Gene connected to human cognitive trait

In two independent studies, scientists have for the first time linked a specific gene with a facet of human thought.

Genes typically come in pairs, with each parent providing one copy. However, the deletion of one copy of a particular gene on chromosome 7 disrupts a person's ability to visualize and mentally manipulate parts of objects, the investigators find. Without this capacity, assembling a model or a simple piece of furniture proves nearly impossible.

The functions of the chromosome 7 gene, called LIM-kinase 1, remain unclear. though researchers theorize that the protein it produces may help to coordinate the development of brain cells involved in some visual and spatial skills.

One of the new studies, directed by geneticist Mark T. Keating of the University of Utah Health Sciences Center in Salt Lake City, appears in the July 12 CELL. The other, directed by geneticist Mayada Tassabehji of St. Mary's Hospital in Manchester, England, is in the July NATURE GENETICS.

"These are genuinely exciting findings because they take us to the point where we can begin to examine the relation of specific genes to cognitive capacities," asserts Helen B. Tager-Flusberg, a psychologist at the University of Massachusetts in Boston.

Both research teams noted a missing copy of LIM-kinase1 in people diagnosed with some or all of the symptoms of Williams syndrome. This rare condition includes mild or moderate mental retardation, impaired visuospatial construction skills, heart and blood vessel defects, elevated concentrations of calcium in the blood during childhood, and an elfin face.

A specific personality also characterizes people with Williams syndrome. They exhibit a hearty friendliness toward acquaintances and strangers alike, constantly seek out human contact, converse easily with others, and react with extreme sensitivity to other people's feelings. The condition largely spares auditory and verbal memory, often resulting in an impressive vocabulary and keen recall of names and faces.

In 1993, Keating and his colleagues reported that the absence of one copy of a chromosome 7 gene called elastin caused the heart problems and unusual facial features of Williams syndrome. They speculated that genes adjacent to elastin account for the disorder's visuospatial difficulties.

In the CELL report, Keating's team analyzes the structure of a DNA segment that encompasses the elastin gene in 25 people from two families, about half of them exhibiting no signs of Williams syndrome and the rest possessing the typical heart defects but not mental retardation. Tests directed by Carolyn B. Mervis of Emory University in Atlanta found that most of the people with heart problems also displayed severe difficulties with visuospatial constructions.

Chromosome 7 deletions were smaller in this partial version of Williams syndrome than in the full-blown condition but larger than the deletions in individuals who possess the heart condition alone, the scientists contend.

Molecular analysis of the DNA region deleted in partial Williams syndrome revealed that it normally includes the elastin and LIM-kinase1 genes, Keating

Tassabehji and her colleagues likewise found that each of 20 people with Williams syndrome lacked single copies of both elastin and LIM-kinase 1.

These findings are "a welcome development," but researchers must now establish whether LIM-kinase1 orchestrates visuospatial constructions entirely on its own, asserts Barbara Pober, a geneticist at Yale University School of Medicine.

Complex cognitive capacities such as visuospatial construction probably involve multiple genes, Tager-Flusberg remarks.

- B. Bower

Finding a new definition of photosynthesis

Plants may take more than one approach to photosynthesis, the age-old process that produces the world's oxygen, a new study suggests. A newly discovered alternative may resemble the system that photosynthetic organisms used billions of years ago when Earth's atmosphere lacked oxygen.

Photosynthetic plants use the energy of light to make carbohydrates, their energy storage molecules, from carbon dioxide and hydrogen. Botanists have long held that every plant requires two separate systems, working together in the leaf membrane, to carry out photosynthesis. Each system is made up of proteins with embedded pigments. Photosystem I (PSI) is essential to the plant's assimilation of carbon dioxide, and photosystem II (PSII) splits water molecules to produce oxygen and hydrogen, according to the so-called Z scheme, proposed in 1960 and extended by later research.

In the past 10 years, however, researchers have created two mutant forms of a common green alga, Chlamydomonas, that have only PSII.

A year ago, Elias Greenbaum and James W. Lee of Oak Ridge (Tenn.) National Laboratory and their colleagues demonstrated that the Chlamydomonas mutants absorb atmospheric carbon dioxide and split water into hydrogen and oxygen

when exposed to light. They did not show, however, that the algae could grow using this version of photosynthesis.

Now, the scientists have found that the mutants' photosynthesis does enable them to grow, they report in the July 19 SCIENCE. "We aren't saying that the Z scheme is wrong. We're saying we've discovered an alternative or new pathway that accomplishes the same process, explains Greenbaum. He speculates that some photosynthetic organisms may use both the new and old pathways.

This finding "is an exciting result that challenges the research community to discover how these [mutant] cells grow," says John Whitmarsh of the University of Illinois at Urbana-Champaign.

"The results are extremely surprising," says Laurens J. Mets of the University of Chicago, who helped develop the mutant algae and coauthored the earlier report in the Aug. 3, 1995 NATURE. He had thought the mutant algae could not obtain enough energy from photosynthesis to grow. He adds, however, that the team has yet to demonstrate that the mutants lack all signs of PSI.

Moreover, just because these laboratory-made mutants can grow using photosynthesis doesn't mean that normal plants can do so without PSI, Whitmarsh and Mets agree.

Greenbaum's group grew the mutant



A scanning electron microscope image of a mutant Chlamydomonas, 10 micrometers in diameter.

algae with and without oxygen and found that the plants perform photosynthesis less effectively in the presence of oxygen and a strong light. This and other evidence suggest that when photosynthesis first evolved, plants had only PSII, he says. PSI evolved to help plants cope with the oxygen, he suggests.

The team did the experiment as part of an effort to find more efficient ways to produce hydrogen for use as a renewable energy source. Normal algae make excess hydrogen when grown without carbon dioxide, the group's earlier studies demonstrated. - T. Adler

JULY 20, 1996 SCIENCE NEWS, VOL. 150 39