AMYLOIDOSIS ALLIANCE
THE VOICE OF PATIENTS

2ND EUROPEAN BERLIN ATTR MEETING
Berlin, 2-4 September, 2019

SALIENT POINTS AND MAJOR FINDINGS
From a patient's perspective

ALLIANCE POLICY AND PRACTICE

The policy and plans of the Amyloidosis Alliance to further evolve from a federation of mainly European, ATTR focused Patient Organizations in the direction of a **global Alliance for all types of Amyloidosis** (including AL and AA), was met with approval. Nonetheless, for reasons of cost-effectiveness and convenience, international meetings with a more regional focus and/or with focus on particular type(s) of Amyloidosis, may still be considered.

- **A 'medication map'** was presented in the Patients' meeting showing which ATTR medication is available in which country, and accessible under which conditions (clinical trial, open label. compassionate use, or approved for marketing)- should be regularly updated and displayed on the Alliance website. The update of this map should be done with the support of the pharmaceutical companies because there were some errors in the information provided by the patients associations.

- The Alliance can/should provide support to local patient organizations and groups in their advocacy work to get access to appropriate medicines and treatment.

- Raising awareness is an Alliance priority, and in cooperation with Alnylam it is working on video material and a toolkit on awareness raising that can be used by Patient Organizations in their home languages.

RESEARCH PRIORITIES

- There are good reasons to assume, and some experiences in that direction would indicate, that a **combination of medicines** can be more effective than a single medicine, such as a TTR stabilizer in combination with a RNA interfering medicine. The industry and health authorities should provide flexible support to physicians to test this idea.

- In a reaction to a presentation on the proven effectiveness of new medication in prolonging a patient's life. one patient observed that he welcomes the extension of life but not ending it "being crazy and blind'. In this, he was referring to the fact that some medicines seem less effective in preventing TTR **deposits in the eyes and brains**. Patients see studies aiming at tackling this problem as a research priority.

- The industry should allow and facilitate expertise centers to undertake **'comparative' research**, so as to find out which medicine is the most effective for which type of Amyloidosis and for which type of patient.
- The information provided on healthy nutrition for Amyloidosis patients was much appreciated. Yet, further studies seem to be necessary to identify which nutrition, diets and lifestyle could be most effective in diminishing the production of amyloidogenic proteins, and treatment of symptoms.

Note for further discussion :
*Is the use **of RCTs(Randomised Placebo Controlled Controlled Trials) the only way to provide a robust data base for assessing the efficacy of a medicine ?** Nowadays new data collection tools (like sensors) developed by health technology allow to monitor closely impact and side effects of a medicine. Could it be possible to collect robust data on efficacy without the use of a placebo group ? From an ethical point of view, the administration of placebo can put the well being and life of patients at risk by delaying his/her acces to the medicine, they badly need. A patient should have "the right to try", i.e. have the freedom to get the medicine which needs further testing , provided he/she is informed about the risks as far as these are known. How can we make clinical trials more cost-effective and less cumbersome for patients and participating clinics ?*

PATIENT REGISTRIES

- There was a presentation by IQVIA, a specialist in the design of effective rare disease registries, including Amyloidosis, and in protecting individual patient privacy. The question arose as to whether the existing THAOS registry is sufficiently suitable for existing Centers to make optimal use of the data collected by the different Amyloidosis Expertise Centers . Can a system could be developed that could be the basis of a 'big data bank' ; a 'Bank which healthcare providers of many countries could tap into for a deeper understanding of the disease and efficacy of treatments ? But which they also feed with the data they have collected ?

Such a Bank, could be of great benefit to patients since it will help to advance on the path towards the best treatment and cure. Are their Expertise Centers willing to participate in a project that will aim at setting up such a system which will allow such Centers and researchers to share data and experiences, and communicate conveniently, regardless of their location, and make optimal use of one another's data ? Could/should the Thaos registry be an integral part of that?
Perhaps IQVIA technicians could be further consulted to explore if they can assist in drawing up such a plan that will aim optimizing the use of patient registries. How can you ensure that the data is fed by reliable an usable data ? And prevent 'rubbish in' resulting in 'rubbish out'' ? Is the widespread use of AI around the corner ? Or do we still have a long way to go ?

DIAGNOSIS

There is not only a problem, in all countries, of **late diagnosis**, but of **misdiagnosis** . It was noted some French doctors, with the support of the French Association, have initiated a process of finetuning and standardizing protocols for the diagnostic process and the Dutch University Hospital in Groningen is also pioneering this field. With the help of the Alliance, this process could be internationalized. The protocol that will emerge, may become an integral part of an internationally accepted 'standard of cure'. The Alliance needs to get the support of ir medical advisors to collect the protocols and define what could be the best protocol(s) available in the different countries

CURE AND CARE

- Several commendable, Industry supported surveys, have been undertaken on the ''burden of the disease', and its effects on the quality of life of patients and their families. The underlying question isπ: 'what kind of accompanying services do patients and their loved ones require to be able to cope with these burdens or what kind of practical as well as socio-psychological support do they need ?

- A pioneer study on the integral approach of cure and care (a combination of medical, para medical and non-medical services) is being undertaken by the social sciences Department of the University of Sevilla / Spain (Ms. Inmaculada Monterdez) and the Expertise Centre of the the Paramiloidose Hospital Geral de Santo António in Oporto, Portugal (Prof.Teresa Coelho).

The patients are facing many issues to live with the disease. They need some support for all the administrative aspects linked to the following categories: Health insurance, Work, Caregivers, Disabilities. The Alliance has to support the access to this information in each country, according the local laws.

At international level, a study might be considered which makes an inventory of the type of accompanying services currently provided by different Expertise Centers in France, Portugal, Germany and the UK; the cost-effectiveness of such services, and how meaningful they are for patients. This could be the basis for the development of a 'standard of care', in synergy with an internationally accepted ' standard of cure' .

The way Amyloidosis is being cured, in the longer term, MAY change substantially, if the CRISPR - mediated targeted gene modification, proves effective. INTELLIA Therapeutics is working on a protocol for a clinical trial to test this out.