Uncovering a hidden genetic landscape in autism, OCD

Scientists are sequencing the three billion DNA letters in the human genome, searching for changes that cause diseases like autism and cancer, and they’ve discovered many important mutations. Still, one type of mutation - called “indels” - has remained largely hidden from view. Now, a team of CSHL scientists has developed a method to reveal these indels, making it possible to discover new mutations associated with disease.

Indels are the addition or deletion of one or more letters in the genome. These often tiny changes cause a shift in the genetic code, rendering proteins dysfunctional and often causing disease. Led by CSHL Assistant Professor Mike Schatz, along with Professor Mike Witting and Assistant Professor Johannes Rehms, the team created a computer formula to identify indels. They used the method to look at families with autism and discovered dozens of new mutations associated with the disease. Read more about this powerful new method here.

New genetic basis for sex determination

Any high school biology student will tell you that the genetic difference between men and women comes down to the X and Y sex chromosomes. But a team of researchers at CSHL has found another genetic basis for sex determination, in fruit flies. The team, including lead author Deplaine Fagegaltier, Ph.D., discovered that a class of very small genes called microRNAs are involved in the fly develops and matures. Even during adulthood, microRNAs send signals that ensure fertility and allow eggs and sperm to develop. In fact, the researchers found that one microRNA preserves sexual identity in male and female flies. Learn more here.

NSF grants to Albeau, Mitra support innovative brain research

The National Science Foundation has awarded Early Concept Grants for Exploratory Research (EAGER) to CSHL Assistant Professor Florin Albeau and Professor Partha Mitra. As part of President Barack Obama’s multi-year BRAIN Initiative, EAGER awards aim to answer fundamental questions about how the brain works. Albeau and Mitra are working to develop new technologies that will provide insight into how neural circuits are configured and how they process and store complex information to control behavior, learning, and memory. Read more here.

LIZ2DAY Walk raises $525,000 for women’s cancers

For the last 10 years, the LIZ2DAY walk has raised funds for breast cancer research and support for patients. This year, the walk expanded its mission to include all women’s cancers, raising $525,000 from more than 600 participants. At a recent luncheon, CSHL was awarded $22,300 which will support breast cancer research in Associate Professor Mikala Epskamp’s laboratory. Learn more here. 

September is Pediatric Cancer Awareness Month

This year, nearly 18,000 kids will be diagnosed with cancer. During the month of September, Cold Spring Harbor Laboratory will join groups across the country to raise awareness about the need for research. Check out the #应收账 campaign, urging Congress to increase research funding or join in a local event like Christina Renna Foundation Walk for a Cure, which benefits pediatric cancer research at CSHL.