PRESS RELEASE:Rett Syndrome Research Trust Awards $6.2 Million in 2017 to Speed a Cure for Rett Syndrome
新闻稿：雷特综合症研究信托基金在2017年为雷特综合症的治疗提供了620万美元

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2018年一月30日，媒体联系人， 莫妮卡. 科恩拉德 - 雷特综合症研究信托基金执行董事：203.445.0041, monica@rsrt.org

TRUMBULL, CT — The Rett Syndrome Research Trust (RSRT) announced today research awards of $6.2 million made in 2017. RSRT's sole and urgent goal is to abolish Rett Syndrome and related disorders. To that end, RSRT has awarded $47 million to research, both basic and clinical, since its launch in 2008.
特朗布尔，CT——雷特综合症研究信托基金（RSRT）今天宣布了2017年完成了620万美元的研究资金。RSRT的唯一和迫切的目标是消除雷特综合症和相关疾病。为了实现这一目标，RSRT自2008年推出以来，已经获得了4700万美元的基础研究和临床研究。

Rett Syndrome is caused by mutations in a gene called MECP2 that result in a cascade of devastating symptoms that worsen over time. These symptoms begin to manifest in early childhood and leave Rett sufferers dependent on 24-hour-a-day care for the rest of their lives.
雷特综合症是由一种叫做MECP2的基因的突变引起的，它会导致一系列毁灭性的症状，随着时间的推移会恶化。这些症状在儿童早期就开始显现，并让雷特的患者在他们的余生中依赖24小时的日常护理。

Last year RSRT implemented a three-year, $33 million, strategic research plan, Roadmap to a Cure that includes four key components: Cure, Treat, Enable, Learn. The 2017 awards strengthen and enhance the Cure and Enable components of the Roadmap. The Cure component features four bold approaches that attack Rett at its core: gene therapy, reactivation of the MECP2 on the inactive X chromosome, RNA modification and protein replacement. The Enable component ensures that potential therapeutics can be successfully measured both in the lab and the clinic; through the generation of cell lines and other valuable resources necessary to test new therapies at the bench, and through the outcome measures and biomarkers development initiative to identify key parameters that predict or correlate with efficacy in patients.
去年，RSRT实施了一项为期3年、3300万美元的战略研究计划，这是一项治疗方案的路线图，其中包括四个关键组成部分：治愈、治疗、有效、学习。2017年的资金巩固并加强了路线图的治疗和有效的组成部分。该治愈组成部分的主要特点是四种大胆的方法，它的核心是：基因疗法，激活了非活性X染色体上的MECP2基因，RNA修饰和蛋白质替换。有效组成部分是确保潜在的治疗可以在实验室和诊所成功地进行测量; 通过细胞株的生成和其他有价值的资源来测试新的治疗方法，并通过结果测量和生物标记发展计划来确定关键的参数，这些参数可以预测或与病人的疗效相关联。

“RSRT’s mission is to develop new therapies and a cure for Rett syndrome. The remarkable progress in the past year has elicited a palpable sense of optimism that a cure for Rett Syndrome is possible. Since it is not possible to predict where therapeutic breakthroughs will first occur, RSRT seeks to increase the odds of success by pursuing the most promising therapeutics in parallel. Thus, our key metric for measuring success is the breadth and strength of our therapeutic development pipeline,” stated Randall Carpenter, MD, Chief Scientific Officer of RSRT.
“RSRT的使命是开发新的治疗方法和治愈雷特综合症。”过去一年取得的显著进步，让人们产生了一种明显的乐观情绪，即治愈雷特综合症是可能的。由于无法预测哪一种治疗的突破将会在何处发生，RSRT以并行方式寻求最有前途的治疗方法来增加成功的几率。因此，我们衡量成功的关键指标是我们的治疗发展管道的广度和强度，”RSRT的首席科学官员兰德尔卡彭特说。

Highlights of RSRT’s 2017 awards:
RSRT 2017年资金的亮点：

- An award of almost $1.6 million was made to James Wilson, MD, PhD who joins our Gene Therapy Consortium with a primary focus of improving delivery of genes to the brain. Dr. Wilson has been a pioneer and a leader in the gene therapy field for three decades making a number of seminal discoveries. He runs the largest academic based gene therapy program in the world. Dr. Wilson’s involvement in our Consortium will leverage deep expertise and resources as well as ongoing gene therapy collaborations with several major pharmaceutical and biotech partner

获得了近160万美元的资金，是由医学博士詹姆斯威尔逊授予的，他加入了我们的基因治疗联盟，主要致力于改善基因向大脑的传递。威尔逊博士在基因治疗领域做了30年的先驱和领导者，他做出了一系列开创性的发现。他是世界上最大的基于学术的基因治疗项目。威尔逊博士参与我们的研究小组将利用专业知识和资源以及与几家主要制药和生物技术合作伙伴进行基因治疗合作

- Stuart Cobb, PhD is also part of our Gene Therapy Consortium. Beyond traditional gene therapy the Cobb lab is also pursuing a therapy called Spliceosome-Mediated RNA Trans-splicing (SMaRT). This technology would enable the correction of the vast majority of MECP2 mutations by splicing out exons 3 and 4 and replacing with mutation free exons. The recent award of $290,000 provides funding to accelerate this exciting technology. The SMaRT reagents will need to be delivered to the brain via vector so our gene therapy efforts will also be very relevant for this approach.

斯图尔特.科布博士也是我们基因治疗联盟的成员。除了传统的基因疗法，科布实验室还在研究一种叫“拼接-介导”的RNA转接（SMaRT）疗法。这项技术将通过拼接外显子3和4并替换突变的无突变外显子，来纠正绝大多数MECP2突变。最近为加速这项激动人心的技术提供29万美元的资金。SMaRT试剂需要通过载体传递到大脑，因此我们的基因治疗工作也将与这种方法密切相关。

- An award of $600,000 was made to Rudolf Jaenisch, PhD that leverages a novel technology, CRISPR, that has taken the scientific world by storm. CRISPR will be used

to deliver epigenetic molecules to the inactive MECP2 with the goal of activating it by synergistically removing methyl tags and adding acetyl tags. It's important to note that the CRISPR guides and epigenetic molecules will need to be delivered to patients via vectors in much the same way as gene therapy. Any valuable insights learned from our gene therapy work will be applicable to this strategy as well.

鲁道夫贾.恩斯奇博士获得了60万美元的资金，他利用了一项新技术——CRISPR，这一技术已经席卷了科学界。CRISPR将被用于将表观遗传分子输送到非活性的MECP2，目标是通过协同移除甲基标记和添加乙酰基标签来激活它。需要注意的是，CRISPR指南和表观遗传分子将需要通过载体传递给患者，就像基因疗法一样。从我们的基因治疗工作中获得的任何有价值的见解也将适用于这一策略。

- Funding of $142,000 will fund a pilot study in the lab of Ben Philpot, PhD to explore reactivation of the silent MECP2 with a novel approach using genetic tools, called zinc finger proteins, that directly interact with the chromosome region responsible for switching the MECP2 gene on and off, called the promoter. In diseased Rett neurons the promoter of the normal MECP2 gene copy is kept in the "off" state by certain modifications that prevent binding of proteins, called transcription factors. Zinc finger proteins are designed to specifically target the MECP2 promoter and locally deliver transcription factors to override the silenced state and jump-start gene activation. Ultimately, promising zinc finger/transcription factor complexes will need to be delivered to the brain. Vectors being developed by our Gene Therapy Consortium can be easily adapted to enable delivery.

142000美元的资金将基金试点研究实验室的本.菲尔波特博士探索激活沉默的MECP2新方法利用基因工具,称为锌指蛋白,直接与负责开关MECP2基因的染色体区域,称为启动子。在患病的雷特神经元中，正常MECP2基因拷贝的启动子被保留在"关闭"状态，通过某些修饰来阻止蛋白质的结合，称为转录因子。锌指蛋白的设计是专门针对MECP2启动子，并在局部传送转录因子来覆盖沉默状态并启动基因的激活。最终，有希望的锌指/转录因子复合物需要传递给大脑。我们的基因治疗联盟开发的载体可以很容易地适应于有效的传递。

- An award of $500,000 was made to the biotechnology company, Q-State Biosciences, and $54,000 to collaborator Michael Greenberg, PhD to advance our goal of precision medicine for individuals with Rett Syndrome. The company has developed methods that use cells from individuals with mutations in MECP2, to study disease, explore possible treatments, and, hopefully predict an individual's response to specific therapeutics. In the present study, the processes for generating the "brain in a dish" will be optimized and the changes that result from Rett mutations will be fully characterized. Drugs and biological therapeutics will then be tested in these cells. This platform has implications for all of our programs – from our curative programs such as gene therapy, RNA editing, MECP2 reactivation and RNA trans-splicing to downstream targeted drugs such as ketamine.

向生物科技公司，Q-State生物科学公司提供50万美元资金，以及合作伙伴迈克尔格林伯格获得了5万4千美元的资金，以推进我们为患有雷特综合症的患者提供精确药物的目标。该公司开发了一些方法，利用MECP2基因突变的个体的细胞，研究疾病，探索可能的治疗方法，并

有望预测个人对特定疗法的反应。在目前的研究中，将对生成“培养皿里的大脑”的过程进行优化充分利用，并将由来自雷特基因突变的变化进行充分的显示。药物和生物疗法将在这些细胞中进行测试。这个平台对我们所有的项目都有影响——从我们的治疗方案，如基因治疗、RNA编辑、MECP2再激活和RNA转接到氯胺酮等下游靶向药物。

- An award of almost $2.4 million has made possible the launch of the Rett Syndrome Clinical Trial Consortium with the primary objective of providing expert Rett physicians with the personnel and resources necessary to execute high-quality clinical trials in a timely and efficient manner. This Consortium will initially fund the following Rett Syndrome Clinics: Boston Children’s Hospital; Drs. David Lieberman & Mustafa Sahin | Children’s Hospital of Philadelphia; Dr. Eric Marsh | Vanderbilt University Medical Center; Dr. Jeff Neul | Children’s of Alabama; Dr. Alan Percy | Center for Rare Neurological Diseases (Atlanta); Dr. Daniel Tarquinio

近240万美元的资金使雷特综合症临床试验联盟的成立成为可能，其主要目标是为专业的雷特医生提供必要的人力和资源，以及时、有效的方式实施高质量的临床试验。该联盟最初将资助下列雷特综合征诊所：波士顿儿童医院; 费城儿童医院的大卫.利伯曼和穆斯塔法.沙欣博士; 埃里克.马什,范德比尔特大学医学中心; 阿拉巴马州儿童医院的杰夫.纽尔博士; 艾伦.珀西博士,罕见神经疾病中心（亚特兰大）; 丹尼尔.塔尔奎尼奥博士

“I have an 8-year-old daughter with Rett Syndrome. What I care about is research that can profoundly change her life and the lives of all children and adults who fight this disorder so bravely every day. RSRT’s 2017 research awards aim to do just that, and I’m honored and proud to be associated with them. Every single family that fundraises for us and every one of our donors make this research possible, and for that they have my deepest thanks. These awards are also a testament to how Monica Coenraads and Randy Carpenter have recruited a team of world-class scientists to attack Rett head on,” states Tim Freeman, RSRT’s Chief Development Officer.
“我有一个8岁的女儿患有雷特综合症。我所关心的是能够深刻地改变她的生活和所有儿童和成年人的生活，他们每天都勇敢地与这种疾病抗争。RSRT的2017年研究资金旨在做到这一点，我很荣幸也很自豪能与他们联系在一起。每一个为我们筹款的家庭和我们的每一个捐助者都使这项研究成为可能，为此他们向我表示深深的谢意。这些资金也证明了莫妮卡.柯恩拉德和兰迪.卡彭特是如何招募了一个世界级的科学家团队来攻克雷特的，”RSRT的首席发展官员提姆.弗里曼说。

List of 2017 Awards by Roadmap to a Cure category:
针对一个治愈种类“路线图”的2017年资金清单：
CURE
治愈
Gene Therapy Consortium James Wilson (University of Pennsylvania) $1,585,886
基因治疗联盟詹姆斯.威尔逊（宾夕法尼亚大学）1 585,886美元
Optimizing Gene Therapy for Rett Syndrome Kathrin Meyer (Nationwide Children’s Hospital) $221,004
为雷特综合症优化基因疗法的凯瑟琳.梅耶（全国儿童医院）提供221,004美元

Additional Support for RNA-trans splicing Efforts in Rett Syndrome Stuart Cobb (University of Edinburgh) $290,000
对雷特综合症的RNA-trans拼接工作的斯图尔特.科布（爱丁堡大学）提供额外支持，290，000美元
Reactivation of MECP2 with Epigenome Editing Tools to Rescue Rett Syndrome Rudolf Jaenisch (Whitehead Institute for Biomedical Research) $599,850
用表观基因组编辑工具重新激活MECP2基因，以拯救雷特综合症患者的鲁道夫.耶尼施（怀特海德生物医学研究所）599，850美元
Pilot Study for Reactivation of Silenced Mecp2 by Artificial Transcription Factors Ben Philpot (University of North Carolina – Chapel Hill) $141,912
通过人工转录因子重新激活沉默Mecp2的本.菲尔波特（北卡罗莱那大学-教堂山分校） 141，912美元的资金，。

ENABLE
有效
Development of an In-Vitro Cell System For Discovering and Evaluating the Effects of Therapeutic Candidates on Neurons Produced Using Rett Patient Ips Cells Q State BioSciences and Michael Greenberg (Harvard University) $553,967
开发一种体外细胞系统，用于发现和评估在神经元上产生的候选治疗药剂的效果，使用雷特病人干细胞Q国家生物科学和迈克尔.格林伯格（哈佛大学）553,967美元
Clinical Trial Consortium David Lieberman & Mustafa Sahin, Eric Marsh, Jeff Neul, Alan Percy, Daniel Tarquinio $2,375,000
临床试验联盟大卫.利伯曼和穆斯塔法.沙欣，埃里克.马什，杰夫.纽尔，艾伦.珀西，丹尼尔.塔尔奎尼奥2,375,000美元

Tri-State Rett Syndrome at Montefiore Aleksandra Djukic $103,000
蒙特菲奥里的三太雷特综合症103，000美元

MECP2 Duplication Syndrome
MECP2重复综合症

Investigating the Potential of Antisense Oligonucleotide Therapy for MECP2 Duplication Syndrome Huda Zoghbi (Baylor College of Medicine) $299,898
摘要探讨了抗敏性寡核苷酸治疗的潜力，针对对MECP2重复综合症的胡达.祖比（贝勒医学院）299，898美元。

Our Partners RSRT’s partners in supporting this work include every affected family that fundraises for us and every individual and organizational donor who shares with us a sense of urgency to change the lives of all who suffer with Rett and related disorders. Some of the parents’ organizations around the world that play a key role in making this research possible include Reverse Rett (UK), Rett Syndrome Research & Treatment Foundation (Israel), Rett Syndrome Ireland, and Rett Syndrom Deutschland. Major parent-led organizations in the U.S. that support our work include Girl Power 2 Cure, Eva Fini Fund at RSRT, Kate Foundation for

Rett Syndrome Research, New Jersey Rett Syndrome Association, and Rett Syndrome Association of Massachusetts.
我们的合作伙 - RSRT支持这项工作的合作伙伴包括每一个为我们筹款的家庭，以及每个与我们共享的个人和组织捐助者，他们与我们一起分享改变所有遭受雷特和相关疾病折磨的人的生命的紧迫感。世界各地的一些家长组织在使这项研究成为可能的过程中发挥了关键作用，包括雷特（英国）、雷特综合症研究与治疗基金会（以色列）、雷特综合症爱尔兰和德国雷特综合症。美国主要的家长主导组织支持我们的工作，包括女童权利2治疗、RSRT的爱娃菲尼基金、雷特综合症研究的凯特基金会、新泽西雷特综合症协会和马萨诸塞州的雷特综合症协会。

About the Rett Syndrome Research Trust The Rett Syndrome Research Trust (RSRT) is a nonprofit organization with a highly personal and urgent mission: a cure for Rett Syndrome and related MECP2 disorders. RSRT operates at the nexus of global scientific activity enabling advances in knowledge and driving innovative research. In March of 2017 RSRT announced Roadmap to a Cure, a threeyear, $33 million strategic research plan. The plan prioritizes four curative approaches with gene therapy as our lead program. In June of 2017 the biotechnology company, AveXis, announced its intent to advance RSRT’s gene therapy program to clinical trials. Since 2008, RSRT has awarded $47 million to research. To learn more, please visit www.reverserett.org
雷特综合症研究信托基金（RSRT）是一个非盈利性组织，具有高度个人化和紧急的使命：治疗雷特综合症和相关的MECP2疾病。RSRT在全球科学活动的联系中发挥作用，促进知识的进步和推动创新研究。在2017年3月，RSRT宣布了一项为期3年、3300万美元的战略研究计划。该计划将四种治疗方法列为我们的主要项目。在2017年6月，生物技术公司AveXis宣布了将RSRT基因治疗方案推向临床试验的意图。自2008年以来，RSRT已经为研究提供了4700万美元。想了解更多，请访问www。reverserett.org

About Rett Syndrome Rett Syndrome is a genetic neurological disorder that affects predominately girls but can rarely also affect boys. It strikes randomly, typically at the age of 12 to 18 months, and is caused by random mutations of the MECP2 gene on the X chromosome. Rett Syndrome is devastating as it deprives young children of speech, hand use, and normal movement often including the ability to walk. Symptoms can also include anxiety, seizures, tremors, breathing difficulties, and severe gastrointestinal issues. While their bodies suffer, it is believed that their cognitive abilities remain largely intact. Although most children survive to adulthood, they require total round-the-clock care.
雷特综合症是一种遗传性神经紊乱症，主要影响女孩，但也影响少数男孩。它是随机的，通常是在12到18个月的时候，是由X染色体上的MECP2基因的随机突变引起的。雷特综合症是毁灭性的，因为它剥夺了幼儿的语言、用手和正常的运动，包括走路的能力。症状还包括焦虑、癫痫、震颤、呼吸困难和严重的肠胃问题。当他们的身体遭受痛苦时，他们的认知能力基本上保持不变。尽管大多数孩子能活到成年，但他们需要全天24小时的护理。