Biology

Gene tied to childhood brain disorder

A child between the ages of 5 and 10 years suddenly goes blind. Seizures occur with increasing frequency, and the child inexorably loses his or her mental faculties. Death generally strikes by age 20.

This tragic sequence typifies Batten disease. Afflicting an estimated 1 in 25,000 newborns, it's the most common neurodegenerative disorder striking children. The only way physicians have been able to distinguish Batten disease from other disorders is by noticing the accumulation of certain pigments and proteins within neurons and many other cell types—an accretion that may cause the illness or be simply a by-product of the true problem.

"A lot of times the diagnosis is difficult or even missed," says Terry J. Lerner of Massachusetts General Hospital in Boston.

The International Batten Disease Consortium, to which Lerner belongs, has finally tracked down the mutated gene responsible for the devastating brain disorder. In 1989, investigators placed the blame somewhere on chromosome 16. Now, Lerner and her colleagues have found a gene on that chromosome that is mutated in all the Batten patients they've studied so far. Most of them have an identical chunk of the gene deleted, the consortium reports in the Sept. 22 CELL.

Identification of the gene should help physicians diagnose the disease quickly. A treatment for the condition may emerge if investigators can determine how the protein encoded by the gene works in healthy individuals.

"We would like to find out what it is doing in the normal cell," says Lerner, who notes that investigators will examine whether the gene's protein accumulates abnormally in Batten patients.

Telling left from right in chick embryos

The left and right halves of an embryo in its earliest stages of development may look perfectly balanced, but on a molecular level its genes are already choosing sides.

Researchers have found that certain proteins are made on either the left or the right side of the embryo, marking the first few steps in a molecular pathway that determines left-right asymmetry in the adult animal.

Cliff Tabin of Harvard Medical School in Boston and his colleagues report in the Sept. 8 CELL their discovery that proteins from three genes—activin receptor IIa (*cAct-RIIa*), sonic hedgehog (*Shh*), and chicken nodal-related 1 (*cNR-1*)—form a sort of two-pronged biochemical cascade. On the left side, the Shh protein spurs cNR-1 protein synthesis, while on the right side, a molecule called activin suppresses synthesis of Shh and stimulates production of cAct-RIIa.

The researchers knew that these genes influence a wide range of developmental events, which amplifies the importance of their discovery of different protein pathways on the two sides, Tabin says. The genes' functions "made it seem plausible that these things were involved in controlling asymmetry," he says.

This idea held up when the group manipulated protein synthesis to see how it affected development of the chick embryo's heart. When they stimulated Shh production on both sides, not just the left, they saw that the newly formed heart tubes—which usually loop to the right—opted to curl to the left half of the time. The 50-50 pattern was consistent with the heart tube's receiving signals from both the left and right sides, essentially leaving the direction of its growth to chance. Moreover, suppressing Shh production on both sides gave the same results.

Further work in this area could include finding additional steps in the pathway, determining the signals that actually make the heart tube bend, and measuring how far the signals travel from their source, Tabin says.

Physics

Lasing turned upside down

There's a new kind of excitement in the laser world.

Getting a beam of coherent light out of a conventional laser generally requires a significant input of energy. Much of this energy goes into maintaining a majority of the atoms or molecules of the lasing material in an excited state. The result is a population inversion in which there are more particles in the higher-energy state than in the ground state.

Now, a team of physicists from the United States, Russia, and Germany have for the first time experimentally demonstrated laser oscillation without population inversion. The researchers take advantage of a quantum interference effect to generate a sustained laser beam of infrared light in a vapor of rubidium atoms.

Marlan O. Scully of Texas A&M University in College Station and his collaborators report their achievement in the Aug. 21 PHYSICAL REVIEW LETTERS. More recently, a second team has observed lasing without inversion in a sodium atomic beam.

In experiments performed at the National Institute of Standards and Technology in Boulder, Colo., Scully's team used external laser beams to produce what are, in effect, two different paths from the ground state to a particular excited state of a rubidium atom. Under the right conditions, these two quantum paths interfere with each other to block absorption but not emission of photons of the proper energy. This allows lasing to occur without having large numbers of atoms in the excited state.

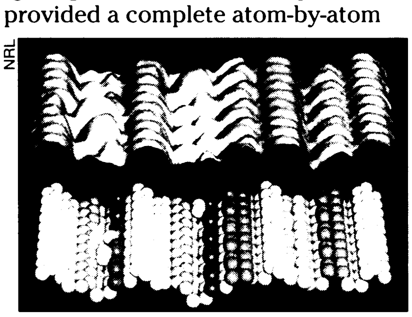
Such a technique offers the possibility of circumventing some of the difficulties researchers have faced in the past in producing laser light at ultraviolet and X-ray wavelengths.

Unveiling silicon's new face

Slicing a silicon crystal is a violent process that tears apart bound atoms, leaving a forest of dangling atomic bonds on the fresh surfaces. To reach some degree of stability, these surface atoms quickly rearrange themselves into new configurations to incorporate these bonds. The details of what happens depend on the direction of the slice through the crystal lattice.

Because silicon is the basis of modern microelectronics, researchers have studied a number of these surfaces in great detail, particularly those parallel to the main axes of silicon's crystal lattice. But much less is known about the structures of surfaces that result when a silicon crystal is cut to expose a surface tilted at some large angle to these standard planes.

Now, researchers have provided a complete atom-by-atom description of a recently discovered, stable surface of silicon having just such an extreme tilt. Deeply and evenly grooved, this surface could serve as a natural template for fabricating various types of one-dimensional structures and thin films for novel applications in microelectronics.


NRL

Scanning tunneling microscope image (top) and computed reconstruction (bottom) showing atomic structure of a newly characterized silicon surface.

Lloyd J. Whitman, Alison A. Baski, and Steven C. Erwin of the Naval Research Laboratory in Washington, D.C., describe the surface, labeled Si(5 5 12), in the Sept. 15 SCIENCE.

"This face of silicon has one of the largest periodic structures ever observed and is stabilized by a complex rearrangement involving every atom on the surface," the researchers note.