Mutant gene offers cholesterol resistance

Some people can eat whatever they want and never seem to suffer ill effects. A prime example of that maxim was the 1991 report of an 88-year-old man who ate about 25 soft-boiled eggs each day.

Despite his voracious appetite for cholesterol-rich eggs, this man did not appear to suffer from atherosclerosis, the deposition of cholesterol and fatty debris on artery walls (SN: 4/13/91, p.236). Researcher Fred Kern Jr. at the University of Colorado School of Medicine in Denver attributes this man's avoidance of heart trouble to unusually efficient methods of cholesterol metabolism.

Indeed, the man has a mutant gene that allows him to eat high-cholesterol foods with no corresponding rise in low-density lipoprotein (LDL), the carrier molecule that deposits fat-like cholesterol on artery walls, which can cause a heart attack, says Richard B. Weinberg. Weinberg was studying the normal form of the gene and suspected a genetic explanation for the man's apparent cholesterol immunity. He found the mutant form in a frozen sample of the man's blood, which he had obtained from Kern.

Now, Weinberg is reporting findings from a pilot study that confirms previous evidence that the gene plays a role in the way the body manages dietary cholesterol. Weinberg, who is at the Bowman Gray School of Medicine at Wake Forest University in Winston-Salem, N.C., and his colleagues presented their data last week at the American Heart Association's 66th scientific sessions in Atlanta.

The mutant gene produces a protein that differs by a single amino acid from the protein coded for by the normal gene, he says.

Weinberg and his colleagues began their study by recruiting 23 medical students. With a simple blood test, the team looked for the mutation, determining that 11 students produced the flawed version of the protein. The remainder made the normal protein.

For the first two weeks of the study, the investigators told the students to eat a diet that limited cholesterol intake to 200 milligrams (mg) per day. The researchers then told the recruits to eat four eggs per day, a diet that provided the students with 1,000 mg of cholesterol daily. In both phases of the study, students consumed the same amount of fat.

The team discovered that in students who had the normal gene, the concentration of LDL cholesterol in the bloodstream increased from 92.2 mg per deciliter (mg/dl) at the end of the first two weeks to 111.4 mg/dl at the end of the egg-rich dietary phase.

For the 11 students with the mutant gene, that high-cholesterol diet seemed to do little to their LDL amounts. Their LDL rose from 93.8 mg/dl to 95.3 mg/dl during the same period, Weinberg reports.

The students with the mutant gene appeared to escape the deleterious effects of eating lots of dietary cholesterol, at least in this pilot study, Weinberg says.

"If the results are confirmed, it would be a very important finding," comments Kern.

There may be an evolutionary advantage to inheriting the mutant gene, which is relatively common in some parts of the world. The altered gene may help protect people from clogged arteries, Weinberg speculates. Previous studies have shown the mutation occurs in about 15 percent of people living in societies (such as the United States) that favor high-fat, high-cholesterol diets. By contrast, the mutation appears uncommon in Asia and Africa, where low-fat, low-cholesterol diets are the rule.

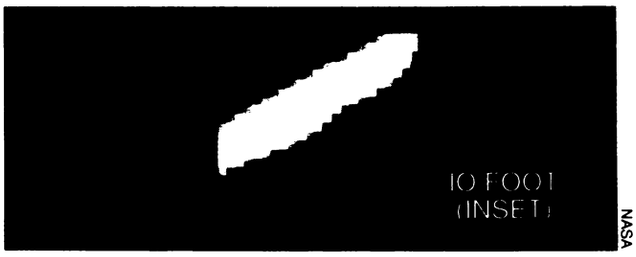
If this gene does play a role in a person's response to dietary cholesterol, it may lead to a more individualized approach to preventing heart disease, Weinberg says. It may be that people with the mutant gene can tolerate a high-cholesterol diet and should focus on other risk factors for heart disease, such as smoking, he suggests.

Much about this gene remains mysterious. Researchers have yet to find the mechanism by which the flawed gene shields the body from dietary cholesterol. It may be that the protein produced by the mutant gene inhibits absorption of cholesterol passing through the intestine, Weinberg says. Indeed, Kern, who is retired now, points out that his former patient absorbed just a fraction of the cholesterol in those daily servings of 25 eggs.

And nobody really knows whether the mutant gene alters the body's response to saturated fat in the diet, which can also lead to high concentrations of cholesterol in the bloodstream (SN: 6/13/92, p.390). If it does, the positive effect on arterial health may be even more significant.

– K.A. Fackelmann

Jupiter and Io: Infrared spots mark link



NASA

Inset shows infrared emission near Jupiter's south pole caused by an electrical interaction between the planet and Io.

Astronomers have discovered a pair of ghostly spotlights that sweep across Jupiter in synch with the motion of its moon Io. Researchers say the faint infrared emissions, detected at NASA's Infrared Telescope Facility atop Hawaii's Mauna Kea, stem from the interplay between Io and the magnetic field of Jupiter.

Researchers have long suspected that Io and Jupiter influence each other through an electrical interaction in which magnetic field lines from the planet strike the moon, notes John E.P. Connerney of NASA's Goddard Space Flight Center in Greenbelt, Md. But scientists hadn't located the exact places in Jupiter's polar regions from which the magnetic field lines enter and exit the planet.

In marking the two magnetic footprints, the infrared spots "provide a magnetic road map of the planet," Connerney says.

He and Richard Baron, Takehiko Satoh, and Tobias C. Owen of the University of Hawaii in Honolulu, report their study in the Nov. 12 SCIENCE.

The finding, says Connerney, upholds a theory proposed in 1969 that Jupiter's rotating magnetic field acts as an electrical generator on Io, inducing a voltage across the moon. The voltage creates 5 million amperes of current that flow back to Jupiter through two separate circuits – magnetic field lines that originate from its north and south polar regions. The infrared emissions might stem from the electrical heating generated by the current, he says.

Alternatively, Connerney adds, as the flow of electrons in the current crashes into Jupiter's ionosphere, it may excite ions there to glow in the infrared. In either case, the interaction appears the likely cause of variations in the radio signals emitted by the planet.

Both infrared spots remain about 8 degrees closer to the equator than the luminous polar patches associated with Jupiter's aurora, indicating that Io plays no major role in generating this electromagnetic disturbance, Connerney says.

Alexander J. Dessler of the University of Arizona in Tucson says the study indicates that magnetic maps of Jupiter generated from Voyager data are more accurate than those from Pioneer 11. Tracking the motion of the spots, he adds, will provide a sensitive test of whether Jupiter reverses its magnetic field every decade or so, like the sun, or roughly every half million years, like Earth.

– R. Cowen