Insulin-like growth factor-1 and Duchenne muscular dystrophy

Yavaş, A.

List of Publications

Yavas A, Weij R, van Putten M, Kourkouta E, Beekman C, Puoliväli J, Bragge T, Ahtoniemi T, Knijnenburg J, Hoogenboom ME, Ariyurek Y, Aartsma-Rus A, van Deutekom J, Datson N. Detailed genetic and functional analysis of the hDMDdel52/*mdx* mouse model. PLoS One. 2020 Dec 23;15(12):e0244215. doi: 10.1371/journal.pone.0244215.

Yavas A, van Putten M, Aartsma-Rus A. Antisense Oligonucleotide-Mediated Downregulation of IGFBPs Enhances IGF-1 Signaling. Journal of Neuromuscular Diseases. 2024;11(2):299-314. doi:10.3233/JND-230118

Yavas A, van Putten M, Ariyurek Y, Aartsma-Rus A. Antisense oligonucleotide-mediated redirection of *Igf1* alternative polyadenylation, *submitted*

List of Abbreviations

2OMePS	2'-O-methyl phosphorothioate
AAV	Adeno-associated viruses
ALS	Acid-labile subunit
ANOVA	Analysis of variance
AON	Antisense oligonucleotides
AS	Alternative splicing
BAM	Binary alignment map
BMD	Becker muscular dystrophy
DE	Differentially expressed
DGE	Differential gene expression
DMD	Duchenne muscular dystrophy
DAPC	Dystrophin-associated protein complex
DSB	Double-strand break
DSE	Downstream elements
EBD	Evan's blue dye
ECM	Extracellular matrix
EMA	European Medicines Agency
E-peptides	Extension peptides
ESE	Exonic splicing enhancer
ESS	Exonic splicing silencer
FDA	Food and Drug Administration
FISH	Fluorescent *in situ* hybridization
GH	Growth hormone
GO	Gene ontology
gRNA	guide RNA
HDAC	Histone deacetylase
HDR	Homology-directed repair
IGF-1	Insulin-like growth factor-1
IGF-1R	IGF-1 receptor
IGFBPs	IGF-binding proteins
IRS-1	Insulin receptor substrate-1
ISE	Intronic splicing enhancer
ISS	Intronic splicing silencer
MD	Muscular dystrophies
MGF	Mechano growth factor
NHEJ	Non-homologous end joining
PAS	Polyadenylation signal
PMO	Phosphorodiamidate morpholino

rhIGF-1	Recombinant IGF-1
SMA	Spinal muscular atrophy
SRSF1	Serine-arginine protein splicing factor-1
TALEN	Transcription activator-like effector nuclease
TGF-β	Transforming growth factor-beta
USE	Upstream elements
UTR	Untranslated regions

Curriculum Vitae

Alper Yavas was born on the 4th of September 1989, in İnegöl, Türkiye. He completed his Bachelor's degree in Molecular Biology and Genetics in Istanbul Technical University, and his Master's degree in Middle East Technical University in Ankara. During his graduation studies, he worked on several topics including plant functional genomics and microbial genetics. In 2019, he moved to the Netherlands to embark on his PhD journey in the Department of Human Genetics, at the Leiden University Medical Center (LUMC) in the group of Prof. dr. Annemieke Aartsma-Rus. In his PhD studies, he focused on developing AON-mediated strategies for treating secondary defects in Duchenne muscular dystrophy by modulation of IGF-1 signaling and posttranscriptional events; alternative splicing and polyadenylation. He also worked on genetic and functional characterization of the humanized mouse model (hDMDdel52/*mdx*) that allows testing human-specific AONs in mice. In February 2024, he began working as a postdoctoral researcher in the Pang Lab, at the Department of Cell and Chemical Biology, LUMC. He is currently studying the function of the non-coding human genome using state-of-the-art genome-editing tools.

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