alpha-1
ORGANISATION AUSTRALIA INC

# Alpha Times

Newsletter of Alpha-1 Organisation Australia inc *Issue 7 Summer 2021-22*

## From the President's Pen

Welcome to our Summer Edition.

Alpha-1 antitrypsin deficiency is one of more than 7000 known rare diseases. Each year Rare Disease Day is observed on 28 February with rare disease communities around the world showing their colours and stripes- purple is for Alpha-1 in conjunction with zebra stripes, the international symbol for rare disease.

Rare disease day raises awareness with the general public and decision-makers that people living with a rare disease need affordable access to treatment and that rare disease negatively impacts peoples' lives in many ways. Despite relentless advocacy for government subsidised augmentation therapy, which is the only treatment available for Alpha-1 lung disease, Australians continue to be denied subsidised augmentation treatment and have unmet needs. We therefore, encourage you to be courageous this year and show your stripes on 28 February.

Put your hands up!
Show your colours for Rare Diseases Day!

## Riding for Awareness

Mark and Matt will be riding from Sydney to Melbourne!

See next page for article

*Mask design by Leonie Robison*

## Riding for a Cause

Mark Lloyd is organising this fundraiser on behalf of Alpha-1 Organisation Australia Incorporated. Mark and his friend Matt Nicholas will be cycling from Sydney to Melbourne over the course of just under 2 weeks to raise awareness of Alpha-1 Antitrypsin Deficiency and funds for Alpha-1 Organisation Australia Inc. There is now an increased urgency to raise awareness around the condition due to COVID-19.

This is an epic ride of almost 1000 km! The route will take our intrepid riders through two Australian states- New South Wales and Victoria, through the towns of Sydney - Narellan - Goulburn - Yass - Cootamundra - Wagga Wagga - Albury - Whitfield - Mansfield - Yea - Coburg - Melbourne- and will involve them in many mental and physical challenges.

The two will be carrying all their supplies with them on the bikes and will have no support crew.

Where possible, they will be staying in local towns but also plan to camp in the more remote areas.

They have both put a huge effort to get themselves cycling fit as they will be averaging around 100km per day throughout the trip!

They are hoping to raise $10 000 through their efforts and are already well on the way with this.

If you would like to support these two courageous cyclists, you can donate through their GoFundMe page:
https://www.gofundme.com/f/the-sydney-to-melb-alpha1-awareness-cycle?utm_campaign=p_lico+share-sheet&utm_medium=copy_link&utm_source=customer

You can also follow their adventures through this link:
https://www.instagram.com/marklloyd600/

### Happy Riding Mark and Matt!

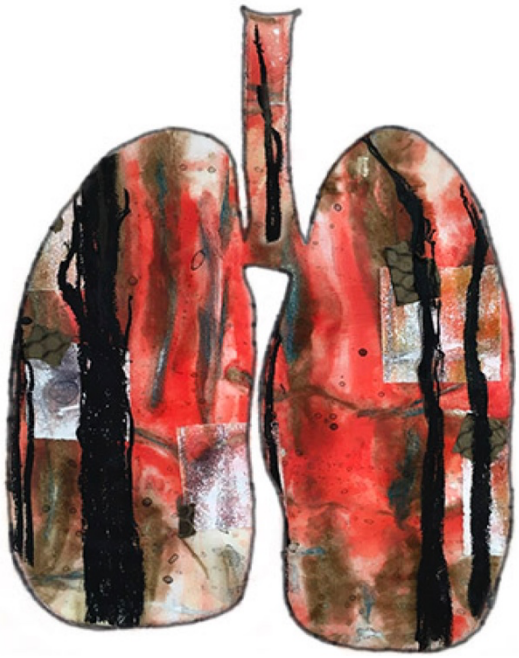
## Snapshot on Alpha-1 Antitrypsin Deficiency

- Alpha-1 is a genetic disorder leading to serious lung and liver disease
- Is caused by an alteration (mutation) in the gene that makes antitrypsin
- Approximately 11% of Australians could have a faulty gene
- Disease can occur when antitrypsin doesn't reach the lungs to protect them or if antitrypsin becomes stuck in the liver. Other organs can be affected.
- Most individuals have not been diagnosed or have been misdiagnosed with adult asthma or COPD
- Researchers are looking for a cure and clinical trials are underway
- Identifying individuals with one or two faulty genes in the gene pair can help them to choose a low risk lifestyle to protect their lungs and liver, to seek medical help at the earliest sign of infection, and to monitor their health for signs of deterioration
- A simple blood test can detect your antitrypsin level and a genotype test can indicate your level of risk for lung and liver disease

## Summer is bushfire season.

### Look after your lungs

Stay away from smoke and fires.



## New Trial for Alpha-1 Related Liver Disease

Hope is important for Alpha-1 sufferers, and we were delighted last year to be able to announce the availability of a new Alpha-1 liver trial for PiZZ patients – the Estrella Trial, commencing this year at St Vincents' Melbourne. This Alpha-1 study is one of many being undertaken internationally looking at potential new treatments and possibly a cure for Alpha-1.
To assist people to determine their eligibility for trial inclusion and make a decision about whether to apply or not, we have included additional information about the Estrella trial in this newsletter.
Such studies offer hope to all Alphas.
There is growing recognition that hope and optimism are important in chronic illness and that positive psychological functioning influences health. There is evidence that individuals with greater optimism and hope engage in healthier behaviours, regardless of how sick they are. Such behaviours could relate to healthy eating, exercise or activities targeting mental health e.g. regular mindfulness or meditation. If you have been diagnosed with Alpha-1 and have hope please contact us and share your story of hope, which may inspire others.

### The ESTRELLA Clinical Study

It is always exciting when a company begins an investigation into providing a potential therapy for diseases. The Estrella Clinical Study is part of Dicerna Pharmaceuticals, Inc.'s clinical development program called SHINE. Dicerna is investigating whether its study drug, belcesiran, is safe, well-tolerated, and will stop the liver disease from progressing for those Alpha-1 PiZZ persons who are liver affected. Belcesiran is a subcutaneously administered GalXC™ ribonucleic acid interference (RNAi) investigational therapeutic and is designed to reduce the build-up of abnormal alpha-1 antitrypsin (AAT) in the liver. Belcesiran is designed to target the gene responsible for production of abnormal AAT protein to reduce production of this protein in the liver. Unlike gene therapy and gene editing, Dicerna's GalXC RNAi investigational therapies are fully reversible after cessation of treatment. Belcesiran has completed the dosing and treatment period in a Phase 1 clinical study and follow-up monitoring is continuing. In Phase 1, the safety of the drug candidate is assessed in healthy volunteers. Belcesiran is now undergoing Phase 2 clinical study, which tests the safety and effectiveness in patients living with the disease being studied. The Phase 2 ESTRELLA clinical study is a double-blind study of belcesiran compared to placebo in patients with AAT deficiency-associated liver disease (AATLD) in which participants will be randomly assigned to receive either placebo or belcesiran. The double-blind nature of this trial means neither trial participants nor the clinicians running the study know which intervention (placebo or belcesiran) participants are receiving.
In the ESTRELLA trial, participants will receive multiple doses of belcesiran or placebo and will be followed after administration of the last dose. There is a screening component to the study, which will determine eligibility in which the duration of the screening testing period will depend on the availability of the staff and equipment at the study site. The study includes two liver biopsies, the first of which is completed during screening and the second after the 24- or 48-week treatment period. The treatment period of 24 or 48 weeks is assigned in a computer -generated alternating manner at the time a participant is confirmed as eligible for the study. Participants will be required to make multiple visits to the study site during the treatment period. These site visits will include a visit to receive an initial injection, three visits for health

checks after receiving the first injection, and 6 or 12 additional visits depending on the study treatment group to which participants are assigned. The second liver biopsy will occur at the last study visit. The study doctor will explain all the tests that will be required and any preparations to make prior to each visit, such as fasting before a test.

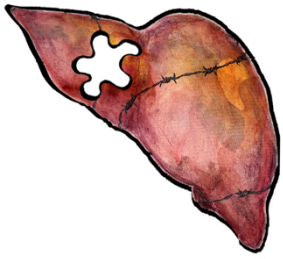
You may be eligible to participate in the ESTRELLA trial if you:

- Are male or female, aged 18 to 70 years old
- Have a confirmed diagnosis of alpha-1 antitrypsin (AAT) deficiency and AAT deficiency-associated liver disease (AATLD)
- Have been tobacco-free for at least three months and are willing to remain tobacco-free for the entire study
- Agree to undergo liver biopsies (at screening and after receiving the last dose in the study
- Are genetically confirmed as PiZZ
- Meet other study-specific criteria.

The principal investigator for this trial is Dr. Matthew Conron, and his study site coordinator is Sue Brenton at St. Vincent Hospital in Melbourne. Reimbursement plans include travel, meals, lodging and a stipend based on a set of requirements and financial/medical needs. If eligible for the study, participants should be sure to discuss any reimbursement questions with the study coordinator.

Other helpful information:

- There are no planned overnight hospital stays
- Patients in Australia can be seen at St. Vincent's
- Currently there are active clinical sites in New Zealand, Spain, and soon in the UK
- Unfortunately, patients who have received a lung transplant are not eligible for the clinical study



For further information:

- Clinicaltrials.gov - NCT04764448
- https://shinetrials.com
- https://estrella-nz.researchstudytrialau.com/
- Contact Sue Brenton, ESTRELLA Study Coordinator, St. Vincent's Hospital:

**St Vincent's Hospital Melbourne**
**Principal Investigator: Dr. Matthew Conron**
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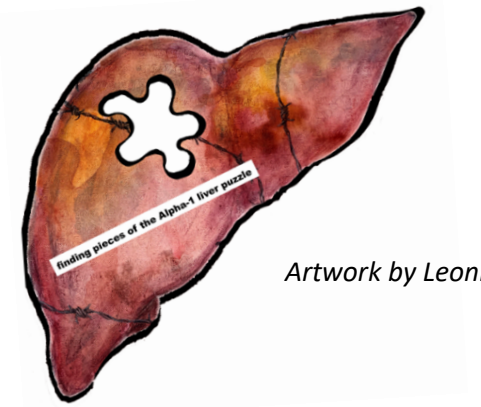


*Artwork by Leonie Robison*

 

*Artwork by Gaynor Heading*
*For use on Mark's and Matt's bike shirts*

*Mask design by Leonie Robison*

## Aunty Alpha

Dear Aunty Alpha,
I have Alpha-1 and find myself getting breathless when I do any sort of physical activity. I am wondering if it is a good idea to exercise and can this improve my lung capacity?
Regards, James

Dear James,
Is sounds like you could have emphysema, which is one of the main diseases of Alpha-1. It is possible you also have COPD or bronchiectasis or a lung infection. Have you been tested for these?
Improving fitness levels is beneficial for everyone and can often improve health and quality of life.
I would suggest you start with some gentle exercise, such as a short walk, and gradually build up to longer walks and more strenuous activity. If you can't walk, there are arm, leg and seated exercises that can help. Specific exercises for chest and lung strength might also help you.
It would be a good idea to consult your doctor first and possibly see a physiotherapist for some suggestions. Your doctor may refer you to pulmonary rehab which patients report as being beneficial. If you can't attend, there are pulmonary rehab videos online that could be useful.

Regards, Aunty Alpha

*Have a question about Alpha-1 Antitrypsin Deficiency?*
*Write to Aunty Alpha at contactus.a1oa@gmail.com*

 

**An Alpha Story - David James Lloyd by his son Mark**

My dad first started to notice a tightness in his chest when we were on a ski holiday back in the mid 90's. I was only a young kid at the time, but I remember him struggling to catch his breath every time he skied down the mountains. We were also renovating our house in the UK and the dust really got to his chest.

This kicked off a series of ongoing doctor's appointments, with him finally being misdiagnosed with asthma. He was put on inhalers for 4 years but unfortunately, they didn't help, and his condition slowly worsened. He was referred to a chest physician at Brompton Hospital in the UK, where he had a blood test and confirmed he had Alpha-1 Antitrypsin.

His condition was managed on steroids and inhalers, and he was informed there was no cure. His lung capacity gradually deteriorated to below 30% and he started on portable oxygen. The decision was then made that a lung transplant would be the best option for him, and he was put on the waiting list at Harefield Hospital in England. The surgeon there was only prepared to offer my dad one lung. This was not an ideal solution due to the systemic nature of the disease affecting both lungs. The complications of weaning him off life support post-operation with one good lung and one diseased lung would have been an impossible physical challenge.

Finally, my dad's physical condition had deteriorated to the point that he had no choice but to go ahead with the op. On 8th February 2009 he was admitted to hospital. The next 6 weeks saw him being treated in Intensive Care as his body fought against the new lungs. I visited him a couple of times, but he was heavily sedated and not able to communicate. He had actually been given two lungs during the operation! Sadly though, on the 18th of March 2009 he passed away. He was 61 years old.

I think it's important to say that he smoked throughout his early life. In addition to this, he had other underlying health problems as a child and had issues with dust from building work - all of which played a part in the severity of his condition.

I still miss my dad a huge amount and think about him every single day. He was a very intelligent man, working as a pharmaceutical consultant throughout his life and I learnt a lot from him. He never once spoke to me about his condition or what was going on at the time. I guess parents' natural instinct is to not want to worry their kids.

My father's experience, which mirrors what many Alphas experience in trying to get diagnosed, and the time that has passed since my dad passed away, has given me the perspective to begin to fully understand and educate myself about Alpha-1.

These difficulties and the lack of access to treatment in Australia, have motivated me to organise my ride to raise awareness about this rarely diagnosed condition.

Halfway through my cycle from Sydney to Melbourne, it will the 13th anniversary of his passing.

My goal is that through doing this cycle, I can play a small part in generating awareness and funding for other Alpha-1 sufferers.

 