

## **The challenges of conducting clinical development in rare / orphan diseases – The industry perspective**

Thomas Meier, PhD

Santhera Pharmaceuticals, Liestal, Switzerland ([www.santhera.com](http://www.santhera.com))

Santhera Pharmaceuticals is a Swiss based, specialty pharmaceutical company with a focus on discovery, development and marketing of small molecules for the treatment of orphan neuromuscular diseases. We have a clinical portfolio of three compounds in seven indications. Our lead compound idebenone (CATENA®) is approved in Canada for use in Friedreich's Ataxia and in clinical development for Duchenne Muscular Dystrophy, Leber's Hereditary Optic Neuropathy, MELAS and primary progressive MS. We have high quality partnerships along the value chain: in-licensing (Novartis), development collaboration (Juvantia) and in marketing (Takeda).

According to the World Health Organization there are more than 5000 rare diseases, affecting less than 10 patients per 10,000 of the population. Most of these have too few patients for companies to develop commercially viable treatments. The USA was the first country to implement Orphan Drug (OD) legislation in 1983, and saw a dramatic increase in registrations for treatments in rare diseases. Other countries have followed this lead – Japan in 1993, Australia in 1998 and Europe in 2000.

There are many challenges in developing drugs for these rare or orphan diseases. Epidemiology is often not well established, making it very difficult to assess the feasibility of clinical studies. Hence running clinical trials and recruiting centers and patients can be extremely difficult. Other challenges include the lack of reliable longitudinal data on the natural progression of the disease, and the validation of clinical endpoints acceptable to health authorities.

At Santhera we have built our business model around working in rare neuromuscular / neurological conditions. Facing all of these challenges we have successfully run several clinical studies and benefited from collaborations with many academic groups around the world. In this presentation Santhera's business concept and approach to drug development in rare neuromuscular diseases will be presented.