




rett syndrome
research trust

RETT SYNDROME RESEARCH TRUST AWARDS BY YEAR

/ TOTAL AWARDS **\$62 MILLION** (2008-2021 YTD)

2021 (to date)

TOTAL AWARDS \$1,469,143

Antonio Bedalov / Kyle Fink

Fred Hutchinson Cancer Research Institute /
University of California Davis
Reactivation of MECP2
\$1,090,919

Jackson Laboratories

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

Bryce Reeve, PhD

Duke University School of Medicine
Development of the Observer-Reported Communication
Ability (ORCA) for Rett Syndrome
\$15,294

2020

TOTAL AWARDS \$1,299,972

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

Citizen

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

Due to the global pandemic and the ensuing fundraising uncertainties we were cautious in taking on additional commitments. Furthermore we undertook a detailed analysis of our portfolio and were able to reduce our commitments by \$6 million. This reduction allows us to focus our resources on curative projects with the greatest likelihood of success in the nearer term.

2019

TOTAL AWARDS \$8,570,181

Adrian Bird, PhD / Michael Greenberg, PhD / Gail Mandel, PhD

University of Edinburgh / Harvard University /
Oregon Health and Sciences University
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

California Institute of Technology

A system for dosage-independent control of MECP2 expression in Rett Syndrome 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, PhD

Fred Hutchinson Cancer Research Center

Mouse model maintenance

\$20,000

Thorsten Stafforst, PhD

University of Tübingen

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

The Jackson Laboratory

Generation and phenotypic assessment of mouse models for Rett Syndrome

\$417,690

Coriell Institute

Rett Syndrome biorepository

\$135,000

Emerald Innovations

Passive monitoring of Rett patients with Emerald

\$164,670

Beth McCormick, PhD

University of Massachusetts 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

Miscellaneous Pilot Studies

\$135,522

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

University of Massachusetts Medical School

Development of siRNA based compounds to potently silence MECP2 towards the treatment of MECP2 Duplication Syndrome

2018

TOTAL AWARDS **\$9,956,283**

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

University of Massachusetts Medical School
RNA and genome editing for treatment of Rett Syndrome
\$2,403,735

**Guoping Feng, PhD / Feng Zhang, PhD /
Robert Desimone, PhD**

Massachusetts Institute of Technology / Broad Institute /
Harvard University
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

University of California, Davis
*New molecular tools for directed editing of MECP2 mutations associated
with Rett Syndrome*
\$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

University of Massachusetts Medical School
*Read-through of premature termination codons for treatment
Rett Syndrome*
\$323,000

Antonio Bedalov

Fred Hutchinson Cancer Research Institute
Reactivation of MECP2
\$38,000

Clinical Trial Consortium

David Lieberman, MD, PhD
Boston Children's Hospital
\$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

2017

TOTAL AWARDS **\$6,166,762**

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 Syndrome

\$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 for Biomedical Research

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 Syndrome

\$299,897

2016

TOTAL AWARDS **\$7,571,438**

Adrian Bird, PhD / Michael Greenberg, PhD / Gail Mandel, PhD

University of Edinburgh / Harvard University / Oregon Health and Sciences University

MECP2 Consortium

\$3,454,921

Stuart Cobb, PhD / Steve Gray, PhD / Brian Kaspar, PhD / Gail Mandel, PhD / Alysson Muotri, PhD

University of Glasgow / University of North Carolina Chapel Hill / Nationwide Children's Hospital / Oregon Health and Science University

/ University of California San Diego

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

\$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

University of California San Diego

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

\$1,001,000

Alysson Muotri

University of California San Diego

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

\$12,500

David Katz

Case Western Reserve University 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

2015

TOTAL AWARDS **\$8,741,782**

Antonio Bedalov, 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, PhD

Massachusetts General Hospital / Harvard University

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 Reserve University

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

\$14,154

Jackson Labs

Development of mouse models

\$42,052

Hermano Igo Krebs, PhD

Massachusetts Institute of Technology

Pilot Study

\$8,000

Tim Benke, PhD / Aleksandra Djukic, PhD / Alan Percy, PhD / Daniel Tarquinio, PhD
Children's Hospital Colorado / Montefiore Medical Center /
University of Alabama Birmingham / Children's Healthcare of Atlanta
Outcome measures and biomarkers development
\$4,500,000

John Foxe, PhD / Sophie Molholm, PhD
University of Rochester / Albert Einstein College of Medicine
From sensory-perceptual representations to cognitive processing in Rett Syndrome
\$533,607

Sasha Djukic, MD, PhD
Albert Einstein College of Medicine
Support for continuing work at the Rett Syndrome Center
\$88,000

2014

TOTAL AWARDS **\$5,809,107**

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, PhD
Massachusetts General Hospital / Harvard University
Re-awakening the silenced normal MECP2 allele with small molecules to treat Rett Syndrome
\$465,000

Antonio Bedalov, 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 Science University
MECP2 Consortium
\$250,000

Ali Khoshnan, PhD / Sarkis Mazmanian, PhD
California Institute of Technology
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

Neurolix
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 Syndrome
\$25,000

Daniela Tropea, PhD

Trinity College Dublin
 Expression of nuclear MeCP2 is dependent on neuronal stimulation and application of IGF1
\$13,000

Miscellaneous Pilot Projects

\$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

2013

TOTAL AWARDS **\$7,167,097**

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

University of Massachusetts 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, PhD

Massachusetts 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

Massachusetts Institute of Technology
 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

2012

TOTAL AWARDS **\$4,235,266**

Benjamin Philpot, PhD

University of North Carolina Chapel Hill
A chemical genetic approach for activating the dormant gene associated with Rett Syndrome
\$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, 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

2011

TOTAL AWARDS **\$3,609,479**

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****Jonthan Kipnis, PhD**

University of Virginia

*Immune modulation as a new therapeutic approach for Rett Syndrome***\$440,000****Jeannie Lee, 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**

Massachusetts Institute of Technology

*mGluR5 dependent synaptic protein synthesis in Rett Syndrome***\$85,896****Jeffrey Macklis, 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 Bissonette, PhD**

Oregon Health and Sciences University

*Respiration in MECP2 deficient mice***\$15,147**

2010

TOTAL AWARDS \$1,322,052**Ronald Crystal, MD, PhD**

Weill Medical College of Cornell University

*AAV mediated gene transfer for the treatment of Rett Syndrome***\$605,121****Brian Kaspar, PhD / Gail Mandel, PhD**

Nationwide Children's Hospital / Oregon Health and

Sciences University

*AAV9 gene therapy for Rett Syndrome***\$80,000****Antonio Bedalov, PhD**

Fred Hutchinson Cancer Research Center

*Chemical genetic approach to reactivate the silenced MECP2 gene on the 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****Sasha Djukic, MD, PhD**

Albert Einstein College of Medicine

*Support for ongoing work at Rett Syndrome Center***\$36,654****Rett Syndrome Clinic**

University of Southern California

*Support for Rett Syndrome Clinic***\$22,022**

2009

TOTAL AWARDS **\$552,683**

Monica Justice, PhD

Baylor College of Medicine

Identification of gene modifiers that ameliorate Rett Syndrome

\$236,038

Stavros Lomvardas

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

2008

TOTAL AWARDS **\$2,278,000**

Adrian Bird, PhD

Baylor College of Medicine

Identification of gene modifiers that ameliorate Rett Syndrome

\$236,038

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, PhD

Fred Hutchinson Cancer Research Center

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

\$140,000