



ALPHA-1 Association of Australia

# Who am I ?

- Steven Knowles, no medical credentials or background.
- Background is primarily banking & finance, also recruitment with a focus on getting medical professionals into Australia & NZ, primarily family physicians.
- Interest in Alpha-1 is an MZ phenotype, with daughter MM, daughter MZ, son ZZ.
- Founded Alpha-1 Association of Australia in 2005  
[www.alpha1.org.au](http://www.alpha1.org.au)
- Member of steering committee on Alpha-1 Global.
- Jenni Nankervis, also here at the 7<sup>th</sup> Global Patient Congress, also actively involved, especially raising awareness of Alpha-1 and fundraising.



# Alpha-1 Register in Australia

## Current status

- No official register in Australia yet exists.
- There is a hospital-specific collation of patient details, about 105 as at March 2019, maintained by Sue Brenton of St Vincent's Hospital in Melbourne.
- As at 5 Apr 2019 the AAA's online discussion forums has 234 registered users across Australia and New Zealand, most of whom are Alpha-1 patients.



# Alpha-1 Register in Australia proposed

- The AAA continues to seek the funds (we need another AUD 30,000 [~USD 21,000] of a total AUD 40,000) needed for Australia to establish a register modeled on the Alpha-1 Research Registry.
- Murdoch Children's Research Institute (MCRI)
- Based in Melbourne.
  - Is a REDCap partner.
  - Has agreed to help develop and act as host Alpha-1 register, based on the Alpha-1 Research Registry.
  - Has determined that a Project Coordinator / Research Assistant is needed for 2 days per week for a year, primarily to seek ethics committee approval, and also manage the technical aspects of adapting the Alpha-1 Research Registry framework for Australia.



# Newborn screening

- Newborn bloodspot screening (NBS) has operated for about 50 years.
- Began with screening for phenylketonuria (PKU), to now around 25 rare genetic conditions. Growth due to introduction of tandem mass spectrometry into NBS between 1998 to 2005.
- NBS National Policy Framework established 8 Dec 2017.
- AAA to submit application for inclusion of Alpha-1, however it's been suggested it may be an uphill battle due to cost / benefit of detection of Alpha-1 in newborns.
- Inclusion of Alpha-1 in newborn screening should answer question of epidemiology, and raise awareness.



# Testing for Alpha-1 in Australia

- Australian government (via Medicare) currently covers the cost of two different tests:
  - Quantitation in serum, urine or other bodily fluids.
  - Isoelectric focussing or similar methods for determination of AATD phenotype in serum.
- The AAA has made a submission to Medicare to have genetic testing funded also. Genetic testing is currently available but at a cost to the patient.



# Research

- Four known researchers currently.
  - Assoc Prof Ashley BUCKLE - Monash University, Melbourne.
    - Grant awarded by Alpha-1 Foundation
    - Working to produce a synthetic molecule for Alpha-1 treatment
  - Dr David CHAPMAN – University of Technology, Sydney
    - 2018 TSANZ / LFA / CSL Behring Research Award
    - Researching autoimmunity and Alpha-1
  - Dr Gang LIU – University of Newcastle, NSW
    - 2019 TSANZ / LFA / CSL Behring Research Award
    - Targeting long non-coding RNAs as a novel therapy
  - One other – awaiting details



# Augmentation therapy in Australia

- Augmentation therapy is available in Australia, but is not yet funded by the Australian government:
  - CSL Behring's Zemaira was registered with Therapeutic Goods Administration (TGA) on 13 Feb 2017.
  - Grifols' Prolastin-C was registered with Therapeutic Goods Administration (TGA) on 10 Nov 2016.
- Both manufacturers submitted applications seeking government funding of augmentation therapy to the National Blood Authority in Oct 2017. A decision is awaited, expected some time during 2019.





# Augmentation therapy in Australia

- On 5 April 2019 the AAA released version 1.0 of its Position Paper on the argument for augmentation therapy funding in Australia, entitled:  
**A Normal Life Expectancy From Alpha-1 Proteinase Inhibitor: Methodological Considerations Related to Research on Augmentation Therapy in Alpha-1 Antitrypsin Deficiency**
- Available at [www.alpha1.org.au/doc/ProTherapy.pdf](http://www.alpha1.org.au/doc/ProTherapy.pdf)
- The AAA hopes this document will help other countries with the argument for funding.
- A dynamic document, your feedback is most welcome.

