Breast cancer: Hope for a genetic test

The elation sparked by last fall's discovery of a gene linked to breast cancer gave way to pessimism when scientists reported finding 22 distinct mutations in the gene. This made routine screening for defects in the gene seem far beyond reach (SN: 12/3/94, p.372).

Now, scientists say they're up to 38 mutations in the gene, called BRCA1. But they also sound a note of hope. Most mutations appear to lead to a shortened version of the protein for which BRCA1 holds the code. If that trend holds for many patients, it could lead to a simple diagnostic test for the defect, says geneticist Mark H. Skolnick of Myriad Genetics and the University of Utah School of Medicine, both in Salt Lake City.

"We're hoping we can do something other than just detect specific mutations. It'll be much less expensive, more reliable, more automatable," says Skolnick. He and 41 colleagues report their findings in the Feb. 15 JOURNAL OF THE AMERICAN MEDICAL ASSOCIATION.

Breast cancer kills some 46,000 American women each year. The cause of most breast cancers remains unexplained, but 5 to 10 percent stem from a defect in BRCA1 or another gene. Women with a mutant BRCA1 have about an 85 percent chance of developing breast cancer.

Skolnick's team pooled data on BRCA1 mutations from nine laboratories in North America and the United Kingdom. The data, some newly reported, included complete scans of BRCA1 in 372 women with breast or ovarian cancer who had a familial risk of these cancers. Three labs also looked for two mutations in the DNA of 714 other patients with these cancers, most of them with no known family history of risk.

Analysis showed 38 variations among 80 women with a mutation in BRCA1. Significantly, of the 63 patients whose defective gene was scanned completely, 86 percent appeared to carry errors coding for a shortened protein. Its role in the body remains unclear.

"It means that many people with many, many different mutations should have the same effect from that gene," Skolnick says. This might enable scientists to identify these people by screening for the snippet of BRCA1 that yields a short protein instead of poring over the entire gene, he says.

But his group still has a lot of work ahead. First, it must find out whether the truncated protein code is common to most mutations, including those not yet discovered. It also needs to learn how great a cancer risk the shortened protein poses. Even then, the test could hit technical snags, warns Francis S. Collins of the National Center for Human Genome Research in Bethesda, Md.

"It's definitely a big maybe," says one of the study's authors, Lawrence C. Brody, also of the center. *— J. Kaiser*

Infant CP protection

Giving magnesium sulfate to pregnant women at risk of delivering very low birthweight babies may help lower their babies' chances of developing cerebral palsy (CP), a new retrospective study suggests. Very low birthweight infants make up about 25 percent of the 5,000 babies diagnosed with CP in the United States each year.

Two researchers compared the prenatal care of 42 children who weighed less than 3.3 pounds at birth and who developed CP to the care of 75 children who had similar birthweights but no CP, they report in the February PEDIATRICS.

Seven percent of the women who bore CP children had received magnesium sulfate during pregnancy, while 36 percent of the mothers of the children without this central nervous system disorder had received the drug, report Karin B. Nelson of the National Institute of Neurological Disorders and Stroke in Bethesda, Md., and Judith K. Grether of the California Birth Defects Monitoring Program in Emeryville.

Physicians had prescribed magnesium sulfate either to prevent convulsions associated with the women's preeclampsia, a pregnancy-induced form of high blood pressure, or to stop premature contractions. The authors couldn't explain why the drug might reduce the incidence of CP, but other studies suggest it may prevent cerebral bleeding, they note. *— T. Adler*

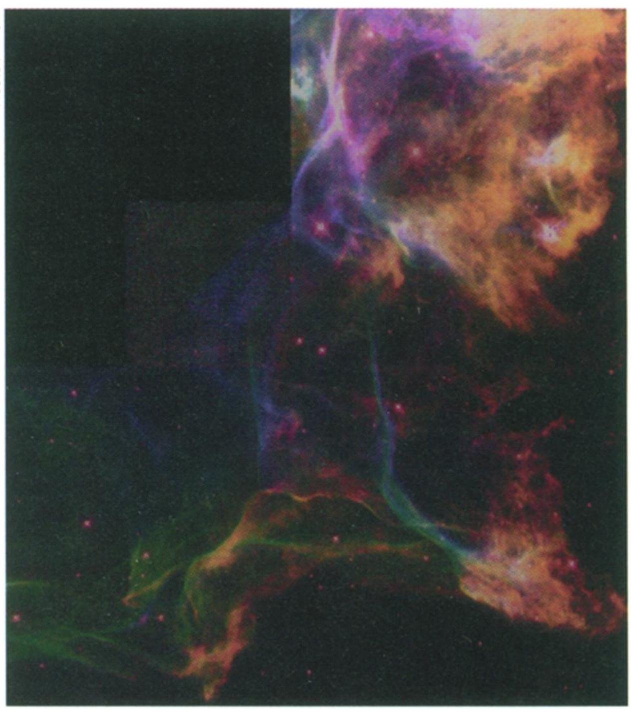
A supernova remnant's shocking trail

Plowing through its surroundings, the racing shock wave from a supernova explosion sets interstellar gas aglow. Gas recently struck by the wave generally radiates at a higher temperature, while gas farther behind the wave glows cooler. Images taken by the unrepaired Hubble Space Telescope had revealed such a pattern for the Cygnus Loop, a supernova remnant 2,500 light-years from Earth (SN: 3/13/93, p.168).

Now, the sharp-eyed, repaired Hubble has captured this remnant in finer detail. The new picture shows a narrow zone directly behind the shock wave, in which hydrogen atoms — rather than ions — radiate most of the light. John J. Hester of Arizona State University in Tempe discussed the image this week in Atlanta at the annual meeting of the American Association for the Advancement of Science.

Hester notes that before the shock hurtled past, the gas consisted mainly of neutral atoms, electrons, and protons. When the wave blasted through, it energized the electrons and protons, which began to collide with the atoms. Even after a few collisions, some atoms remained un-ionized, holding onto their outermost electrons. But the collisions did cause the atoms to radiate. This accounts for the zone of atomic emissions, which spans a width a few times the Earth-sun distance, Hester says.

As the charged particles continued to bombard the atoms, they succeeded in ionizing them, generating the high-temperature emissions directly behind the thin region of atomic hydrogen radiation. As the ions radiated, they began to cool, creating the low-temperature region behind the hotter layer. *— R. Cowen*



Hester/NASA

In this false-color image, shock wave in Cygnus Loop moves from left to right. Green indicates light emitted by hydrogen atoms just behind the wave; blue denotes doubly ionized oxygen; red depicts singly ionized sulfur.