Genes shed light on common blindness cause

Newly identified genes responsible for two rare, degenerative diseases of the retina could help researchers target the cause of age-related macular degeneration, an eye disorder that constitutes the leading cause of blindness among older individuals in developed countries.

Two independent groups of researchers have pinpointed the genes that, when altered, lead to Best's disease and North Carolina macular dystrophy – two inherited forms of macular degeneration that affect individuals early in life. A team led by ophthalmologist Edwin M. Stone and pediatrician Val C. Sheffield of the University of Iowa in Iowa City report in the July NATURE GENETICS that the Best's disease gene lies on chromosome 11. In the July GENOMICS, researchers led by ophthalmologist Kent W. Small of Duke University in Durham, N.C., report tracing the North Carolina macular dystrophy gene to chromosome 6.

Macular degeneration is a group of largely untreatable disorders characterized by destruction of the macula, the central portion of the retina responsible for fine visual detail. Patients with age-related macular degeneration often develop progressively widening distortions or blank spots in their vision, beginning in their 50s or 60s. According to the National Eye Institute, each year 16,000 people in the United States with age-related macular degeneration become legally blind, which means that eyeglasses cannot correct their vision.

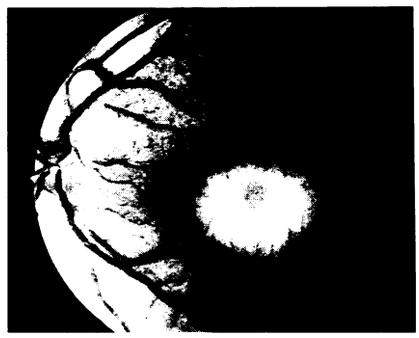
In Best's disease, the macula becomes coated with a yellowish substance called lipofuscin, which obstructs progressively larger regions of the retina as an individual ages. Researchers estimate that this inherited disorder causes only 1 percent of all cases of macular degeneration. However, they cannot accurately assess the prevalence of Best's disease because many cases are misdiagnosed as age-related macular degeneration.

The Iowa researchers found the gene responsible for Best's disease by analyzing DNA taken from 45 members of a five-generation family with 29 Best's disease patients. By comparing the DNA from affected and unaffected family members, they narrowed down the genetic defect responsible for the disorder to a specific region on the long arm of chromosome 11.

The Duke researchers used a similar strategy to pin down the gene for North Carolina macular dystrophy, which affects 96 members of various ages from a single large family whose ancestors first settled in that state in 1830. Small and his colleagues pinpointed the cause of this early-onset form of macular degeneration to the long arm of chromosome 6.

The discoveries are the first steps toward developing tests and treatments for Best's disease and North Carolina macular dystrophy, says Iowa's Stone. But even more important, he adds, the findings will lead to a better understanding of all types of macular degeneration, of which he estimates "well over 50 percent" have a genetic cause.

"We want to find five or six different genes that affect the central retina and then see how they relate to age-related macular degeneration," Stone says.



A photograph of a Best's disease patient's retina reveals degeneration (white blotch) of the central retina, or macula.

Stone/NATURE GENETICS

Duke's Small agrees that the new genes "could have far-reaching ramifications for age-related macular degeneration and other, similar eye disorders." – *C. Ezzell*

Mt. Pinatubo's cloud shades global climate

It's a cooler world, thanks to Mt. Pinatubo, the Philippines volcano that blasted into prominence last summer in one of the largest eruptions of the century. A variety of recent measurements confirm predictions that the volcanic debris would act, at least temporarily, to cool global climate.

In a series of eruptions starting on June 15 of last year, Mt. Pinatubo ejected an estimated 20 million tons of sulfur dioxide gas into the stratosphere, where it formed tiny droplets of sulfuric acid (SN: 8/31/91, p.132). Such droplets, or aerosols, remain suspended in the upper atmosphere for several years following an eruption. Mt. Pinatubo's aerosol cloud, which circled the globe within a few weeks of its emergence, led scientists to speculate that incoming sunlight might be scattered or blocked, resulting in cooler temperatures on the ground.

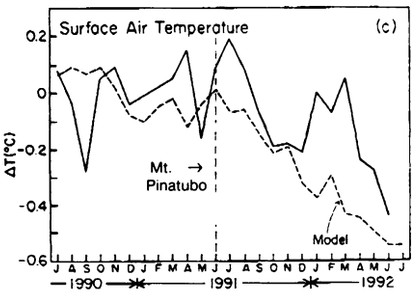
Volcanic aerosols have now been caught in the act, the National Oceanic and Atmospheric Administration (NOAA) announced earlier this month.

Ellsworth G. Dutton, a meteorologist with NOAA's Climate Monitoring and Diagnostics Laboratory in Boulder, Colo., traced the effects of Pinatubo's cloud with ground-based instruments that directly measure the strength of sunlight. Dutton says his results show a 20 to 30 percent decline in the amount of solar radiation that reaches the ground without being scattered or reflected, and a 2 to 4 percent decline in total solar radiation.

Temperatures have already started to drop, both at ground level and in the lower atmosphere, says James K. Angell of NOAA in Silver Spring, Md. Angell told SCIENCE NEWS his analyses of weather balloon data show that the first half of 1992 was 0.4°C cooler, overall, than the first half of 1991. He notes that the volcano's effect may be greater than suggested by these observed temperature shifts, since this year's El Niño warming would normally *raise* average temperatures by 0.2°C (SN: 1/18/92, p.37).

Weather satellites confirm cooling in the lower atmosphere, recording a global drop of more than 0.5°C since last June, with this June being 0.2°C cooler than average, according to John Christy of the University of Alabama at Huntsville and Roy Spencer of NASA's Earth Science Lab at the Marshall Space Flight Center in Huntsville. Christy says their data indicate that the greatest cooling, 1.0°C, occurred in the northern midlatitudes – an area that includes the continental United States – while temperatures in the southern hemisphere have dropped by only 0.3°C.

James E. Hansen, a climate modeler at NASA's Goddard Institute for Space



James Hansen and Helene Wilson

Average monthly global temperatures compared with model predictions. Zero equals mean temperatures for July 1990 through June 1991.

Studies in New York City, eagerly follows such weather reports. His computer models of the volcano's effects have predicted that the upper atmosphere would warm as aerosol particles scattered and absorbed solar energy, while the partially shaded lower atmosphere would cool. Although some initial temperatures did not clearly follow Hansen's predictions, the recent reports fit well with his model.

According to Hansen's model, average surface temperatures will continue to drop, for a maximum cooling of 0.6°C by the end of this year. Then, temperatures will gradually return to normal by 1994 as the volcanic aerosols slowly settle back down to Earth. – *K. Hoppe*