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SMA Research Effort Funded by Loren Eng and Dinakar Singh

Loren Eng and Dinakar Singh, the wife and husband who were the driving force behind the creation of the Spinal Muscular Atrophy (SMA) Center at CUMC in 2002, have made a substantial gift through the SMA Foundation to help recruit Dr. Umrao Monani, a leading SMA researcher who will join CUMC this summer.

Dr. Monani, a molecular geneticist, has participated in groundbreaking SMA research by helping develop one of the most widely used mouse models of SMA, a feat that makes further basic research on the disease possible. Dr. Monani was recruited to Columbia by Dr. Darryl De Vivo, the Sidney Carter Professor of Neurology at CUMC and director of the SMA Clinic.

Dr. Timothy A. Pedley, Henry and Lucy Moses professor of neurology and chairman, says: "Columbia already has an unparalleled group of clinical and laboratory investigators working on motor neuron disorders. This offers an exceptionally fertile environment for Dr. Monani's research, which will extend, and in turn be amplified by, studies ongoing here. The SMA Foundation has given us an extraordinary opportunity to find effective treatments and ultimately a cure for SMA."

"Our belief in the depth and quality of the work being done at Columbia under Dr. De Vivo's leadership is tremendous," says Ms. Eng, founder and president of the New York-based Spinal Muscular Atrophy Foundation and underrecognized, underdiagnosed and underfunded. We are very excited about the research possibilities this new funding opens up." SMA is the leading genetic cause of infant and toddler death.

Arya Singh

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Every parent watches his or her infant genetic, neuromuscular disease that has been compared to Lou Gehrig's disease.

SMA is characterized by a wasting away of nerve cells in the spinal cord. The degeneration leads to increasing muscular weakness and affects a child's

Progress is being made, however. Researchers have isolated the gene—survival of motor neurons, or SMN1—necessary for motor neurons to function normally. Its absence may trigger the disease's onset. In addition to SMN1, people have a backup gene, SMN 2, that produces a smaller amount of protein. Nearly everyone with spinal muscular atrophy has a mutated SMN1 gene. It is the number of backup genes that determines the severity of the disease. Several studies aimed at manipulating the SMN2 gene are under way in the United States.

To hasten progress in bringing meaningful help to children with SMA, Ms. Eng, Mr. Singh and the SMA Foundation envision continued support in the areas of clinical care, translational research (the process by which laboratory findings are translated into drugs and treatments) and basic science research.

"Columbia has the potential to become a world-class SMA center because of the talent that already is here and is being brought here," says Ms. Eng, who believes a cure for SMA is tantalizingly within reach. The NIH has identified SMA as the neurological disease that has the greatest potential for a treatment or cure in the near term and has commenced work on a therapeutics development project for SMA.

Still, grabbing that golden ring will take dedication and commitment by the best scientific minds. "We at the SMA Foundation rely on Dr. De Vivo and the superb clinical and scientific team he has

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mother of 4-year-old Arya, a patient of Dr. De Vivo's who has one of the milder forms of SMA. Of her and her husband's recent $860,000 commitment Ms. Eng says, "This new gift makes clear that we consider the SMA team at Columbia to be at the forefront of the research taking place on this disease today."

According to Dr. De Vivo, "SMA is a relatively common disease that has been and toddler for signs that the child is reaching appropriate milestones such as rolling over, sitting up, and starting to walk. When a young child is delayed in reaching these markers, or perhaps reaches them and then slows or reverses in development, parents grow concerned and look for explanations. For about 1 out of every 6,000 children that explanation will come as a diagnosis of SMA, a ability to crawl, walk, control the head and neck, and breathe. The disease may be present in varying degrees of severity. In milder cases, a child might be able to sit but not walk (SMA type 2) or be able to walk and stand alone (SMA type 3); in severe cases (SMA type 1) breathing is inhibited and death is almost certain before the age of 2 years. No treatment, aside from palliative care, is available.

assembled, the collaborations he is making with other researchers across the university and even outside the university," Ms. Eng says. "Our commitment to CUMC is ongoing and heartfelt."

To learn more about SMA research at Columbia University, contact Director of Development Lori Funicello at 212-304-7200.