

## The concerns and problems facing Alpha-1 Antitrypsin Deficiency patients

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Alpha-1 Antitrypsin Deficiency (AATD) is probably the most common life threatening inherited disease in Europe. It is estimated that in excess of 100,000 individuals on the Continent suffer from it.<sup>1</sup> It fatally affects the lungs (emphysema) and liver (cirrhosis). As the only therapy for liver AATD is transplantation and as lung disease is far more common, I shall confine my remarks to lung related AATD for which replacement (augmentation) therapy has been developed and is fairly widely used. This is a plasma derived product and is IV delivered. An inhaled product is currently being developed also.

However, most patients do not know that they have the condition and most doctors do not know that some of their patients have the condition either. This is true even with symptomatic patients. They are usually diagnosed with Asthma or COPD. This in turn, means that they are being inappropriately treated with therapies that do little for their condition of Alpha-1 Antitrypsin Deficiency.

It has been estimated in the USA that it takes an average of five doctors, over a seven year period, to diagnose the condition in a patient. Even after that long delay there is no guarantee that an Alpha-1 patient will be diagnosed. To date, only c. 5% have been diagnosed in the countries that are looking for them.

AATD is a deficiency of a vital lung protecting protein. Therefore, it would seem that the obvious therapy is to replace or augment that which is lacking. This therapy has been developed and is now being widely used in the USA and in Europe. In Europe it is licensed and prescribed in the following countries:

Germany	Austria
France	Spain
Italy	Portugal
Ireland	

It is important to note that its prescription and use varies very much from area to area and from country to country. So we cannot say that it is universally available

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<sup>1</sup>In a recent study in Dublin, on a random population sample of 1,000 individuals we had to double the estimate of the AATD population in Ireland. Therefore, the estimate given may be very wide of the mark.

to patients in Europe. It is available on a named patient basis in some EU countries other than the above.

Some physicians will not prescribe the therapy because they believe that its efficacy has not been proven in a double blinded placebo controlled clinical trial. Such a study is being carried out currently internationally by CSL Behring. Countries involved in this study include Ireland, Poland, Nordic countries and Australia. The main producer for this replacement therapy for Europe is Talecris Biotherapeutics (Prolastin). However, it is also being produced in France and Spain.

It is interesting that the therapy is not generally available in the Nordic countries or the UK and this causes much concern to AATD patients in these countries who know that their peers in France, Spain, Germany et al are receiving it and that it is beneficial to them.. It is my opinion that it is contrary to the spirit of the Mutual Recognition Programme directive of the EU Commission that it is not licensed and reimbursed in all EU countries. In AATD we have a “second class citizen” situation rather than equity of availability.

It is possible that, as a result of the clinical trial mentioned above, that augmentation therapy will gain wider acceptance among regulators and clinicians across Europe and thereby become accessible to all AATD patients. Until that day arrives thousands of patients will have to suffer without its benefit.

High self esteem contributes in no small way to patient wellbeing and often to recovery. Alpha 1 Antitrypsin Deficiency affects patients in a serious way in their 40s and 50s and can be very debilitating at this time of their lives. Hospitalisation and infections lead to loss of work days and eventually to retirement/unemployment.

People on appropriate therapies:

1. stay at work longer and continue to pay taxes – at mid 40s to retirement normally at the highest level, thus generating revenue for the state
2. are not dependent on social welfare – thus saving on unemployment benefit and social welfare costs
3. are less often hospitalised – another saving, when we consider the cost of a night in hospital
4. are productive in work and thus contribute to national wealth.

Healthy patients save the exchequer significant sums of money. Much more importantly, their self esteem is greatly improved. Few, if any of us, enjoy paying taxes but being employed and a net contributor is better for self esteem than being unemployed and a net receiver.

However, even where therapies are licensed and reimbursed not all Alpha-1 Antitrypsin Deficiency patients will receive the treatment as 95% of them have yet to be identified and correctly diagnosed. The common mantra, that patients who receive early diagnosis and therapy fare better in health and lifestyle than those who do not, holds true here also. National patient groups and institutions as well as Alfa Europe are urgently promoting detection and screening programmes. Currently there are numerous screening and detection programmes in progress in various

countries. Lobbying is also being used to raise awareness of the need for screening and detection among various groups such as: patient families, medical practitioners, statutory healthcare givers, local politicians and ministers, the EU Parliament and the healthcare industry.

To date, we have been fairly successful in getting financial help for these programmes from both industry and state e.g. Ireland: Targeted detection Programme, Germany: Screening Programme and Italy: Area Screening Programme.

As AATD is a genetic condition it is vital that family members of AATD patients be also tested for the condition. Alfa Europe strongly encourages families of patients to participate in testing and screening programmes. However, there are problems to be faced such as:

Psychological, Some people just do not wish to know. Some parents feel huge guilt for “giving” their children the condition.

Financial/Social, Some people fear the effect a diagnosis will have on their own and their children’s insurance premiums, mortgage availability etc.

Therefore, it is essential that good information and counselling is made available to newly diagnosed patients and their families.

Other therapies such as, pulmonary rehab and physiotherapy are available across Europe. However, there is no central plan for AATD patients and services can be sketchy particularly in less densely populated areas but that is true for most therapies for most medical conditions. Oxygen therapy is widely available for patients when the condition becomes more severe. This, in turn, can cause problems when travelling especially by air.

Outreach services are sometimes available in some countries but by no means, in all. It will be necessary for Alpha-1 Antitrypsin deficiency to be planned for in national health plans and in EU directives on health.

It seems to me that there are three immediate requirements to be prioritised in the diagnosis and treatment of alpha-1 patients.

1. Availability of a full package of therapies and remuneration in all European Countries.
2. Adequate counselling services for newly diagnosed patients and their families.
3. Accredited national screening or detection programmes for Alpha-1 patients.

It is incumbent on all states in the EU to afford equal access to adequate services and therapies to people with Alpha-1 Antitrypsin Deficiency.

In the meantime we who represent patient groups, medical care providers and the healthcare industry need to:

- Continue political lobbying
- Raise awareness in the general population
- Raise awareness among General Practitioners
- Inform pulmonary healthcare providers
- Continue working closely together and in harmony.