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New Grant Seeks to Fill Knowledge Gaps Regarding Spina Bifida

It's well known that folic acid reduces risk, but not how or why; NIH funding will support genomic investigations and international patient registry

Researchers at University of California San Diego School of Medicine, in collaboration with Rady Children's Institute for Genomic Medicine, have been awarded a five-year, $8.3 million grant from the National Institutes of Health's Eunice Kennedy Shriver National Institute of Child Health and Human Development to further illuminate the causes of spina bifida, the most common structural defect of the central nervous system.

Spina bifida occurs when the developing spine and spinal cord do not form properly. It's a defect of the neural tube, the structure in developing embryos that eventually becomes the baby's brain, spinal cord and surrounding tissues.

In spina bifida, a portion of the neural tube doesn't close and the backbone protecting the spinal cord doesn't form correctly, often resulting in damage to the cord and nerves that leads to physical and neurological disabilities, including paralysis from the waist down and hydrocephalus or excessive fluid buildup inside the brain.

The condition is relatively rare, occurring in approximately one in 3,000 births worldwide. Treatment depends upon the severity of the condition, often involving surgeries before and after birth.

The exact cause of spina bifida is not known. It likely involves multiple factors: genetic, nutritional and environmental. Previous research has established that folic acid, known as vitamin B9, can halve the risk of spina bifida when taken by women prior to conception of a child. As a result, folic acid is a common nutritional supplement in grain-based foods in the United States and other countries.

"And yet spina bifida persists," said Joseph Gleeson, MD, Rady Professor of Neuroscience at UC San Diego School of Medicine and director of neuroscience research at the Rady Children's Institute for Genomic Medicine. "We know B9 reduces risk, but the how remains a mystery and the disease remains incurable."

The new grant will be used by Gleeson and colleagues to set up an international registry of patients with spina bifida and fund new studies investigating how B9 reduces disease risk. In cooperation with the Spina Bifida Association, Shriner's Children's Hospitals and spina bifida clinics throughout the world, UC San Diego and Rady physicians and scientists will apply whole genome sequencing to identify potential causes and underlying mechanisms of the disease.

Whole genome sequencing is a process of determining the complete DNA sequence of a patient's genome or genetic material at a single time. It can be used to identify subtle differences in a similar group or guide development of personalized therapies based upon genetic information.

Members of the grant team include Vineet Bafna, PhD, professor in the Department of Computer Science at UC San Diego; Christopher Kintner, PhD, and Joseph Ecker, PhD, both molecular biologists at Salk Institute for Biological Studies; and Lee Niswander, PhD, a developmental biologist at University of Colorado Boulder.

For more information on the spina bifida grant and program, email contact@gleesonlab.org.

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