PRESS RELEASE: James Wilson, MD, PhD, from the University of Pennsylvania, Joins the Rett Syndrome Research Trust Gene Therapy Consortium
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2018年一月25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) is pleased to announce that James Wilson, MD, PhD, director of the University of Pennsylvania Gene Therapy Program and the Rose H. Weiss Orphan Disease Center, has joined our Gene Therapy Consortium.
特朗布尔，CT——雷特综合症研究信托（RSRT）很高兴地宣布，詹姆斯威尔逊，医学博士，宾夕法尼亚大学基因疗法项目主任，罗斯h.维斯孤儿疾病中心，已经加入我们的基因治疗联盟。

Rett Syndrome is a severely disabling childhood neurological disorder caused by mutations in a gene called MECP2, which leads to diminished MeCP2 protein levels. Rett is not neurodegenerative and preclinical research has shown that the disorder is dramatically reversible once protein levels are restored. Thus, therapeutics that restore MeCP2 protein levels have the potential to provide profound benefit and potentially cure Rett Syndrome. In addition to delivery of the MECP2 gene, alternative curative approaches currently funded by RSRT such as RNA editing, RNA trans-splicing, and MECP2 reactivation require delivery of genes to the brain, and may also be considered “gene therapies”.
雷特综合症是一种严重致残的儿童神经系统疾病，它由一种叫做MECP2的基因突变引起，这导致MECP2蛋白水平下降。雷特并不是神经退化和临床前研究表明，一旦蛋白质水平恢复，这种紊乱就会急剧逆转。因此，恢复MeCP2蛋白水平的疗法有可能带来深远的益处，并有可能治愈雷特综合症。除了提供MECP2基因之外，目前由RSRT资助的替代疗法，如RNA编辑、RNA转接和MECP2重新激活，都需要将基因传递给大脑，也可以被认为是“基因疗法”。

In 2014 RSRT launched the original Gene Therapy Consortium, a collaboration of two gene therapy labs, Brian Kaspar, PhD (then at Nationwide Children’s Hospital, now at AveXis) and Steven Gray, PhD (then at University of North Carolina Chapel Hill, now at University of Texas Southwestern) and two MECP2 labs, Gail Mandel, PhD (Oregon Health and Sciences University) and Stuart Cobb, PhD (then at University of Glasgow, now at University of Edinburgh). Based on the encouraging data generated by this Consortium the biotech company, AveXis, is now advancing the lead MECP2 gene therapy candidate toward clinical trials.
2014年RSRT发起了最初的基因疗法的联盟,两个基因疗法的合作实验室,布莱恩·卡斯帕·博士(当时在国内儿童医院,现在在AveXis)和史蒂文•格雷博士(当时在北卡罗莱纳大学教堂山分校,现在在德克萨斯大学西南)和两个MECP2实验室,盖尔·曼德尔,博士(俄勒冈健康与科学大学)和斯图尔特·科布博士(当时在格拉斯哥大学,现在在爱丁堡大学)。基于该联盟所产生的令人鼓舞的数据，生物科技公司 - AveXis现在正将领先的MECP2基因治疗候选方案推向临床试验。

Dr. Wilson has been a pioneer and a leader in the gene therapy field for three decades and has played a pivotal role in the renaissance that the field is currently experiencing. He made a number of seminal discoveries including the discovery and development of the adeno-associated virus (AAV) that will be used by AveXis in the upcoming clinical trial. Dr. Wilson directs the Gene Therapy Program at The University of Pennsylvania, where one prominent research focus is improving delivery of genes to the brain.
威尔逊博士在基因治疗领域已经有30年经验的先驱和领导者，并且在这个领域正在经历的文艺复兴式的变革中发挥了关键作用。他做了许多开创性的发现，包括发现和开发腺相关病毒（AAV），这将在即将到来的临床试验中被AveXis所使用。威尔逊博士是宾夕法尼亚大学基因治疗项目的负责人，该项目的一个突出研究重点是改善大脑的基因传递。

“Recent advances by RSRT Consortia researchers are providing insights for novel genetic therapeutics that will very likely benefit from enhanced delivery and regulation. I believe this exciting collaboration will leverage our collective expertise and increase our ability to develop novel and improved gene therapies,” stated Dr. Wilson.
“RSRT联盟的研究人员最近的研究成果为新型基因疗法提供了一些见解，这些疗法很可能从增强的传递和规则中获益。我相信这种令人兴奋的合作将会利用我们的集体经验，并提高我们开发新的和改良基因疗法的能力。”威尔逊博士说。

Beyond traditional gene therapy, RSRT is also pursuing RNA approaches and biological approaches to reactivate the silent MECP2 gene on the inactive X chromosome. The reagents for these approaches will also have to be delivered into cells via vectors so lessons learned via the Gene Therapy Consortium will be highly relevant.
除了传统的基因疗法，RSRT还在研究RNA方法和生物方法，以激活不活跃的X染色体上的沉默的MECP2基因。这些方法的试剂也必须通过载体传送到细胞中，因此通过基因治疗联盟获得的经验将是高度关联的。

“Dr. Wilson’s pioneering research has produced numerous breakthrough scientific discoveries that serve as the foundation for the success of many gene therapy programs. Current efforts to improve brain delivery in his lab are bolstered by major funding from several pharmaceutical and biotech partners and leveraged by collaboration with the expert scientists in these companies. This collaboration with Dr. Wilson has potential to accelerate development of our entire portfolio of gene therapies,” commented Randall Carpenter, MD, Chief Scientific Officer of RSRT.
”威尔逊博士的开创性研究已经产生了许多突破性的科学发现，这些发现为许多基因治疗项目的成功奠定了基础。目前在他的实验室里改善大脑的工作得到了一些制药和生物科技合作伙伴的资助，并通过与这些公司的专业科学家的合作大展拳脚。与威尔逊博士的合作有可能加速我们整个基因疗法组合的开发。”RSRT的首席科学官员兰德尔卡彭特评论道。

The Wilson lab is receiving an award of $1.6 million over two years for research specific to Rett Syndrome. The total RSRT investment in the Gene Therapy Consortium is $4.7 million.
威尔逊实验室将在两年内获得160万美元的奖励，特定用于雷特综合症的研究。基因治疗联盟的RSRT总投资是470万美元。

“Last year RSRT announced Roadmap to a Cure, our three-year $33 million strategic research plan. The enthusiastic support of Rett families in the USA and around the world has made it possible for RSRT to recruit scientists, like Dr. Wilson, who are tops in their field to aggressively pursue curative approaches. As a mother of an adult daughter who suffers daily from the effects of Rett I know that time is of the essence. I am immensely grateful to our families and their network of supporters who make our work possible,” said Monica Coenraads, Executive Director of RSRT.
“去年，RSRT宣布了一项治疗方案的路线图，这是我们3年3千3百万美元的战略研究计划。在美国和世界范围内，雷特患者家庭的热心支持使得RSRT能够招募到像威尔逊博士这样的科学家，这些科学家在他们的领域里积极寻求治疗方法。作为一个成年女儿的母亲，她每天忍受着雷特病患的影响，我知道时间是最重要的。我非常感谢我们的家人和他们的支持者，他们让我们的工作成为可能。”RSRT执行董事莫妮卡.科内拉德说。

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，生物科技公司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，但也会罕见地影响到男孩。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。