alpha-1 ORGANISATION AUSTRALIA INC

Alpha Times

Newsletter of Alpha-1 Organisation Australia inc

Issue 10 Spring 2022

From the President's Pen

Hi everyone
Our volunteers have been busy in recent months getting ready for November *Alpha-1 Awareness Month.* We have a special project underway which will be launched during November 2022 so keep an eye out. We have also developed some wonderful new resources to support people with Alpha-1 antitrypsin deficiency (Alpha-1), including resources for doctors. We have distributed these resources to hundreds of Australian doctors who are likely to be managing people with Alpha-1, as discussed in this edition. These are available for download from the Alpha-1 Organisation Australia website - Alpha-1 Organisation Australia | Resources (a1oa.org.au). So, word is getting out about Alpha-1 across Australia.
Sometimes there are announcements for research grants that aren't directly related to Alpha-1 but have huge relevance e.g., the recent announcement by *GrantConnect* (Australian Government) on the NHMRC-NIHR *Collaborative Research Grant Scheme*. This grant will support an international collaboration on various lung related topics including rescue packs in chronic obstructive pulmonary disease (COPD). As many Alpha-1 patients have COPD (e.g., emphysema) and have previously been prescribed medication to manage flare-ups (e.g., an exacerbation) as part of their agreed COPD management plan, it will be interesting to see how the research results impact Australian COPD action plans and the appropriate use of rescue packs, a topic that has been in the media in recent times.
As the weather warms up (it still feels like Winter in many parts of Australia) I hope that everyone finds time for some enjoyable simple quality-of-life activities.
Wishing you all the best,
Gaynor Heading
President A1OA

A1OA New initiatives

New products and a mental health first aider.

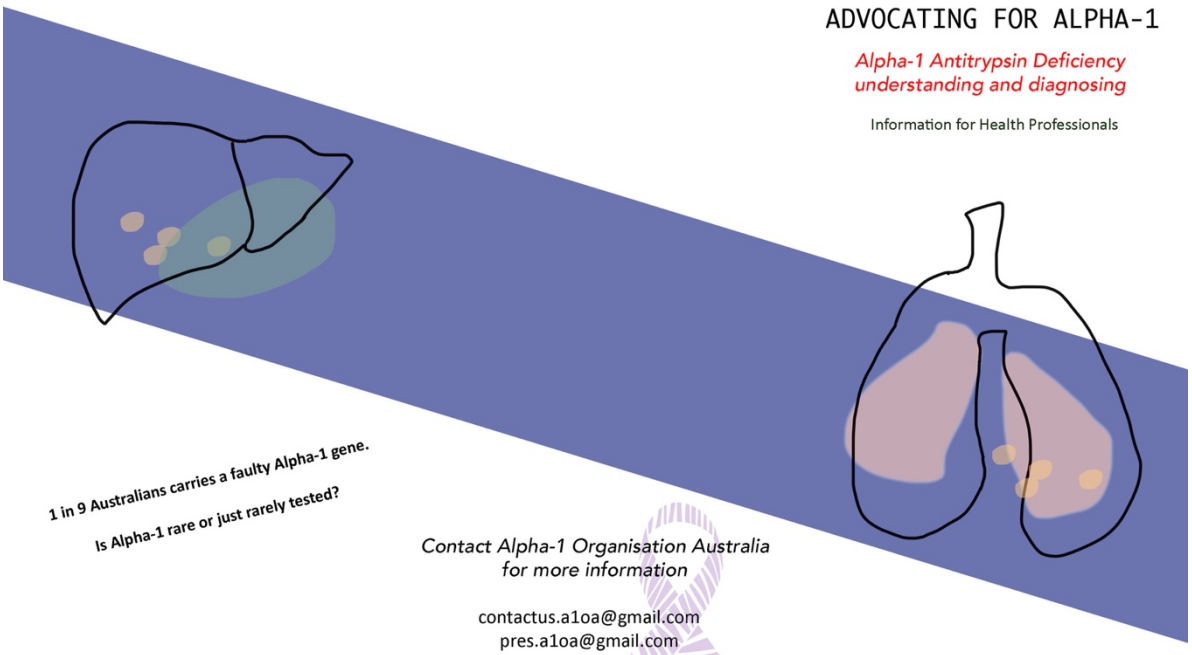
Firstly, we developed brochures for medical practitioners and patients, and emailed or posted these to around 300 lung specialists and GPs with an interest in lung health. These are double sided trifold brochures and contain tips for doctors on what to look out for in patients who are potentially Alpha-1 affected, to obtain a diagnosis of Alpha-1 Antitrypsin Deficiency, and hints for patients suffering specific symptoms. See these below.

We also developed small portable double-sided cards with some simple questions and answers that can be given to someone asking about Alpha-1. We call these "wallet cards" as they are the size of a credit card or business card and fit easily into a wallet. These can also be seen below.

The brochures and wallet cards are available to be downloaded from our website *Resources* page to be printed. This is best done through an online professional printing service but could be done "at home" if you have the right resources.

https://www.a1oa.org.au/resources/

Brochure for Health professionals to aid in diagnosis of Alpha-1



ADVOCATING FOR ALPHA-1

Alpha-1 Antitrypsin Deficiency understanding and diagnosing

Information for Health Professionals

1 in 9 Australians carries a faulty Alpha-1 gene.

Is Alpha-1 rare or just rarely tested?

Contact Alpha-1 Organisation Australia for more information

contactus.a1oa@gmail.com
pres.a1oa@gmail.com

or see the fact sheets on
www.a1oa.org.au

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Alpha-1 Antitrypsin Deficiency (A1AD)

A1AD is a genetic condition, caused by a mutation in the Serpina 1 gene on chromosome 14.
There is a large number of variants with a small number of these causing medically significant problems. The two most common of the latter are designated Z and S, with M being normal. Other rare genotypes are hard to detect as the usual gene test only looks at Z and S, so must be ascertained by gene sequencing.

The most common symptom of A1AD is emphysema, which becomes noticable in adults who were previously thought to be healthy, usually between 30 and 50 years of age. This is often mistaken for adult-onset asthma or early onset COPD.
Any patient presenting in this way should be tested for A1AD.

The gene coding for the Z variant causes a misshapen molecule of alpha-1 antitrypsin to be produced in the liver, which then becomes trapped therein and can lead to cirrhosis or lung cancer when it builds up to toxic levels.
Patients with unexplained liver problems should be tested for A1AD.

Estimates suggest that 10% or fewer of people with a severe deficiency in alpha-1 antitrypsin have been diagnosed.

Does your patient have unexplained emphysema?
Does your patient have adult onset asthma?
Does your patient have unexplained bronchiectisis?
Does your patient have recurring lung infections?
Does your patient have a family history of breathing problems?

Have you considered alpha-1 antitrypsin deficiency?

Does your patient have unexplained cirrhosis of the liver?
Does your patient have abnormal itching, a symptom of liver disease?

Have you considered alpha-1 antitrypsin deficiency?

This genetic condition is thought to be the most common rare disease, but is often not considered or not included in diagnostic tests when diagnosing patients.

Lung function tests and clinical examination alone cannot identify Alpha-1.
It is important to have an alpha-1 antitrypsin bloodtest. to determine the level of alpha-1 antitrypsin.
If found to be low this should be followed up with a genotyping test.

Contact Alpha-1 Organisation Australia for more information.

contactus.a1oa@gmail.com
or see the fact sheets on
www.a1oa.org.au

Levels of alpha-1 antitrypsin expected with different genotypes

MM	Normal
MS	80% normal
MZ	60% normal
SS	60% normal
ZZ	10% normal
NullNull	zero

Individuals deficient in the protein, alpha-1 antitrypsin, lack protection in the lungs against the enzyme neutrophil elastase, which flares up with infections.

Those at risk should be prescribed antibiotics at the first sign of infection.

ADVOCATING FOR ALPHA-1

Why test for Alpha-1?

Patients will benefit from:
Ceasing smoking
Avoiding lung irritants
Immunisation
Lung Rehabilitation
Clinical trial participation
Rapid treatment of lung infections
Health lifestyle - exercise, healthy diet and weight
Family and peer support
Health monitoring

Brochure for patients for display in Doctor's surgeries

Alpha-1

Why to test?

Affected individuals need to know they will benefit from:

Ceasing smoking
Avoiding lung irritants
Immunisation
Lung Rehabilitation
Clinical trial participation
Rapid treatment of lung infections
Healthy lifestyle - exercise, healthy diet and weight
Family and peer support
Health monitoring

Alpha-1 Antitrypsin Deficiency

Short of breath?

Do you have unexplained shortness of breath, COPD, or adult onset asthma?

Itchy?

Do you have unexplained liver problems or itchiness?

These can be symptoms of the genetic condition Alpha-1 antitrypsin deficiency.

Ask your doctor for a blood test if this sounds like you.

Contact Alpha-1 Organisation Australia for more information

contactus.a1oa@gmail.com
pres.a1oa@gmail.com

or see the fact sheets on
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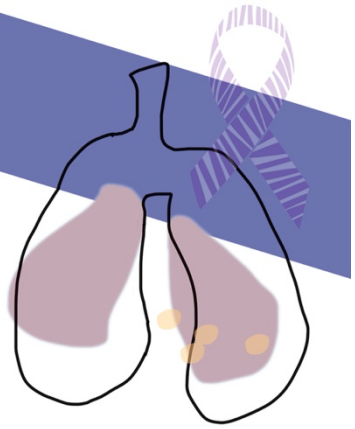

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The most common symptom of A1AD is emphysema, which becomes noticable in adults who were previously thought to be healthy, usually between 30 and 50 years of age. This is often mistaken for adult-onset asthma or early onset COPD.

Smoke and Dust

Lung irritants can cause rapid lung destruction leading to emphysema so should be avoided.

Individuals deficient in the protein alpha-1 antitrypsin lack protection in the lungs against the enzyme neutrophil elastase. This enzyme flares up with infections and breathing in lung irritants, causing further lung destruction.

Those at risk should be prescribed antibiotics at the first sign of infection.

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a1ad

Wallet Cards

alpha-1
ORGANISATION AUSTRALIA INC

Together making a difference

FInd out about our organisation and Alpha-1

www.a1oa.org.au
contactus.a1oa@gmail.com

Alpha-1 Organisation Australia is a registered charity
ABN: 89131526593

What is Alpha-1 Antitrypsin Deficiency? (A1AD)

1 **Genetic lack of a protein,** (mostly made in the liver.) It's main function is to protect lungs. Other organs can be affected.

2 **What are the symptoms?** A1AD mainly affects lungs (emphysema, COPD, often misdiagnosed as asthma) and liver (fatty liver, cirrhosis, liver cancer).

3 **Should I get tested?** Talk to your doctor if you have adult-onset asthma or emphysema but never smoked, or you have unexplained fatty liver or raised liver enzymes.

4 **How is it tested?** A simple blood test to determine your antitrypsin level, followed by a genetic test if your level is low.

Mental Health First Aid

MENTAL HEALTH FIRST AIDER™ ACCREDITED

Alpha-1 Organisation Australia now has an accredited Mental Health First Aider who is ready to help if you are not coping after a diagnosis of A1AD for yourself or a family member. A new diagnosis can cause mental distress, anxiety, or depression. Please reach out to mentalhealth.a1oa@gmail.com

Recently Diagnosed? Looking for information?

The website of A1OA has many resources to help you: information brochures and fact sheets, and downloadable posters. https://www.a1oa.org.au/resources/

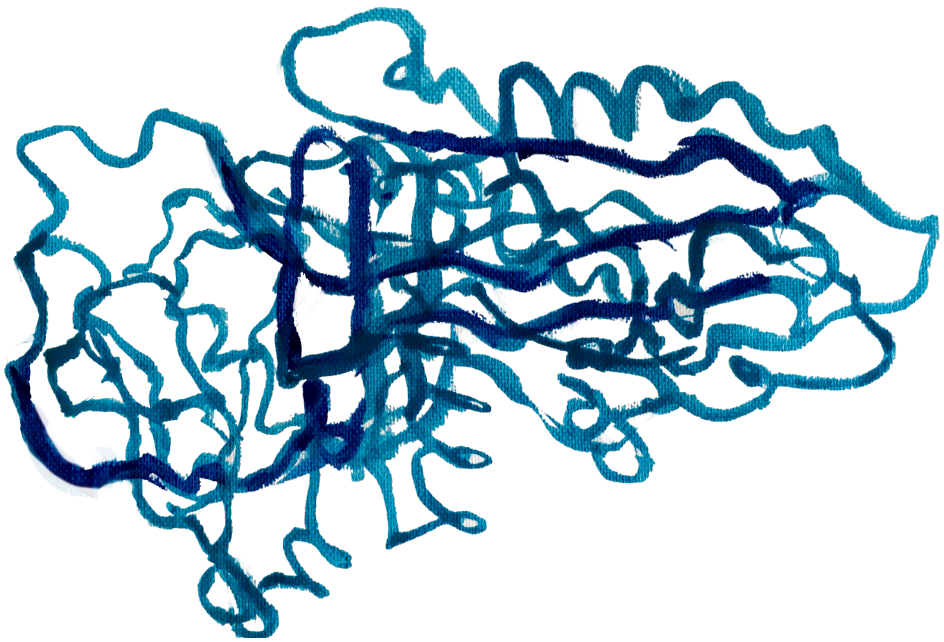
Here are two easily read articles that might also be of help:

American Thoracic Society Patient Education Series: MZ Alpha-1 Antitrypsin Deficiency (April 2022) PDF

This article describes inheritance of Alpha-1 Antitrypsin Deficiency (A1AD)- the gene being passed from parent to child. People with A1AD have decreased levels of a protein called Alpha-1 Antitrypsin (AAT) in their blood, that is they have a deficiency. Low levels of AAT can lead to lung and/or liver problems. There are different types of A1AD and each of these carries different degrees of risk. This fact sheet describes the MZ form of A1AD.

StatPearls Continuing Education Activity: Alpha 1 Antitrypsin Deficiency (19 July 2022)

In this article, read about the clinically under-recognised genetic disorder, A1AD, that causes the production of defective alpha-1 antitrypsin protein. AAT protein protects the body from the neutrophil elastase enzyme, which is released from white blood cells to fight infection. The article is written as an educational activity discussing the evaluation and management of A1AD and highlights the role of the inter-professional team in managing and treating the condition to improve patient care.



Artist's interpretation of the complex alpha-1 antitrypsin molecule

Aunty Alpha

Dear Aunty Alpha,
I have been getting frequent headaches, often to the point where I experience “brain fog”, eye issues, dizziness, nausea, making it difficult to function in my daily life. I have been using asthma puffers for a number of years, with an ever-increasing dose as my lungs worsen with Alpha-1. Are these headaches migraines and could there be a correlation between the headaches and puffer use as the frequency and severity of the headaches seems to have increased along with the increased use of my puffer.
Regards
Shayne

Dear Shayne,
Your headaches sound like migraines, which as you have experienced can have an impact on the sufferer’s ability to function normally.
You don’t specify what types of inhalers (puffers) you are using. Inhalers fall into three main categories- relievers, preventers, and mucous-reducing anticholinergics.

It is the relievers that can cause migraine. This is a known side-effect along with nervousness, shakiness, throat irritation, muscle aches. More serious, though less common, side effects include a rapid heart rate, or fluttering or pounding heart.

All medications have side effects, which are generally mild and considered acceptable as they are outweighed by the benefits of the drug. Sometimes, though, the side effects can be more severe such as in your case. You may need to adjust your dosage or try a different brand or type of puffer. When making changes of this kind, always consult with your GP or specialist.

Regards
Aunty

Examples of reliever “puffers”

A second opinion

It has been said that a second opinion may help someone with a rare disease, particularly, when there are few or no treatment options. Another opinion (not an internet search) could in fact, provide information, answers and management / treatment options which could help delay disease progression and improve quality of life and be worth the additional costs.
Some of the conversations that A1OA board members have recently had with patients and specialists indicate that there are a lot of unanswered questions among Alpha-1 patients, such as:

- Which inhaler/s would be best for me and what dose?
- Is nebulising 3% saline (opposed to 6% which may be considered too strong) better in its dual beneficial role in damaging the biofilm on bacteria breathed in everyday, while acting as a mucolytic agent?
- Should I have a 4D chest image, where a CT scan is used in combination with fluoroscopic screening (a specialized type of x-ray imaging procedure) to produce moving images of the lungs to see if one lung is working harder than the other, if the diaphragm is moving and if there is less air movement due to loss of lung elasticity?
- Should I consider lung volume reduction using valves or “glue”?
- Is the evidence on lung glue “in”?
- Would I benefit from thrice-weekly Azithromycin (e.g., for chronic bronchitis, bronchiectasis, emphysema), due to its immunomodulatory properties that inhibit pro-inflammatory cytokine production and inhibits neutrophil influx?
- Are any off-label treatments beneficial (i.e., a drug not approved for one’s condition by the Therapeutic Goods Administration)?
- What tests should I have to check/monitor for heart failure (associated with COPD)?
- When walking (with COPD but not on supplemental oxygen) and my heart rate goes high (sinus tachycardia), should I stop walking or need to see a heart specialist?
- How often should I have a sputum test if I have had a history of serious lung infections?
- Is regular liver function monitoring required?
- If I have high liver enzymes, what should I do?
- Are my bones thin and if so, what should I do?
- Could any complementary medicines be useful e.g., collagen?

Seeking a second opinion is common and within your rights. In an era of information overload, emerging evidence, and new technologies, a second opinion may be just what the doctor ordered.

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

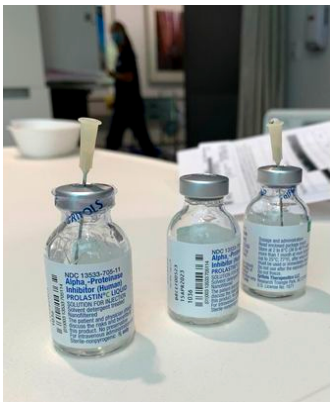
NEWS FLASH

RESPIRATORY DISEASE MEETING – Including Alpha-1 Antitrypsin Deficiency (A1AD).

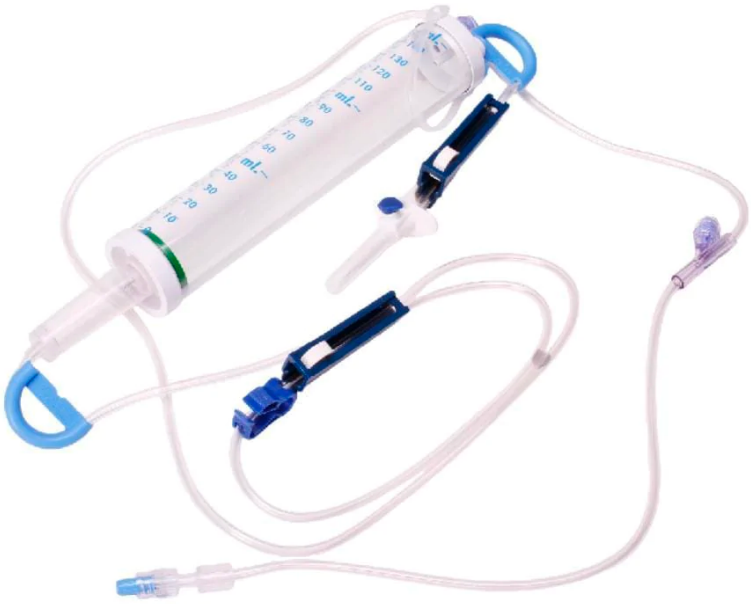
The 16th Annual NSW Asthma Meeting (titled NAMe 16) is being held November 17th-18th 2022. The meeting will cover all aspects of respiratory diseases, convening sessions inclusive of COPD, COVID-19, pulmonary fibrosis, A1AD, immunometabolism and diet and microbiome interventions amongst many others.

The good news is that there is an online Zoom participation option, and it is FREE to register – you just need to register using the link below.
NAMe Meeting 2022 | Centenary Institute

If you wish to attend in person it is being held at the Centenary Institute NSW. The full program will be announced in the coming weeks.



Vials of alpha-1 antitrypsin ready for infusion into a patient



Burette and tubing for infusing