Study: Stuttering may be genetic

For many years, stuttering has been identified primarily as an emotionally based disorder — an audible sign of extreme anxiety or nervousness. Evidence compiled recently, however, indicates that stuttering is not only a physical abnormality but one that may be passed from one generation to the next.

A study of 555 stutterers and more than 2,000 of their close relatives "suggests that susceptibility to stuttering is genetically transmitted," reports Kenneth L. Kidd, professor of human genetics at Yale University School of Medicine. Results of the study also show that males appear to be more susceptible to the disorder than are females. Kidd presented his findings recently at the annual meeting of the American Association for the Advancement of Science.

The research confirms that stuttering "runs in families" — the frequency of stuttering among relatives of subjects is "much greater than the frequency in the population at large," Kidd says. Although this does not in itself constitute conclusive proof that stuttering is transmitted genetically, Kidd says he has ruled out two major psychological/cultural possibilities as modes of transmission.

First, the explanation that the child stutters as a way of *imitating* a parent could account for a maximum of only 10 percent of the cases, according to Kidd. "Ninety percent [of stutterers] had two fluent parents" at the time of birth, Kidd told SCIENCE NEWS. In many such cases, either the father or mother had at one time stuttered and recovered prior to the baby's birth and/or other relatives had been stutterers.

Second, the suggestion that familial anxiety can be internalized by the child to cause stuttering also may be ruled out, he says. Since anxiety is known to exacerbate the severity (measured, in this case, by frequency) of stuttering, any hypothesis pinpointing anxiety as a cause would mean that more severe stutterers would have a higher proportion of stuttering relatives, Kidd says. "But we found no such correlation," he says.

Further indication that the affliction is genetically transmitted, he says, is confirmed by the apparent "sex-specific" nature of stuttering. Statistics indicate that up to 5 percent of males, compared with just 2 percent of females, stutter for at least six months sometime during childhood. But beyond that, Kidd has found that although female stutterers are less common, they "have significantly more relatives who have ever stuttered." This suggests, he says, that "whatever contributes to susceptibility to stuttering, more of those factors would need to be present for a female to surpass the stuttering threshold than for a male to cross the threshold...if more factors of promoting stuttering are required to make a female stutter, families of female stutterers would have more of those factors, and hence, more stutterers" — which, apparently, they do.

"Definite proof" of a genetically transmitted disability, Kidd concedes, "is elusive. Yet all available evidence suggests that susceptibility to stuttering is genetically transmitted." He hypothesizes that the transmission may occur through either a single gene or "through many different genes [each] with a very tiny effect." In the latter case, the accumulation of a certain number of such genes above a critical point causes the individual to surpass the threshold and develop a stutter, according to the researcher.

Physiologically, stuttering involves a combination of factors that cause the person to "breathe abnormally or move his vocal cords inappropriately," says Martin Adams of the University of Houston. About 80 percent of the children who ever stutter recover before adulthood, Kidd estimates. Little research has been undertaken to determine what, if any, differences exist between those children and the other 20 percent who continue to stutter as adults. But, says Kidd, "once it persists, it is very hard to overcome."

Just how the Yale findings could help stuttering victims is unclear. "It probably won't mean anything immediately," Kidd says. "But if we can find out what the problem is [genetically and physiologically], it could have major implications for therapy and... possibly prevention." □

Lab-grown virus for a diarrhea vaccine

A major cause of serious diarrhea in infants and young children is a virus known as rotavirus. The virus is thought to be responsible for a large share of the millions of deaths annually from diarrhea, among children in developing countries. The first step toward an urgently needed rotavirus vaccine was reported in the Jan. 11 SCIENCE. A team headed by Richard G. Wyatt of the National Institute of Allergy and Infectious Diseases succeeded in growing a strain of human rotavirus under laboratory conditions. The keys to the success were patience and piglets. Virus taken directly from human stool specimens does not grow in cultured cells. So the scientists instead orally infected a newborn, germfree piglet with "type 2" rotavirus from feces of a young patient. They then transferred the virus from piglet to piglet for 11 passages, until the accumulated mutations allowed the virus to propagate in laboratory-grown monkey kidney cells. With the new procedure, the investigators plan to analyze the rotavirus genetic structure and hope to create a weakened virus suitable for use as a vaccine. □

A protein linked to photosynthesis

Joseph Priestly first twisted the key that unlocks the secrets of photosynthesis when he demonstrated in 1772 that a mouse in a closed jar could survive with a plant but not alone. Although Priestly did not realize what was happening, his experiments were the first to show that plants produce oxygen. Ironically, what was first observed about photosynthesis remains one of the last mechanisms of the energy-converting process to be fully understood by scientists. Now, however, two researchers may be hot on the trail of the protein responsible for oxygen evolution in photosynthesis.

Douglas Winget of the University of Cincinnati and Mark Spector of Cornell University have isolated from the thylakoids or inner chloroplast membranes of spinach a protein necessary for photosynthetic oxygen evolution. The protein has an apparent molecular weight of 65,000 and contains two manganese atoms per molecule. Whether this particular protein is the enzyme that catalyzes the photosynthetic splitting of water to generate oxygen remains to be seen. What has been observed, though, is that removal of the specific protein from thylakoid membranes stops oxygen evolution; addition of the same protein restores oxygen evolution.

The trail to the elusive protein involved in oxygen evolution has been blazed for some time. "So far, for many people who have tried to find such a protein, there have been some tantalizing suggestions of findings, but never before has anyone been able to pull out the protein and purify it," Winget says.

The success story of the two protein pinpointers, which will appear in the February PROCEEDINGS OF THE NATIONAL ACADEMY OF SCIENCES, is just one chapter in the continuing saga of enzyme isolation. The guidepost for enzyme purification is some measurement of what the enzyme should be doing — if the enzyme catalyzes the splitting of water, for example, oxygen evolution is measured. For soluble enzymes, or enzymes that float inside cells and organelles, the cell is simply broken, the enzyme extracted and the enzyme-specific activity measured. As the enzyme becomes increasingly purified, more specific activity can be measured.

But a membrane protein, such as the protein involved in photosynthetic oxygen evolution, poses more of a problem for enzyme isolators. Extracting the protein from its membrane often renders it inactive. "In order to see the activity of the membrane protein, one has to put it back in the membrane where it feels more at home," Spector says, explaining the rationale behind a procedure pioneered by other researchers — Yasuo Kagawa and

Efraim Racker of Cornell University, for example. Winget and Spector cite use of this membrane reconstitution procedure as one reason for their successful isolation of a membrane protein involved in photosynthetic oxygen evolution.

Each portion of the photosynthetic process decoded ultimately will have numerous applications — control of plant growth and the development of herbicides, for example. Meanwhile, Winget prefers to view the isolation of a protein required for photosynthetic oxygen evolution as "a gain in knowledge on a basic level."

"I can't say, 'Now that we have this enzyme, we will be able to solve a food problem, an agriculture problem, a cancer problem or anything like that,'" Winget says; rather, the protein isolation is more like a long-awaited twist of Priestly's more than 200-year-old key. □

Kepone mimics female hormone

In 1975, male workers in a Virginia plant making the pesticide kepone were found to suffer symptoms that included tremors, irritability, memory loss and sterility (SN: 11/20/76, p. 324). Since then, scientists have learned more about how kepone actually triggers sterility.

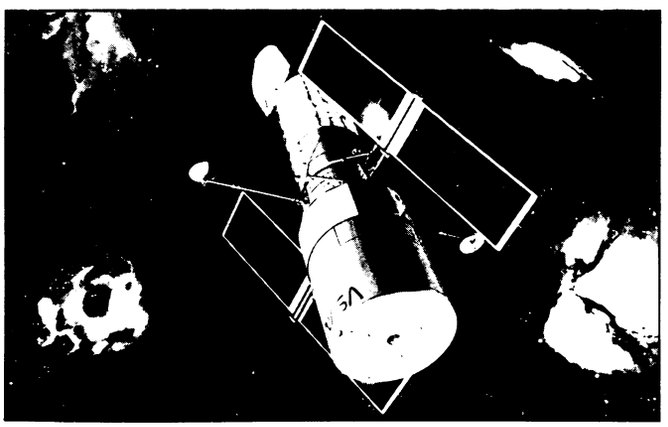
For one thing, kepone creates a constant estrus in female mice. This finding suggests that it exerts sterility by mimicking the female reproductive hormone estrogen. But how does kepone, as an estrogenic mimic, actually bring about constant estrus? The answer now appears to have been found by Bruce Hammond and his co-workers at the University of Illinois at Urbana. They report in the December PROCEEDINGS OF THE NATIONAL ACADEMY OF SCIENCES that kepone competes with estrogen for receptors in the uterus.

The possibility that kepone might exert its estrogenic activities by interacting with estrogen receptors in the uterus of the female rat was especially intriguing because kepone has a chemical structure dramatically different from that of estrogen hormones. Tests were conducted both in live animals and in the test-tube.

Hammond and his team found not only that kepone interacts with estrogen receptors in the rat uterus, but that it competes with the estrogen hormone estradiol for these receptors. Although kepone's ability to bind to estrogen receptors is extremely small compared with that of estradiol, it might still be able to tie up the receptors if it is present in the body for any length of time, the researchers believe. In fact, kepone has already been found to have a long life in the body because of its unusual ability to accumulate in liver and fat tissues, and when female rats eat kepone for seven weeks, all enter constant estrus. □

Institute to run Space Telescope

Late in 1983 or early in 1984, the U.S. space shuttle is scheduled to loft a huge, 10-ton satellite whose activities will center around a single instrument: the Space Telescope. Although the 2.4-meter instrument will be less than half the size of some telescopes now in use on earth, its 500-kilometer-high orbit, well above the obscuring atmosphere, is expected to let it see about 350 times the volume of space that can be studied from the ground, including stars only one-fiftieth as bright as those visible to earthbound observatories. Designed to last through the end of the century, it is to be serviced about every two and a half years by shuttle astronauts, and occasionally brought back to earth for overhaul and relaunching. The device is eagerly awaited both by stellar astronomers, who will be able to see unprecedentedly faint and distant stars, and by planetary observers, who will be able to study the solar system's worlds with detail formerly limited to the brief visits of space probes.


NASA

The 2.4-meter Space Telescope, as it will appear in orbit after being carried aloft by the space shuttle.

Because of the heavy demand anticipated (and already in evidence) for the instrument's services, it will not be operated like one of the National Aeronautics and Space Administration's typical scientific satellites or probes. In fact, neither the scientific programs nor the day-to-day operations of the facility will be handled by NASA at all. Instead, the agency has asked potentially interested parties — universities, aerospace companies, consortia, etc. — to bid for a contract to design, set up and run a Space Telescope Science Institute. Housed at a site of the contractor's choosing, the institute would manage the various science programs, select the observers, plan and schedule operations, and even (from NASA's Goddard Space Flight Center in Maryland) operate the telescope itself. The bids, which will actually be detailed proposals for the institute's design and operation, are due at NASA by March 3, where the winner is expected to be chosen by August or September.

One reason for the institute system, says program manager Don Burrowbridge, is that the heavily instrumented device's full and complicated observing schedule (it may run virtually 24 hours a day) could prove too much of a coordination burden (and an expense) for NASA to do the job "in-house." Although each of the telescope's five initial instruments will have its own principal investigators, for example, only the first two months of observing time will be exclusively theirs. Responding to the widespread interest in using the device, NASA plans to allow greater and greater numbers of outside observers to take part. After the first two months, the "development-phase science team" and the European Space Agency (which is providing one of the instruments as well as the telescope's solar panels) will be allowed only about 60 percent of the schedule time, with the rest going to outside astronomers. Half a year later, the balance will shift further, to 35/65, and twelve months after that to 25/75. By the time the telescope has been aloft for 30 months, the original science team will be out of the picture entirely, leaving the institute to coordinate the needs of astronomers from around the world, who may be using the instrument in numbers as high as 10 persons a day.

The institute approach, Burrowbridge adds, should also enable the Space Telescope program to be more "responsive" to the scientific community at large, an advantage echoed by some astronomers who feel that it gives a chance for the science to be managed in a less parochial way than if it were administered from within NASA.

Remarkably, the Space Telescope also appears to be on schedule and within its budget, according to a report to Congress this month from the U.S. Comptroller General. The report also concludes, however, that over its nearly 20-year lifetime, inflation and other factors may cause the overall program to cost as much as twice NASA's $1.1 billion estimate. The report urges that more complete cost data be given to Congress. □