FRANK WILLERSINN, M.D.
Founder and President of Alpha-1 Plus Belgium
Member of the Board of Directors of the Alpha-1 Foundation, Miami
Member of the Alpha-1 Global Steering Committee

Living An Active Life With Alpha-1 Antitrypsin Deficiency

INTERVIEW WITH FRANK WILLERSINN, M.D.

BY DR. IRINA ODNOLETKOVA, PPTA DIRECTOR, HEALTH ECONOMICS & OUTCOMES

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Q What is Alpha-1 antitrypsin deficiency?

Alpha-1 antitrypsin deficiency (AATD) is a rare genetic disorder that causes defective production of alpha-1 antitrypsin. Depending on the form and degree of deficiency, it may lead to lung disease, liver disease, or skin disease. Among the rare diseases, AATD is one of the most common conditions. But not everyone with AATD develops a clinically significant disease. Probably about a tenth of the alphas require plasma protein augmentation therapy. The different genotypes will lead to different serum levels of alpha-1 antitrypsin (A1AT). Most patients with clinical disease are homozygous SS or ZZ. They have the lowest serum levels of A1AT.

Q How were you diagnosed with AATD?

I was a heavy smoker for more than 15 years and I had chronic bronchitis. Like a lot of others, I was diagnosed by chance in 1998, when my alpha-1 globulin levels were tested due to a mistake in the laboratory request form. Without special symptoms, I did not go to the doctor immediately. All I knew was that AATD had a genetic origin. But at that point, I was much more preoccupied with adopting a new way of life after I lost my arm two years earlier, a consequence of a severe and rare form of streptococcal infection. Statistically there was no chance of survival. I was brought in in a coma, and my heart even stopped beating once. By a miracle, I survived but lost my infected arm. Since then I experience every day as a gift.

Q What moved you to start patient advocacy work?

Ten years after the diagnosis, when I started with alpha-1 antitrypsin augmentation therapy, I wanted to learn more about AATD and find other people with this rare disease. Together, we contacted Members of the European Parliament (MEPs) and explained to them that the problem of AATD is not sufficiently addressed in Belgium. After this meeting, the MEPs reached out to the Belgian Minister of Health, and we realized that better care may be achieved through a dialogue with health care policymakers. In 2013, I founded the Alpha-1 patient association in Belgium, which was made up of eight members with AATD and gave us an identity and a voice with policymakers.

Q How do you see the mission of your patient association?

The mission is to improve the health and quality of life of other alphas. The founder of AlphaNet and COPD [chronic obstructive pulmonary disease] Foundation in the U.S., John W. Walsh, was a great source of inspiration. Currently, I am involved in patient advocacy at different levels, like Alpha-1 Global, which is run by eight volunteers. We support national associations through developing information and advocacy tools to improve early diagnosis, education of specialists, establishment of treatment centers, etc. We find patients and patient associations throughout Europe and help to develop new associations in countries where they don't currently exist. Along with a group of top experts in Europe, we are also actively involved in drafting EU recommendations for AATD. The upcoming 6th Alpha-1 Global Patient Congress will be held in Lisbon, Portugal on April 5-8, 2017 (see http://www.alpha-1global.org/ for more information).

Q What have you been able to achieve?

In more than 30 countries in Europe, there is no reimbursement for AATD treatment. Through educating physicians in countries such as Romania and Poland, and supporting them with diagnostic tools, we identify local alphas and bring them together. They can find national patient associations and we help them in their advocacy work. Bringing them in contact with other well-established national structures—such as rare disease foundations supporting immunodeficiency and hemophilia—is important to build their health policy experience. Similarly, we set up Alpha-1 Switzerland, which started with twelve enthusiastic alpha-1 volunteers and now, six months later, they have 35 members.

Q What are the main challenges in your advocacy work?

The main challenge is that every country requires a different approach. To start, all work needs to be done in the national language and there are 24 official languages in Europe. Furthermore, each country has its own health care system and policy for rare diseases, which creates inequalities. For example, in the Netherlands, the replacement therapy is reimbursed to people with severe AATD (phenotype OO), in Belgium the reimbursement stopped several years ago, leaving those alphas diagnosed after June 2010 without treatment. We have taken several actions against this discrimination and will continue this work. Also, in the UK, no reimbursement was granted by the National Institute for Health and Care Excellence. The budget impact of AATD in the UK would be high, indeed, with about 1,300 patients waiting for treatment and an annual treatment cost per patient of approximately 80,000 € (about $85,400). However, patients and clinicians don't give up their struggle and hope.

Q What is your perspective on the plasma sector?

The plasma protein products industry has a great potential to open new markets, both geographically and in terms of new indications, because AATD also has an influence on other metabolic balances.

I believe that more targeted (personalized) treatments will be possible when the underlying causes of diseases are better understood. An important challenge for the future is to ensure more plasma collection in Europe. Currently, most of the plasma is coming from the U.S., which worries me as a patient with European residence. In Europe, a solution must be found to motivate young people to donate plasma. It will require a re-thinking of the communication campaigns to make clear to people how important their contribution will be for saving other people's lives. •