nature portfolio

https://doi.org/10.1038/d41573-024-00020-8

Supplementary information

Facilitating development of AAV gene therapies for rare diseases of no current commercial interest

In the format provided by the authors

Supplementary Table 1

Project Title	Principal Investigator	Institution
Rep, Cap, and Adenovirus Synthetic RNAs for Manufacturing Recombinant Aden-Associated Virus Vectors	Alan Davis	Baylor College of Medicine
Stable adeno-associated virus vectors for human gene therapy	Fred Bunz	Johns Hopkins University
Bottlenecks in AAV cellular entry, trafficking and nuclear delivery	Michael Chapman	University of Missouri
Dissecting and piloting the intracellular trafficking of AAVs	Antonella deMatteis	Telethon Institute of Genetics & Medicine
Investigating Innate Sensing and Antiviral Restriction of AAV vectors in the Human Central Nervous System	Anna Kajaste-Rudnitski	San Raffaele Telethon Institute for Gene Therapy
Characterizing the impact of CpG methylation on AAV genome packaging, expression, and the innate immune response toward improved gene therapy vector production	Brian Davis	GE Healthcare
Quantification of AAV dose-response with single cell resolution	Leah Byrne	University of Pittsburgh
Increasing rAAV transgene size by host factor modulation	Anna Maurer	University of California Berkeley

Supplementary Table 2

Affected Organ System	Disease Name	Affected Gene	AAV serotype	ROA
Ocular	Congenital Hereditary Endothelial Dystrophy	SLC4A11	AAV8	Intracorneal
	NPHP5-Retinal Degeneration	NPHP5	AAV5	Subretinal
	Retinitis pigmentosa 45	CNGB1	AAV5	Subretinal
Neurological	Multiple Sulfatase Deficiency	SUMF1	AAV9	Intracisterna magna
	Spastic paraplegia 50	AP4M1	AAV9	Intrathecal
	Charcot Marie Tooth disease type 4J	FIG4	AAV9	Intrathecal
Systemic	Propionic Acidemia	PCCB	AAV9	Intravenous
	Morquio A Syndrome (Mucopolysaccharidosis IVA)	GALNS	AAV8	Intravenous