Full Name
Annemieke Aartsma-Rus

Company/Organization
Leiden University Medical Center

Speaker Bio
Prof. Dr. Annemieke Aartsma-Rus is a professor of Translational Genetics at the Department of Human Genetics of the Leiden University Medical Center (LUMC, the Netherlands). She played an important role in the development of antisense mediated exon skipping for Duchenne muscular dystrophy during her PhD research (2000-2004) at the LUMC. In 2020 she co-founded the Dutch Center for RNA Therapeutics (DCRT), a non-for-profit academic collaboration aiming to develop clinical treatment with exon skipping therapies for eligible patients with unique mutations. In 2022 she became a board member of the N-of-1 collaborative (N1C), a global umbrella organization aiming to facilitate development of antisense oligonucleotide therapies for patients with very rare, eligible mutations.

Her work currently focuses on developing antisense-mediated exon skipping as a therapy for Duchenne muscular dystrophy and rare brain diseases. This involves work in cell and animal models to improve efficiency of exon skipping, studies in muscle pathology, the identification of biomarkers, studying the basics of pre-mRNA splicing and transcript processing and the generation and detailed analysis of mouse models.

Finally, she aims to bridge the gap between stakeholders (patients, academics, regulators and industry) involved in drug development for rare diseases and to develop exon skipping therapies for patients with unique mutations.

She has published over 220 peer-reviewed papers, 11 book chapters and 15 patents. She has given many invited lectures at scientific conferences and patient organization meetings, where she is known for her ability to present science in a clear and understandable way.

In 2011 she received the Duchenne Award from the Dutch Duchenne Parent Project in recognition of her dedication to the Duchenne field. In 2020 she received the Black Pearl Science Award from Eurordis for her work in educating patients. In 2021 she received the Ammodo Science Award for her contribution to developing exon skipping therapies for Duchenne, the outstanding achievement award from the Dutch Society of Gene and Cell Therapy for her work and the Rosalind Franklin in Science award for her work for the journal Nucleic Acid Therapeutics.

Since 2015 she is the most influential scientist in Duchenne muscular dystrophy in the past 10 years (https://expertscape.com/ex/muscular+dystrophy%2C+duchenne).