination with Shingrix vaccine is recommended for over 50s- 2 doses 6 months apart.
- Covid 19 vaccine is currently recommended for everyone who is eligible, regardless of Alpha-1 status. If Alphas contract Covid 19 then early access to antiviral drugs, either Lagevrio or Paxlovid is recommended.

Caregiving

- Caregiving can be isolating and stressful.
- Caregivers need to take time to look after themselves and have things to look forward to. They should do something for themselves every day, read, go for a walk, watch a movie.
- Caregivers need to look after their own health, set boundaries and know when to say no.
- Social support is essential. Make sure to keep in touch with family and friends.

Stressed caregivers can show the following signs:

- Tired but can't sleep
- Sleeping too much or too little
- Feeling overwhelmed
- Having trouble focusing or remembering
- Feeling isolated
- Not enjoying activities they previously liked
- Feeling worried, sad, or angry all the time
- Feeling resentful

Things that could help caregivers:

- Breathing and grounding techniques can help.
- Living in the moment. Avoiding unhelpful or negative thoughts that can overwhelm you.
- Find things you can change to improve the situation.
- Join a support group for caregivers.

Do you have a question about any of the issues raised?

Write to Aunty Alpha, who is always happy to answer your questions or contact the A1OA through contactus.a1oa@gmail.com

Aunty Alpha

Dear Aunty Alpha,
I am a ZZ Alpha. I have recently developed some lumps on my skin that are sore to touch as well as some purplish patches. These are all mainly on my arms and legs. Could this be panniculitis?
Regards
George

Dear George,
That does sound like it could be panniculitis, which can be a complication of Alpha-1. A visit to your doctor will confirm this for you via a biopsy.
Panniculitis symptoms are typically:

- Large bumps (nodules) that are tender to touch and can vary in size
- Bumps on the legs, feet, hands, and arms
- Bumps that produce an oily fluid
- Sometimes bruising or patches of broken skin can occur

Available treatments such as tetracycline antibiotics can be helpful but the best results are obtained with Augmentation Therapy, which currently is not subsidised in Australia for lung affected Alphas but may be prescribed for panniculitis, or privately purchased to preserve lungs if the sufferer meets the Special Access Scheme criteria.
Regards
Aunty Alpha

Development in Access to Augmentation Therapy

Grifols Private Pay Access Pathway Program

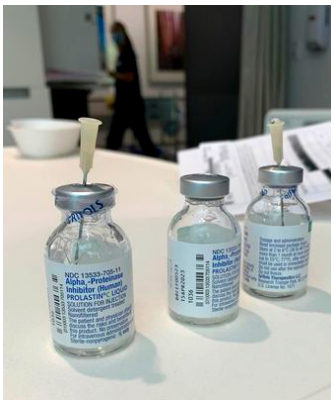
Following discussion with Grifols, the company now offers a private pay access pathway program, allowing eligible Alpha-1 patients (typically PiZZ) to privately purchase augmentation therapy from Grifols via a pharmacy.

This is a great breakthrough as plasma supply from CSL Behring has been disrupted in recent times. To access the program, specialist centres (e.g., hospitals / infusion lounges) or general practices can be approached to organise access to infusions. Some medical insurance schemes will cover the cost of a private hospital infusion lounge, but the cost of Prolastin-C will most likely not be covered. Infusion costs will differ by location e.g., a GP surgery may charge a fee for the infusion nurse/equipment, but a private hospital infusion lounge may be covered by private insurance.

The recommended dosing regimen of Prolastin-C is 60 mg/kg body weight by IV infusion administered once weekly. The current cost of one vial of Prolastin-C is just under $500. Since dosage is weight related, private purchase may not be an option as most people would be looking at $1,500+ per week.

To access the program, an application to the Special Access Scheme (at the TGA) is made by your respiratory specialist. Approval is usually very quick, and once approved the nominated supportive pharmacy can order the product on the patient's behalf, and arrangements put in place to have the product taken to the infusion location, ready for the patient's visit.

A few NSW Alpha-1 patients have started using this pathway e.g., are receiving infusions at a private hospital infusion lounge and at a general practice. If you would like to chat about this private purchase pathway, please contact us at contactus.a1oa@gmail.com.



An Alpha Story

Ian Saunders

While we're all different and are certainly affected by A1AD to different degrees, from what I've read my story is similar to many. Diagnosed as a ZZ with COPD/Emphysema in my early 50s (after a misdiagnosis of late-onset asthma) I've suffered from a slow and steady decline in my lung function and am now in my early 60s battling to maintain a reasonable lifestyle and delay the need for a lung transplant – as some of you may know this is itself a double-edged sword, since the older you are – and increasingly from the mid-sixties – the more reluctant they are to carry out a transplant.

Luckily I was able to get on the Grifols clinical trial for their augmentation therapy (Prolastin); it was meant to be three years of either a single dose, a double dose or a placebo, but as my lung function continued to decline rapidly during the first year of the trial, it was stopped for me after year two – the positive being that I was then given the therapy for two years, during which time my lung function stabilised and according to some measures even improved marginally, leaving me convinced such therapy works. Of course, all good things come to an end, and for the last 3 years my lungs have continued to get worse after ceasing treatment.

Covid permitting I have also been lucky to be able to attend a pulmonary rehabilitation gym for the last few years, and there is no doubt in my mind regular visits and associated exercise have slowed the decline in my lung function – despite my pre-existing belief that exercise is a dangerous activity 😉.

Of course, the other impact of living with a condition such as A1AD is on the mental state – while we're all living with the terminal condition called life, knowing that we have a condition that will only accelerate the inevitable can at times be a tad depressing, and it's important to be aware of this and to seek the appropriate support if needed to stay positive and to continue to be as active and engaged as possible.

Finally, I try and do what I can for the future generations of A1AD patients (including my son who will be an MZ) by contributing to the A1OA charity, in the hope that not only available treatments will be made generally available, but also that one day soon one of the many 'cures' will prove itself to be effective.

I wish all my fellow Alphas all the very best on their particular journey and would encourage everyone to ask for information and help to ensure their best possible outcome.