



RETT
SYNDROME
RESEARCH
TRUST

RETT SYNDROME RESEARCH TRUST

FUNDS AWARDED BY YEAR

TOTAL AWARDS: \$91.3 MILLION 2008-2025

AAV development and Manufacturing

\$1,100,000

MIT Core Facility

Safety studies for gene replacement using BBB penetrant AAV delivery

\$1,100,000

Tacit Therapeutics

Multi-allelic correction using RNA trans-splicing

\$1,000,000

Profluent

AI Designed improvements to base editor

\$1,000,000

Shape Therapeutics

Single allele correction using small RNAs delivered by AAV

\$300,000

Resplice Therapeutics

Trans-splicing for Rett syndrome

\$130,000

Senti-Bio / Jackson Laboratories

Design and test of an inducible control circuit for base editing activity

\$250,000

Evox

Exosome delivery platform evaluation

\$240,000

Jonathan Watts, PhD

UNIVERSITY OF MASSACHUSETTS

Transvascular Delivery of mRNA and guide RNA to the CNS to Correct MECP2

Mutations that drive Rett Syndrome

\$84,791

Kyle Fink, PhD

UC DAVIS

Long term safety risk linked to constitutive expression of DNA base editing

\$205,647

Kyle Fink, PhD

UC DAVIS / EPICRISPR BIO

X-reactivation of wild type alleles in Rett syndrome

\$269,247

Jackson Laboratories

Partnership with Uncommon Therapeutics to test an ABE (adenosine base editor)

design with low bystander effects

\$219,317

Aleksandra Jacobs, MD, PhD

MONTEFIORE RETT SYNDROME CLINIC

Extending Rett syndrome CSF biomarkers to blood based detection

\$40,123

Bryce Reeve, PhD

DUKE UNIVERSITY SCHOOL OF MEDICINE

Advancing Patient Centered Research

\$215,254

RSRT Biorepository

\$49,817

Rett Syndrome Global Registry

\$107,740

Couragene

\$15,000

Vibrant Study

\$8,741

2025

ProQR Therapeutics

Development of AX-2402 for the Treatment of Rett Syndrome

\$2,650,000

Apertura Gene Therapy

License of Novel Blood Brain Barrier Penetrating Capsid

\$2,000,000

Kai Chen, PhD

UC BERKELEY (JENNIFER DOUDNA LAB)

A New Brain Delivery Platform for Base Editing of MeCP2 Mutations and Gene Knock-in for Rett Syndrome

\$1,158,523

Omar Abudayyeh, PhD and Jonathan Gootenberg, PhD

**BRIGHAM AND WOMEN'S HOSPITAL &
BETH ISRAEL MEDICAL DEACONESS CENTER**

Programmable RNA Writing for Treatment of Rett Syndrome

\$859,990

Jiangbing Zhou, PhD

YALE UNIVERSITY

Non-viral delivery of base editing therapy for Rett syndrome

\$588,306

Jiangbing Zhou, PhD

YALE UNIVERSITY

Non-viral delivery of epigenome editing therapy for Rett syndrome

\$548,306

Shawn Liu, PhD

COLUMBIA UNIVERSITY

Non-viral delivery of epigenome editing therapy for Rett syndrome

\$476,231

RCourage, Inc.

STEP-RNP production of epigenetic and base editors

\$60,000

Kyle Fink, PhD

UC DAVIS

Rett syndrome tRNA screening

\$97,554

Jef Boeke, PhD

NEW YORK UNIVERSITY

Development of a Fully Humanized MECP2 Mouse Model

\$161,753

Vivek Kumar, PhD

JACKSON LABORATORIES

Automated Rett Syndrome Phenotyping and Efficacy Testing

\$258,979

Han Zhang, PhD

UMASS MEDICAL SCHOOL (ERIK SONTHEIMER LAB)

Base Editing Program Support

\$194,732

Rett Syndrome Global Registry

\$88,536

RSRT Biorepository

\$162,873

Aleksandra Jacobs, MD, PhD

MONTEFIORE RETT SYNDROME CLINIC

\$25,000

Adrian Bird, PhD / Jacky Guy, PhD

UNIVERSITY OF EDINBURGH

*Correcting Rett syndrome-causing C-terminal Deletions
using Adenine Base Editors*

\$315,502

**Erik Sontheimer, PhD / Jonathan Watts, PhD /
Scot Wolfe, PhD**

UMASS MEDICAL SCHOOL

Base and Prime Editing Approaches for Rett syndrome

\$2,343,091

Guoping Feng, PhD

MIT

*Single AAV Deliverable and Transiently Inducible Base Editors
for Rett syndrome*

\$3,734,738

Peter Beal, PhD

UC DAVIS

*Directed RNA editing for the repair of MECP2 mutations causing
Rett syndrome*

\$390,506

ProQR

Correction of R270X mutations in MECP2 RNA using Axiomer® Technology

\$1,120,000

Michael Elowitz, PhD / Viviana Gradinaru, PhD

CALTECH

Quantitative, dosage-compensated gene therapy for Rett syndrome

\$500,000

Victor Faundez, MD, PhD

EMORY UNIVERSITY

Correlating Rett Syndrome Brain CSF Proteomes with Blood Plasma Profiles

\$1,150,965

Victor Faundez, MD, PhD / Stuart Cobb, PhD

EMORY UNIVERSITY / EDINBURGH UNIVERSITY

Systems Biology of Rett Syndrome Gene Therapy Outcomes (Supplement)

\$103,120

RSRT Biorepository

Induced Pluripotent Stem Cell and Fibroblast Cell Collections

\$230,949

Rett Syndrome Global Registry

Parent-reported SHARE Study

\$100,750

Emerald Innovations

Digital Technologies for the Assessment of Rett Symptoms

\$20,075

Vivalink

Digital Technologies for the Assessment of Rett Symptoms

\$7,446

**Boston Children's Hospital Rett Syndrome Research
Team**

Rett Clinic Support

\$69,088

Montefiore Rett Syndrome Research Team

Rett Clinic Support

\$50,000

Orrin Devinsky, MD

NYU LANGONE

*Improving Diagnostic Accuracy of Seizure and Non-Seizure Events to Enhance Clinical
Care and Trial Outcomes*

\$50,000

Adrian Bird, PhD / Stuart Cobb, PhD

UNIVERSITY OF EDINBURGH

Data Analysis Equipment

\$87,850

2023

Emerald Innovations

Passive monitoring of Rett patients with Emerald

\$1,106,237

Shawn Liu, PhD

COLUMBIA UNIVERSITY

Multiplex Epigenome Editing to Reactivate & Maintain MECP2 in RTT Neurons

\$482,877

Herophilus

Evaluation of MECP2 Reactivating Effects of Herophilus Lead Small Molecules

\$200,000

David Lieberman, MD, PhD

BOSTON CHILDREN'S HOSPITAL

Boston Children's Hospital Rett Clinic

\$67,345

Samir Mitragotri, PhD

HARVARD UNIVERSITY

Pilot Study to Explore Novel Delivery Technology

\$50,000

John Foxe, PhD

UNIVERSITY OF ROCHESTER

From sensory-perceptual representations to cognitive processing in Rett Syndrome

\$36,690

Coriell Institute

Rett Syndrome biorepository

\$119,461

Harvard Stem Cell Institute

Support for development of patient derived induced pluripotent stem cell lines

\$10,727

2022

Antonio Bedalov, MD, PhD / Kyle Fink, PhD
FRED HUTCHINSON CANCER INSTITUTE / UC DAVIS
Reactivation of MECP2
\$1,090,919

Victor Faundez, PhD
EMORY UNIVERSITY
Systems Biology of Rett Syndrome Gene Therapy Outcomes
\$584,304

Ciitizen
Digital Natural History Study
\$444,000

Joseph Anderson, PhD
UC DAVIS MEDICAL CENTER
Feasibility of a stem cell gene therapy approach for the treatment of Rett Syndrome
\$186,254

Joni N. Saby, PhD / Eric D. Marsh, MD, PhD
CHILDREN'S HOSPITAL OF PHILADELPHIA (CHOP)
Electrophysiological (EEG) Outcome Measures for Rett Syndrome Clinical Trials
\$115,906

David Lieberman, MD, PhD
BOSTON CHILDREN'S HOSPITAL
Clinical Trial Consortium
\$67,821

Stuart Cobb, PhD
UNIVERSITY OF EDINBURGH
Genetic Analysis of the Rett Syndrome Cerebrospinal Fluid Proteome
\$47,014

Coriell Institute
Rett Syndrome biorepository
\$53,612

Harvard Stem Cell Institute
Support for development of patient derived induced pluripotent stem cell lines
\$36,343

The Jackson Laboratory
Generation and phenotypic assessment of mouse models for Rett Syndrome
\$5,620 (additional support)

Bryce Reeve, PhD
DUKE UNIVERSITY SCHOOL OF MEDICINE
Development of the Observer-Reported Communication Ability (ORCA) for Rett
\$15,294

Sasha Djukic, MD, PhD
ALBERT EINSTEIN COLLEGE OF MEDICINE
Support for continuing work at the Rett Syndrome Center
\$25,000

The Jackson Laboratory
Testing of siRNA compounds from Khvorova lab for MECP2 Duplication Syndrome
\$362,930

Davut Pehlivan, MD
TEXAS CHILDREN'S HOSPITAL
Clinical studies in MECP2 Duplication Syndrome as foundation for antisense oligonucleotide drug trials
\$125,000

DSG

Development of the Rett Syndrome Global Registry

\$693,000

James Wilson, MD, PhD

UNIVERSITY OF PENNSYLVANIA

MECP2 gene therapy for Rett Syndrome

\$380,686

Clinical Trial Consortium: David Lieberman, MD, PhD

BOSTON CHILDREN'S HOSPITAL

\$94,176

Bryce Reeve, PhD

DUKE UNIVERSITY SCHOOL OF MEDICINE

Development of the Observer-Reported Communication Ability (ORCA) for Rett syndrome.

\$72,225

Ciitizen

Pilot Study for Digital Natural History Study

\$34,885

Sasha Djukic, MD, PhD

ALBERT EINSTEIN COLLEGE OF MEDICINE

Support for continuing work at the Rett Syndrome Center

\$25,000

2020

Bird / Greenberg / Mandel Labs
EDINBURGH / HARVARD / OREGON HEALTH & SCIENCES
MECP2 Consortium
\$3,359,054

James Wilson, MD, PhD
UNIVERSITY OF PENNSYLVANIA
MECP2 gene therapy for Rett Syndrome
\$765,607

James Wilson, MD, PhD
UNIVERSITY OF PENNSYLVANIA
MECP2 gene therapy for Rett Syndrome, vector production
\$37,999

Stuart Cobb, PhD / Chris Sibley, PhD
UNIVERSITY OF EDINBURGH
RNA trans-splicing therapy in Rett Syndrome
\$235,950

Harvard Stem Cell Institute
Support for development of patient derived induced pluripotent stem cell lines
\$101,912

Michael Elowitz, PhD
CALTECH
A system for dosage-independent control of MECP2 expression in Rett gene therapy
\$212,374

Peter Glazer, PhD / Mark Saltzman, PhD
YALE UNIVERSITY
PNA nanoparticles for gene editing of Rett Syndrome
\$275,000

Alanna Schepartz, PhD
YALE UNIVERSITY
Evaluating cell-permeant miniature proteins (CPMPs) as a strategy for delivering functional MECP2 into model cells and neurons
\$297,716

Joost Gribnau, PhD
ERASMUS MEDICAL CENTER
Human in vitro models for X chromosome reactivation
\$401,000

Antonio Bedalov, MD, PhD
FRED HUTCHINSON CANCER RESEARCH CENTER
Mouse model maintenance
\$20,000

Thorsten Stafforst, PhD
UNIVERSITY OF TUBINGEN
RNA editing for MECP2 mutations via RESTORE
\$359,856

Joseph Jacobson, PhD
MASSACHUSETTS INSTITUTE OF TECHNOLOGY
Correction of MECP2 mutations with engineered ScCas 9 base editors
\$50,000

Beth McCormick, PhD
UMASS MEDICAL SCHOOL
Microbiome study for the advancement of novel nutritional supplements
\$520,316

Sasha Djukic, MD, PhD
ALBERT EINSTEIN SCHOOL OF MEDICINE
Support for continuing work at the Rett Syndrome Center
\$75,000

Ronald Cohn, PhD
THE HOSPITAL FOR SICK CHILDREN
Interrogation of genome editing strategies as a therapeutic modality for MECP2 Duplication Syndrome
\$570,000

Anastasia Khvorova, PhD
UMASS MEDICAL SCHOOL
Development of siRNA based compounds to potently silence MECP2 towards the treatment of MECP2 Duplication Syndrome
\$435,515

The Jackson Laboratory
Generation and phenotypic assessment of mouse models for Rett Syndrome
\$417,690

Coriell Institute
Rett Syndrome biorepository
\$135,000

Misc. Pilot Studies
\$135,522

Emerald Innovations
Passive monitoring of Rett patients with Emerald
\$164,670

2019

**Jonathan Watts, PhD / Scot Wolfe, PhD /
Eric Sontheimer, PhD / Anastasia Khvorova, PhD**
UMASS MEDICAL SCHOOL

RNA and genome editing for treatment of Rett Syndrome

\$2,403,735

**Guoping Feng, PhD / Feng Zhang, PhD /
Robert Desimone, PhD**
MIT/HARVARD/BROAD INSTITUTE

RNA-editing as a gene therapy approach for Rett Syndrome

\$2,332,000

Beam Therapeutics

*Developing a pre-clinical DNA base editing program to precisely correct the genetic
cause of Rett Syndrome in the central nervous system*

\$1,870,660

John Sinnamon, PhD
OREGON HEALTH AND SCIENCE UNIVERSITY

New editing enzymes for RNA

\$345,000

Peter Beal, PhD
UC DAVIS

New molecular tools for directed editing of MECP2 mutations associated with Rett

\$563,870

Stuart Cobb, PhD / Adrian Bird, PhD
UNIVERSITY OF EDINBURGH

Gene Therapy Consortium 2.0

\$653,856

Stuart Cobb, PhD
UNIVERSITY OF EDINBURGH

Purchase of qPCR machine

\$13,945

Andrea Cerase, PhD
QUEEN MARY UNIVERSITY OF LONDON

Reactivation of MECP2 and CDKL5 genes by functional deactivation of Xist RNA

\$351,022

James Wilson, MD, PhD
UNIVERSITY OF PENNSYLVANIA

Gene Therapy Consortium Vector Core

\$131,243

Allan Jacobson, PhD / Jonathan Watts, PhD
UMASS MEDICAL SCHOOL

Read-through of premature termination codons for treatment of Rett Syndrome

\$323,000

Antonio Bedalov, MD, PhD
FRED HUTCHINSON CANCER RESEARCH INSTITUTE

Reactivation of MECP2

\$38,000

Clinical Trial Consortium: David Lieberman, MD, PhD
BOSTON CHILDREN'S HOSPITAL

Reactivation of MECP2

\$74,792

Laurel Joy Gabard-Durnam, PhD
HARVARD UNIVERSITY

Post Doctoral Fellowship, Autism Science Foundation

\$17,500

Hassan Ghasemzadeh, PhD
WASHINGTON STATE UNIVERSITY

Pilot study to examine gait patterns in Rett Syndrome

\$10,000

Sasha Djukic, MD, PhD
ALBERT EINSTEIN COLLEGE OF MEDICINE

Support for continuing work at the Rett Syndrome Center

\$75,000

Huda Zoghbi, MD, PhD
BAYLOR COLLEGE OF MEDICINE

A forward genetic screen to identify druggable modulators of MECP2 levels

\$752,660

James Wilson, MD, PhD
UNIVERSITY OF PENNSYLVANIA

Gene therapy consortium

\$1,585,886

Katherin Meyer, PhD
NATIONWIDE CHILDREN'S HOSPITAL

Optimizing gene therapy for Rett Syndrome

\$152,489

Katherin Meyer, PhD
NATIONWIDE CHILDREN'S HOSPITAL

A gene therapy consortium to develop and evaluate gene therapy approaches in Rett

\$68,515

Stuart Cobb, PhD
UNIVERSITY OF GLASGOW

Additional support for RNA trans-splicing efforts in Rett Syndrome

\$290,000

Rudolf Jaenisch, MD
WHITEHEAD INSTITUTE

Reactivation of MECP2 with epigenome editing tools to rescue Rett Syndrome

\$599,850

Benjamin Philpot, PhD
UNIVERSITY OF NORTH CAROLINA CHAPEL HILL

Pilot study for reactivation of silenced MECP2 by artificial transcription factors

\$145,443

Q State Biosciences

Development of an in-vitro cell system for discovering and evaluating the effects of therapeutic candidates on neurons produced using Rett patient iPS cells

\$498,141

Michael Greenberg, PhD
HARVARD UNIVERSITY

Development of an in-vitro cell system for discovering and evaluating the effects of therapeutic candidates on neurons produced using Rett patient iPS cells

\$55,826

Clinical Trial Consortium: Daniel Tarquinio, DO
CENTER FOR RARE NEUROLOGICAL DISEASES

\$495,000

Clinical Trial Consortium: David Lieberman, MD, PhD
BOSTON CHILDREN'S HOSPITAL

\$395,000

Clinical Trial Consortium: Eric Marsh, MD, PhD
CHILDREN'S HOSPITAL OF PHILADELPHIA

\$487,715

Clinical Trial Consortium: Alan Percy, MD, PhD
UNIVERSITY OF ALABAMA BIRMINGHAM

\$495,000

Clinical Trial Consortium: Jeffrey Neul, MD, PhD
VANDERBILT UNIVERSITY MEDICAL CENTER

\$495,000

Sasha Djukic, MD, PhD
ALBERT EINSTEIN COLLEGE OF MEDICINE

Support for continuing work at the Rett Syndrome Center

\$103,000

Huda Zoghbi, MD
BAYLOR COLLEGE OF MEDICINE

Investigating the potential of antisense oligonucleotide therapy for MECP2 Duplication

\$299,897

2017

Bird / Greenberg / Mandel Labs

EDINBURGH / HARVARD / OREGON HEALTH & SCIENCES

MECP2 Consortium

\$3,454,951

Stuart Cobb, PhD / Steve Gray, PhD / Brian Kaspar, PhD

Gail Mandel, PhD / Alysson Muotri, PhD

UNIVERSITY OF GLASGOW / UNC CHAPEL HILL /

NATIONWIDE CHILDREN'S / OREGON HEALTH & SCIENCE

UC SAN DIEGO

A gene therapy consortium to develop and evaluate gene therapy approaches in Rett

\$1,450,275

Stuart Cobb, PhD

UNIVERSITY OF GLASGOW

Scientific support for gene therapy, splicing therapy and protein therapy programmes in Rett Syndrome

\$210,000

Stuart Cobb, PhD

UNIVERSITY OF GLASGOW

Optimizing MECP2 trans-splicing for human translation

\$330,804

Alysson Muotri, PhD

UNIVERSITY OF CALIFORNIA SAN DIEGO

A drug-screening platform using MECP2-deficient neurons and preclinical testing

\$1,001,000

Alysson Muotri, PhD

UNIVERSITY OF CALIFORNIA SAN DIEGO

Role of an autism-related cytokine in a genetic model of ASD (Autism)

\$12,500

David Katz

CASE WESTERN SCHOOL OF MEDICINE

Preclinical studies of LM22A-4 in mouse models of Rett Syndrome

\$250,000

ArmaGen, Inc.

Protein replacement for Rett Syndrome

\$125,000

Rudolf Jaenisch, MD

WHITEHEAD INSTITUTE FOR BIOMEDICAL RESEARCH

Reversal of Rett phenotype: A screen for compounds that enhance KCC2 expression

\$180,000

Michael Greenberg, PhD

HARVARD UNIVERSITY

Identifying therapeutics for treating Rett Syndrome using nuclear size as a proxy for long gene mis-regulation

\$110,000

Q State Biosciences

Development of an in-vitro cell system for discovering and evaluating the effects of therapeutic candidates on neurons produced using Rett patient iPS cells

\$330,000

Miscellaneous Pilot Projects

\$33,838

Sasha Djukic, MD, PhD

ALBERT EINSTEIN COLLEGE OF MEDICINE

Support for continuing work at the Rett Syndrome Center

\$84,000

Antonio Bedalov, MD, PhD

FRED HUTCHINSON CANCER RESEARCH CENTER

Genetic and pharmacologic reactivation of Mecp2 on the silent X-chromosome as a therapeutic approach to Rett Syndrome

\$824,575

Jeannie Lee, MD, PhD

MASSACHUSETTS GENERAL HOSPITAL / HARVARD

Treating Rett Syndrome by targeting the Xist interactome

\$766,854

Joost Gribnau, PhD

ERASMUS MC

In vivo and in vitro models for X chromosome reactivation

\$177,900

Neurolix, PhD

Clinical development of NLX-101 in Rett Syndrome

\$530,000

Mark Zylka, PhD

UNIVERSITY OF NORTH CAROLINA

High Throughput screen to identify drugs that normalize long gene expression in Rett Syndrome model neurons

\$400,000

Andrew Napper, PhD

NEMOURS DUPONT PEDIATRICS

Discovery and in vivo characterization of compounds promoting MECP2 read-through

\$230,101

Stuart Cobb, PhD

UNIVERSITY OF GLASGOW

Spliceosome-mediated RNA trans-splicing therapy in Rett Syndrome

\$86,208

Stephen Turley, PhD / Adam Lopez, PhD

UNIVERSITY OF TEXAS SOUTHWESTERN MEDICAL CENTER

Exploration of the impact of 2-hydroxypropyl- β -cyclodextrin treatment on lifespan and brain cholesterol metabolism in male mecp2 deficient mice

\$156,180

Miscellaneous Pilot Studies

\$20,000

DiamiR

microRNA biomarkers in Rett Syndrome

\$26,815

David Katz, PhD

CASE WESTERN

Preclinical Studies of LM22A-4 in Mouse Models of Rett Syndrome

\$14,154

The Jackson Laboratory

Development of mouse models

\$42,052

Hermano Igo Krebs, PhD

MIT

Pilot Study

\$8,000

Tim Benke, MD, PhD / Aleksandra Djukic, MD, PhD

Alan Percy, MD / Daniel Tarquinio, DO

CHILDREN'S HOSPITAL COLORADO / MONTEFIORE

UA BIRMINGHAM / CHILDREN'S HEALTHCARE OF ATLANTA

Outcome measures and biomarkers development

\$4,500,000

Michele Fagiolini, PhD

BOSTON CHILDREN'S HOSPITAL

Testing NR2A and NR2B NAMs in mouse models of Rett Syndrome

\$337,336

John Foxe, PhD / Sophie Molholm, PhD

UNIVERSITY OF ROCHESTER / ALBERT EINSTEIN COLLEGE

From sensory-perceptual representations to cognitive processing in Rett Syndrome

\$533,607

Sasha Djukic, MD, PhD

ALBERT EINSTEIN COLLEGE OF MEDICINE

Support for continuing

\$88,000

2015

Monica Justice, PhD

UNIVERSITY OF TORONTO

Identifying genetic modifiers of MECP2 in the mouse

\$715,680

Jeffery Neul, MD, PhD

BAYLOR COLLEGE OF MEDICINE

Identification of genetic modifiers in Rett Syndrome

\$314,456

Jeannie Lee, MD, PhD

MASSACHUSETTS GENERAL HOSPITAL / HARVARD

Re-awakening the silenced normal MECP2 allele with small molecules to treat Rett

\$465,000

Antonio Bedalov, MD, PhD

FRED HUTCHINSON CANCER RESEARCH CENTER

Chemical genetic approach to reactivate the silenced MECP2 gene on the inactive X chromosome

\$290,000

Terry Magnuson, PhD

UNIVERSITY OF NORTH CAROLINA, CHAPEL HILL

Systems genetics approach toward understanding regulation of MECP2 expression

\$200,000

David Katz, PhD

CASE WESTERN RESERVE UNIVERSITY

Preclinical studies of LM22A-4 in mouse models of Rett Syndrome

\$271,700

Adrian Bird, PhD / Michael Greenberg, PhD /

Gail Mandel, PhD

UNIVERSITY OF EDINBURGH / HARVARD UNIVERSITY /

OREGON HEALTH AND SCIENCES UNIVERSITY

MECP2 Consortium

\$250,000

Ali Khoshnan, PhD / Sarkis Mazmanian, PhD

CALTECH

Exploring the link between MECP2 and gut physiology to test a novel probiotic therapy for Rett Syndrome

\$200,000

Lucas Pozzo-Miller, PhD

UNIVERSITY OF ALABAMA BIRMINGHAM

Testing whether LM22A-4 improves hippocampal function in female MECP2 heterozygous mice

\$110,000

Neurolaxis

NLX-101 as a treatment for breathing disorders in Rett Syndrome

\$54,945

Sung-Yon Kim, PhD

LIFE SCIENCE RESEARCH FOUNDATION

Post doctoral fellowship

\$91,500

Steven Gray, PhD

UNIVERSITY OF TEXAS SOUTHWESTERN MEDICAL CENTER

Supplement for gene therapy consortium

\$67,401

Tom Frazier, PhD / David Katz, PhD /

Daniel Sessler, MD, PhD

CASE WESTERN RESERVE UNIVERSITY / CLEVELAND CLINIC

Low-dose ketamine for the treatment of Rett Syndrome

\$1,295,131

Sasha Djukic, MD, PhD

ALBERT EINSTEIN COLLEGE OF MEDICINE

Pharmacological treatment of Rett Syndrome with Lovastatin

\$403,000

Sasha Djukic, MD, PhD

ALBERT EINSTEIN COLLEGE OF MEDICINE

Supplement for copaxone clinical trial

\$47,000

Debra Weese-Mayer, MD / Michael Carroll, PhD

LURIE CHILDREN'S HOSPITAL OF CHICAGO

Outlining the automatic signature of Rett Syndrome

\$157,300

Nurit Ballas, PhD

STONY BROOK UNIVERSITY

Determine the proteome, secretome and transcript changes in astrocytes derived from human Rett patients iPSCs and their effect on interaction with human neurons

\$20,000

DiamiR

microRNA biomarkers in Rett Syndrome

\$6,768

Sasha Djukic, MD, PhD

ALBERT EINSTEIN COLLEGE OF MEDICINE

Support for continuing work at the Rett Syndrome Center

\$140,161

Stephen Turley, PhD

UNIVERSITY OF TEXAS SOUTHWESTERN MEDICAL CENTER

*Exploration of the impact of 2-hydroxypropyl- β -cyclodextrin treatment on lifespan and brain cholesterol metabolism in male *mecp2* deficient mice*

\$20,000

Recursion Pharmaceuticals

High content phenotypic screening of existing drugs for the treatment of Rett

\$25,000

Daniela Tropea, PhD

TRINITY COLLEGE DUBLIN

Expression of nuclear MeCP2 dependent on neuronal stimulation & application of IGF1

\$13,000

Miscellaneous Pilot Projects

Induced Pluripotent Stem Cell and Fibroblast Cell Collections

\$7,000

Huda Zoghbi, MD, PhD

BAYLOR COLLEGE OF MEDICINE

A forward genetic screen to identify druggable modulators of MECP2 levels

\$414,065

Huda Zoghbi, MD, PhD

BAYLOR COLLEGE OF MEDICINE

Antisense oligonucleotide therapy for the treatment of MECP2 Duplication Syndrome

\$230,000

**Adrian Bird, PhD / Michael Greenberg, PhD /
Gail Mandel, PhD**
**UNIVERSITY OF EDINBURGH / HARVARD UNIVERSITY /
OREGON HEALTH AND SCIENCES UNIVERSITY**
MECP2 Consortium
\$3,417,575

**Stuart Cobb, PhD / Steven Gray, PhD /
Brian Kaspar, PhD / Gail Mandel, PhD**
**UNIVERSITY OF GLASGOW / UNIVERSITY OF NORTH
CAROLINA CHAPEL HILL / NATIONWIDE CHILDREN'S
HOSPITAL / OREGON HEALTH AND SCIENCES UNIVERSITY**
Gene Therapy Consortium
\$1,535,942

Michael Green, PhD
UMASS MEDICAL SCHOOL
Testing drugs that modulate X chromosome inactivation to reactivate the silent MECP2
\$750,000

David Katz, PhD
CASE WESTERN RESERVE UNIVERSITY
Preclinical evaluation of therapeutics that modulate the NMDA pathway
\$150,000

Jeannie Lee, MD, PhD
MASS. GENERAL HOSPITAL / HARVARD UNIVERSITY
An oligotherapeutics approach to treat Rett Syndrome
\$100,000

Michela Fagiolini, PhD
BOSTON CHILDREN'S HOSPITAL
Preclinical testing of selective novel NMDA receptor modulators
\$126,741

Mark Bear, PhD
MIT
mGluR5 dependent synaptic protein synthesis in Rett Syndrome
\$45,943

Bruria Ben Zeev, MD
SHEBA MEDICAL CENTER
Copaxone clinical trial
\$197,962

Sasha Djukic, MD, PhD
ALBERT EINSTEIN COLLEGE OF MEDICINE
Copaxone clinical trial
\$412,370

Sasha Djukic, MD, PhD
ALBERT EINSTEIN COLLEGE OF MEDICINE
Support for ongoing work at Rett Syndrome Center
\$72,000

Huda Zoghbi, MD, PhD
BAYLOR COLLEGE OF MEDICINE
A forward genetic screen to identify druggable modulators of MeCP2 levels
\$319,224

Kevin Foust, PhD
NATIONWIDE CHILDREN'S HOSPITAL
RNA interference for the treatment of MECP2 Duplication Syndrome
\$39,340

Benjamin Philpot, PhD

UNIVERSITY OF NORTH CAROLINA CHAPEL HILL

A chemical genetic approach for activating the dormant gene associated with Rett

\$2,204,800

Jonathan Kipnis, PhD

UNIVERSITY OF VIRGINIA

Immune modulation as a new therapeutic approach for Rett Syndrome

\$720,000

John Bissonnette, PhD

OREGON HEALTH AND SCIENCES UNIVERSITY

Respiration in MECP2 deficient mice

\$59,642

Antonio Bedalov, MD, PhD

FRED HUTCHINSON CANCER RESEARCH CENTER

Chemical genetic approach to reactivate the silenced MECP2 gene on the inactive X chromosome

\$55,688

Andrew Pieper, MD, PhD

UNIVERSITY OF TEXAS SOUTHWESTERN MEDICAL CENTER

In vivo identification of pharmacological agents for the treatment of Rett Syndrome

\$69,000

Monica Justice, PhD

BAYLOR COLLEGE OF MEDICINE

Identification of gene modifiers that ameliorate Rett Syndrome

\$757,165

Jay Shapiro, MD, PhD

KENNEDY KRIEGER INSTITUTE

Treatment of osteoporosis in murine Rett Syndrome models

\$20,000

Sasha Djukic, MD, PhD

ALBERT EINSTEIN COLLEGE OF MEDICINE

Support for ongoing work at the Rett Syndrome Center

\$109,771

Greenwood Genetic Center

MECP2 testing

\$3,000

Huda Zoghbi, MD, PhD

BAYLOR COLLEGE OF MEDICINE

Is MECP2 Duplication/Triplication Syndrome reversible?

\$236,200

2012

**Adrian Bird, PhD / Michael Greenberg, PhD /
Gail Mandel, PhD**
**UNIVERSITY OF EDINBURGH / HARVARD UNIVERSITY /
OREGON HEALTH AND SCIENCES UNIVERSITY**

MECP2 Consortium

\$1,840,441

Huda Zoghbi, MD, PhD
BAYLOR COLLEGE OF MEDICINE

Investigating novel therapeutic approaches for Rett Syndrome

\$517,054

Monica Justice, PhD
BAYLOR COLLEGE OF MEDICINE

Identification of gene modifiers that ameliorate Rett Syndrome

\$298,879

Jonathan Kipnis, PhD
UNIVERSITY OF VIRGINIA

Immune modulation as a new therapeutic approach for Rett Syndrome

\$440,000

Jeannie Lee, MD, PhD
**MASSACHUSETTS GENERAL HOSPITAL / HARVARD
UNIVERSITY**

*A high-throughput screen to identify compounds that reactivate the functional MECP2
allele in Rett Syndrome*

\$300,000

Mark Bear, PhD
MIT

mGluR5 dependent synaptic protein synthesis in Rett Syndrome

\$85,896

Jeffrey Macklis, MD, PhD
HARVARD UNIVERSITY

Vitamin D therapy for MECP2 target Irak1/NFkB dysregulation

\$35,352

Sasha Djukic, MD, PhD
ALBERT EINSTEIN COLLEGE OF MEDICINE

Support for ongoing work at Rett Syndrome Center

\$66,710

Benjamin Philpot, PhD
UNIVERSITY OF NORTH CAROLINA CHAPEL HILL

\$10,000

John Bissonnette, PhD
OREGON HEALTH AND SCIENCES UNIVERSITY

Respiration in MECP2 deficient mice

\$15,147

Ronald Crystal, MD, PhD

WEILL MEDICAL COLLEGE OF CORNELL UNIVERSITY

AAV mediated gene transfer for the treatment of Rett Syndrome

\$605,121

Sasha Djukic, MD, PhD

ALBERT EINSTEIN COLLEGE OF MEDICINE

Support for ongoing work at Rett Syndrome Center

\$36,654

Brian Kaspar, PhD / Gail Mandel, PhD

NATIONWIDE CHILDREN'S / OREGON HEALTH & SCIENCES

AAV9 gene therapy for Rett Syndrome

\$80,000

Rett Syndrome Clinic

UNIVERSITY OF SOUTHERN CALIFORNIA

Support for Rett Syndrome Clinic

\$22,022

Antonio Bedalov, MD, PhD

FRED HUTCHINSON CANCER RESEARCH CENTER

Genetic approach to reactivate the silenced MECP2 gene on inactive X chromosome

\$250,000

Jonthan Kipnis, PhD

UNIVERSITY OF VIRGINIA

Immune modulation as a new therapeutic approach for Rett Syndrome

\$187,000

Huda Zoghbi, MD, PhD

BAYLOR COLLEGE OF MEDICINE

Interventional trials in mice models of Rett Syndrome and MECP2 disorders

\$100,000

Marisa Bartolomei, PhD

UNIVERSITY OF PENNSYLVANIA

Analysis of epigenetic modifications of the MECP2 locus

\$41,255

2010

Monica Justice, PhD

BAYLOR COLLEGE OF MEDICINE

Identification of gene modifiers that ameliorate Rett Syndrome

\$236,038

Stavros Lomvardas, PhD

UNIVERSITY OF CALIFORNIA SAN FRANCISCO

Insight into MECP2 function raises therapeutic possibilities for Rett Syndrome

\$140,000

Huda Zoghbi, MD, PhD

BAYLOR COLLEGE OF MEDICINE

*Interventional trials in mice models of Rett Syndrome
and MECP2 disorders*

\$100,000

Marisa Bartolomei, PhD

UNIVERSITY OF PENNSYLVANIA

Analysis of epigenetic modifications of the MECP2 locus

\$40,000

Sasha Djukic, MD, PhD

ALBERT EINSTEIN COLLEGE OF MEDICINE

Support for continuing work at the Rett Syndrome Center

\$36,645

2009

Adrian Bird, PhD

BAYLOR COLLEGE OF MEDICINE

Identification of gene modifiers that ameliorate Rett Syndrome

\$1,380,000

Andrew Pieper, MD, PhD

UNIVERSITY OF TEXAS SOUTHWESTERN MEDICAL CENTER

In vivo identification of pharmacological agents for the treatment of Rett syndrome

\$505,000

Monica Justice, PhD

BAYLOR COLLEGE OF MEDICINE

Identification of gene modifiers that ameliorate Rett Syndrome

\$253,000

Antonio Bedalov, MD, PhD

FRED HUTCHINSON CANCER RESEARCH CENTER

Chemical genetic approach to reactivate the silenced MECP2 gene on the inactive X chromosome

\$140,000

2008