PRESS RELEASE: AveXis to Advance Gene Therapy Program Initiated by the Rett Syndrome Research Trust

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新闻稿：AveXis公司促进基因治疗项目由雷特综合症研究信托基金于2017年6月7日发起 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 excited to announce that AveXis, Inc., has committed to advance a gene therapy candidate for Rett Syndrome into human clinical trials. AveXis, a pioneering clinical-stage gene therapy company, has already advanced a transformative gene therapy treatment for spinal muscular atrophy Type 1, a devastating disease that strikes infants and kills in toddlerhood. Remarkably, in a Phase 1 clinical trial, infants treated with the gene therapy achieved developmental milestones, including the ability to sit unassisted, talk and walk in some cases, – achievements never seen in untreated babies with the disease. It is our shared belief that gene therapy, the introduction of healthy MECP2 genes to compensate for the mutated ones, may also produce benefits for individuals with Rett Syndrome.
特朗布尔，CT——雷特综合症研究信托基金（RSRT）兴奋地宣布，AveXis公司已经承诺将把雷特综合症基因疗法作为候选基因治疗推广到人类临床试验中。AveXis是临床试验阶段基因疗法公司的先驱，已经为脊髓性肌肉萎缩症1型脊髓性萎缩症提供了一种变革性的基因治疗方法。这种疾病是一种致命的疾病，它会在婴儿身上发生，并在蹒跚学步时造成死亡。值得注意的是，在第一阶段的临床试验中，接受基因治疗的婴儿达到了发育的里程碑，包括在某些情况下无辅助的坐、说话和走路的能力，这是在未接受治疗的婴儿身上从未见到的成就。我们的共同信念是基因疗法，即引入健康的MECP2基因来补偿突变的基因，也可能对患有雷特综合症的病患个体产生益处。

This milestone represents the culmination of research originating in 2010 when RSRT funded the collaboration between Dr. Gail Mandel and Dr. Brian Kaspar, the now scientific founder and Chief Scientific Officer of AveXis, to explore the potential of gene therapy for the treatment of Rett Syndrome. Encouraged by the results of that collaboration, in 2014 RSRT conceived of, recruited the scientists for and funded the Gene Therapy Consortium to definitively evaluate the feasibility of gene therapy as a treatment for Rett Syndrome.
这一里程碑标志着2010年发起的研究的高潮，当时RSRT资助了盖尔曼德尔博士和AveXis的科学创始人兼首席科学官员布莱恩卡斯帕博士的合作，探索基因疗法治疗雷特综合症的可能性。在合作成果的鼓励下，2014年RSRT构想的，招募了一些科学家并资助了基因治疗联盟，以确定基因疗法作为治疗雷特综合症的可行性。

The Consortium scientists, Stuart Cobb, Steve Gray, Brian Kaspar and Gail Mandel, exceeded our expectations by developing a gene therapy product candidate with impressive efficacy, safety and delivery characteristics. Importantly, therapeutic benefits in the mouse models are much greater than that of any drug ever tested for Rett Syndrome.
该联盟的科学家，斯图尔特科布，史蒂夫格雷，布莱恩卡斯帕和盖尔曼德尔，超出了我们的预期，他们开发了一种基因疗法候选产品，具有令人印象深刻的功效，安全性和传递特性。重要的是，试验小鼠模型的治疗效果比任何一种用于雷特综合症的药物都要大得多。

Adrian Bird's 2007 reversal study showing that symptoms in mice can be dramatically reversed suggests that a diagnosis of Rett need not translate to lifelong disability. While gene therapy seems an obvious approach for any of the numerous single gene disorders, the quality of the data generated by the Consortium was an essential factor in AveXis' decision to prioritize development of a gene therapy for Rett Syndrome.
艾德里安伯德在2007年的逆转研究表明，试验小鼠的症状可以被显著逆转，建议雷特的诊断不一定会转化为终生残疾。虽然基因疗法似乎是众多单一基因疾病中的一个明显的方法，但该联盟产生的数据质量是AveXis决定优先发展雷特综合症基因疗法的关键因素。

"RSRT recognized early on that the gene therapy approach would move forward only if we aggressively championed it. We brought together scientists with strong track records of success and provided them with the financial support and infrastructure to work effectively and efficiently. Our strategy worked and I am thrilled that AveXis, a leader in gene therapy, is now taking this work forward at an industry scale. I am beyond grateful to the many Rett families who believe in our vision and to the generosity of their networks that make our vital work possible," said Monica Coenraads, Executive Director of RSRT and mother to a young woman with Rett Syndrome.
"RSRT很早就认识到基因治疗方法只有在我们积极地支持它的时候才会向前发展。我们汇集了具有成功经验的科学家，并为他们提供了资金支持和基础设施，使他们能够有效、高效地工作。我们的策略奏效了，我很高兴，基因疗法的领导者AveXis现在正以行业的规模推进这项工作。我非常感激许许多多雷特的家庭，那些相信我们的愿景和他们网络资源的慷慨，这让我们的重要工作成为可能，"RSRT的执行总监和一个患有雷特综合症的年轻女儿的母亲莫妮卡科内拉德说。

"As was made clear in RSRT's recently launched strategic research plan, Roadmap to a Cure, our mission is a cure for Rett Syndrome. Today's announcement is a huge step forward towards our goal. By addressing the core cause of Rett, gene therapy has the potential to be a life-changer. We have complete confidence in the expertise and leadership of the AveXis team and will continue to provide our full support as this program moves forward," said Randall Carpenter, Chief Scientific Officer, RSRT.
"正如RSRT最近启动的战略研究计划——治疗路线图所表明的那样，我们的使命是治愈雷特综合症。"今天的声明是朝着我们的目标迈出的一大步。通过解决雷特的核心发病原因，基因疗法有可能改变人们的生活。我们对AveXis团队的专业知识和领导能力充满信心，并将继续提供我们的全力支持，因为该项目将继续推进。"RSRT首席科学官兰德尔卡彭特说。

"Without RSRT championing gene therapy for the treatment of Rett Syndrome, we would not be where we are preclinically in our understanding of the potential for gene therapy to treat this devastating disease," said Brian Kaspar, Chief Scientific Officer of AveXis. "Bringing our expertise and focus on rare monogenic diseases, we are excited by the possibility that gene therapy may address the needs of individuals with Rett Syndrome."
"如果没有RSRT支持基因疗法来治疗雷特综合症，我们就不会在临床前了解基因疗法治疗这种毁灭性疾病的可能性，"AveXis的首席科学官布莱恩.卡斯帕说。"将我们的专业知识和重点放在罕见的单基因性疾病上，我们对基因疗法可能解决雷特综合症患者的需求感到兴奋。"

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. In March of 2017 RSRT announced Roadmap to a Cure, a three-year, $33 million strategic research plan. The plan prioritizes four curative approaches with gene therapy as our lead program. RSRT operates at the nexus of global scientific activity enabling advances in knowledge and driving innovative research through constant engagement with academic scientists, clinicians, industry, investors and affected families. Since 2008, RSRT has awarded $42 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。在2017年3月，RSRT宣布了一项名为“治愈”的路线图，这是一项为期3年、耗资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。RSRT在全球相关科学活动的联系中发挥作用，通过与学术科学家、临床医生、行业界、投资者和受影响家庭的持续接触，促进知识的进步和推动创新研究。自2008年以来，RSRT已经为研究提供了4200万美元。想了解更多信息，请访问www.reverserett.org。

About AveXis, Inc. AveXis is a clinical-stage gene therapy company developing treatments for patients suffering from rare and lifethreatening neurological genetic diseases. The company's initial proprietary gene therapy candidate, AVXS-101, recently completed a Phase 1 clinical trial for the treatment of SMA Type 1. For additional information, please visit www.avexis.com
AveXis公司是一家临床阶段的基因治疗公司，为患有罕见的和危及生命的神经系统疾病的患者开发治疗方案。该公司最初的候选专利基因治疗AVXS-101，最近完成了第一阶段治疗SMA型1的临床试验。欲了解更多信息，请访问www.avexis.com

About Rett Syndrome, Rett Syndrome is a genetic neurological disorder that almost exclusively affects girls. 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, normal movement often including the ability to walk. As the children enter childhood the disorder brings anxiety, seizures, tremors, breathing difficulties, 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，几乎只会影响到女孩。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，常常包括走路的能力。当孩子们进入童年时，这种疾病会带来焦虑、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。