

## Rare Diseases: No Innovative Therapies Without Research



Individualized therapies and the integration of the patient's perspective are becoming increasingly important.

February 29 is Rare Disease Day. On this occasion, the Austrian pharmaceutical company AOP Orphan Pharmaceuticals GmbH (AOP Health) takes a look at the challenges faced by patients with rare diseases and highlights the trends in their treatment.

With over 30 million Europeans affected, including 400.000 Austrians, rare diseases are becoming an increasingly pressing health issue. It is alarming that, as of yet, no specific therapy options exist for 95 percent of these diseases. AOP Health, specialized in the research and development of therapies for rare diseases as well as in intensive care, has been involved in this market segment for over 25 years, aiming to provide medical solutions for those affected.

### Research is the key to new therapies

Extensive research programs are needed to create new treatment options. Currently, these are impossible without private initiatives. In Austria, 50.4 percent of all research is conducted by private companies<sup>1</sup>. In the field of drug development, this proportion is even higher because the approval or marketing of drugs is applied for or carried out exclusively by pharmaceutical and biotech companies. In order for new therapies to be developed, framework conditions that promote research and development are necessary, as Martin Steinhart, CEO of AOP Health, emphasizes:



If we want to ensure that the supply of medicines in Europe remains guaranteed, we need regional research programs, a strong pharmaceutical industry and clear regulations that promote the development of new therapies and make research attractive for diverse actors.

Dr. Martin Steinhart  
CO-CHIEF EXECUTIVE OFFICER | AOP HEALTH

Steinhart is therefore skeptical of the planned EU pharmaceutical legislation because the draft includes a shortening of the data protection period as well as the market exclusivity duration. This means that companies are left with less time to recover the costly developments through sales. The CEO of AOP Health continues: *"Shorter protection periods, during which pharmaceutical companies can market their development exclusively, make it even harder for those companies that want to get involved in the already challenging field of rare diseases. The planned legislation could therefore make the situation more difficult for patients in Europe and weaken the business location."*

### Trend towards individualized therapy

The common trend towards personalized medicine is also strongly noticeable in the field of rare diseases. Improved diagnostic methods thus enable a more accurate determination of the clinical pictures. This means that clinical pictures can be divided into subgroups with specific characteristics and therapy needs, thereby identifying particularly rare diseases or forms of disease. Treatment has also changed significantly in recent years: Newly discovered therapeutic approaches place special emphasis on individualized therapy. Molecular testing and targeted therapies can consequently improve prognosis and treatment. The "one size fits all" approach is a thing of the past. AOP Health integrates these new insights into its development programs.

Bianca Tan, Director of the Therapeutic Area Cardiology & Pulmonology at AOP Health, explains:

Our goal is to find answers to hitherto unanswered medical questions for our patients. In our research, we therefore also try to understand the diseases' mechanisms at the molecular genetic level to perhaps even achieve disease modification. On top of that, we are examining whether patients can benefit from synergies induced by combination therapies.

Bianca Tan, MSc  
THERAPEUTIC AREA DIRECTOR CARDIOLOGY & PULMONOLOGY



## Treatment in specialized centers yields better results

In addition to individualized therapy, prompt referral of patients to specialized centers is pivotal. Prof. Irene Lang, a specialist in pulmonary vascular diseases at MedUni Vienna, explains: *"Patients with rare diseases benefit enormously from treatment in an expert institution. On the one hand, there are specialists who deal with those affected on a daily basis and therefore have a lot of experience in the diagnosis and selection of therapies. On the other hand, they offer the necessary diagnostic and therapeutic infrastructure."* She thus recommends that colleagues outside special centers immediately refer patients to an EU-approved center if they suspect a rare disease. Lang continues: *"It is crucial for those affected to quickly receive the correct diagnosis and treatment. Treatments for pulmonary hypertension, for example, are expensive and time-consuming, and it is detrimental to our health care system if the wrong patients receive this expensive medication."*

## Let's talk to patients, not about them

Another trend that is visible throughout medicine is the increasing involvement of the patient's perspective in treatment, but also in research work. Represented by so-called patient advocacy groups, those affected are not only involved in the selection of therapies, but also in the preparation of study designs. However, they still face hurdles in their work.

Claas Röhl, chairman and founder of the Patient Advocacy Group NF Kinder and board member of Pro Rare Austria, says: *"The current regulations, which are actually intended to protect patients, make it difficult for us to be involved in the development of medicines and therapies."* It is essential that patients are perceived as experts in their disease by all players within the healthcare system. He adds: *"We want our concerns and needs to be incorporated because it is our lives that are at stake here. Our perspective is unique and cannot be covered by scientists. We rely on training programs, partly at university level, which empower us to talk to the treatment teams, but also to pharmaceutical companies on an eye level, in order to be heard and to communicate the needs of the patients, so that research activities are geared towards them."*

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