

Annual Congress on

Rare Diseases & Orphan Drugs

October 26-27, 2016 Chicago, USA

COST action BM1207: Involving all stakeholders to overcome challenges of genetic therapy development for Duchenne muscular dystrophy

Annemieke Aartsma-Rus

Leiden University Medical Center, Netherlands

Duchenne muscular dystrophy (DMD) is a rare, progressive muscle-wasting disease leading to severe disability and premature death. Treatment is currently symptomatic but multiple experimental therapies are in development. Implemented care standards, validated outcome measures correlating with clinical benefit and comprehensive information about the natural history of the disease are essential for the regulatory approval of any therapy. However, for DMD and other rare diseases, these were not in place when potential therapies entered the clinical trial phase. This has resulted in suboptimal trials for DMD therapy. To address this, a cooperative effort of DMD stakeholders, including representatives from patient groups, academia, industry and regulatory agencies aimed at identifying strategies to overcome challenges, developing the tools required and collecting relevant data. This is ongoing work, but already a huge effort has been made to develop new outcome measures, collect natural history data and to develop potential biomarkers. The open and constructive dialogue among stakeholders has positively influenced therapy development for DMD and this should serve as a paradigm for rare disease therapies' development in general.

Biography

Annemieke Aartsma-Rus has obtained her PhD at Leiden University, Netherlands in 2005. She became a Group Leader in 2007 and she is currently a Professor of Translational Genetics at Leiden University Medical Center, Netherlands. She currently chairs the TREAT-NMD Alliance (an infrastructure network for clinical trial readiness for neuromuscular disorders) and a networking action (funded by Cooperation of Science and Technology (COST)). She has published more than 100 papers in peer reviewed journals, written multiple book chapters and generated and maintains pages to explain Duchenne therapies in lay terms to the patient community. In 2009 she received the Duchenne award from the Dutch Duchenne Parent Project for her dedication to the field.

A.M.Aartsma-Rus@lumc.nl

Notes: