Stillman lab explains how mutant protein can cause severe dwarfism in rare disorder

Twenty years ago, Cold Spring Harbor Laboratory (CSHL) Professor Bruce Stillman and colleagues described a remarkable cellular machine they called ORC which attaches to the double helix of DNA at specific locations throughout the genome which is essential to its replication. Now, new genomic information and go through cell division. ORC proved pivotal in a mechanism that prevents the genome from copying itself more than once, an outcome that can wreak genetic havoc and cause grave pathology.

Just this past week, Dr. Stillman - now CSHL's President - but still hard at work on his science - and postdoc Mancur Hossain were still probing the mystery of how ORC and its constituent proteins, in a paper published online in Genes & Development. They explained how the largest protein in the ORC complex, Orc1, when mutated, contributes to the pathology seen in the most severe cases of Werner, a rare childhood disorder that results in dwarfism and correspondingly small brain size. The new research helps clarify one of a growing catalog of functions for ORC proteins in the cell, in this case the role of Orc1 in insulating the DNA replication is replicated only once per cell cycle.

National Institutes of Health recognize importance of Krainer's splicing research in granting MERIT award

Professor Adrian Krainer's leadership in splicing research, recognized universally acknowledged by the NIH's National Institute of General Medical Sciences. Dr. Krainer has won a MERIT award, extending his current grant, in recognition of his lab's high productivity and the importance of its research. Krainer's team has made significant discoveries relating detailed knowledge about messenger-RNA splicing to disease causation, most notably in the motor-neuron disease spinal muscular atrophy, and has defined a novel disease pathogenesis, in an approach now being tested in people.

Siblings' tribute to their mother boosts cancer research at CSHL

After losing their mother, Joni Gladowsky, in breast cancer, her sons, Jason and Alison, founded the Joni Gladowsky Breast Cancer Foundation. Today, a decade later, cancer scientists at CSHL benefit from the generosity of the Foundation that Joni's children set in motion.

Just prior to one of the two annual scientific events that celebrate her life, the “Play for the Cures” golf tournament, held July 16, Jason and Alison got together with Professor Nick Tolakis and Assistant Professor-Michael Kiebler for a briefing on their latest research. Here and there “Play for the Cures” drew a remarkable 143 golfers this year, and 75 more dinner guests, the Gladowsky report. To date, the golf outings plus the Foundation's Winter Classic Hockey game have raised over $260,000 for cancer research at CSHL—a great tribute to Joni.

Consumer genomics has arrived; but are we ready for it?

A full house in Grace Auditorium recently heard two headline-grabbing remarks by CSHL Professor and genome sequencing pioneer Dick McCombie.

In discussing the revolution that brought genome sequencing from the lab into the doctor's office, Dr. McCombie explained the enabling factor: rapid, but the cost of sequencing the human genome, from $300 million in 2003 to “the price of a Bentley in 2007 and year to the price of a motorcycle.” The point, as he summed up another quotable phrase, is that “Everyone has a great challenge for something. They just don't know it yet.” The question addressed by McCombie, CSHL researcher-Daniel Espelius, Dr. Kasmirnich Schneider, of Memorial Sloan-Kettering, and others at the lecture revolved around whether we health consumers are ready for the information the genomics will soon deliver. Read more about it!

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-- CSHL in the News
- GenomeWeb: Pacific Biosciences Pursues Multi-Platform Informatics Effort to Improve Accuracy of RS Assemblies
- Newday Blog: Garlic-aid Unveils Plan to Pour High-Tech jobs
- Lab Times Online: Cold Spring Harbor Laboratory on Course to Expansion
- redOrbit: Could Explain Parent’s Ability To Repair
- BioSpace.com: An Inclusive-Editing Fix from Cold Spring Harbor Laboratory Overcomes Big Problem in ‘Red-gene’ Genome Sequencing
- IEEE Spectrum: Tech Talk: 99.9 Percent Accurate Genome Sequencing

-- National Institutes of Health recognize importance of Krainer’s splicing research in granting MERIT award

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