

GENETICS

Genetic Disorder Found

Homocystinuria, a genetic disorder caused by a mutant gene and a defective enzyme, has been added to the list of inborn errors of metabolism—By Faye Marley

► A NEWLY RECOGNIZED genetic disorder called homocystinuria has been added to the list of "inborn errors of metabolism." One symptom is dislocated lenses that can cause nearsightedness and detached retina.

A nationwide screening of patients in eye clinics throughout the United States is expected to reveal many more than the 46 cases reported in Bar Harbor, Maine, by Dr. Victor A. McKusick of the Johns Hopkins University School of Medicine, Baltimore, at a short course in medical genetics.

The disease is caused by a mutant gene and a defective enzyme. It can easily be detected through a urine test that shows the presence of homocystine and amino acid.

Symptoms include some of those found in the Marfan's syndrome, such as great height, spider fingers or weakness of the aorta. President Lincoln is believed to have had a true Marfan's syndrome because of his loose jointedness and height. A total of 143 families were screened in the Hopkins study with the cooperation of other universities that helped to locate patients with dislocated lenses or presumed Marfan's syndromes or both.

One feature found in homocystinuria but

not in the true Marfan's syndrome is generalized osteoporosis, usually leading to codfish vertebrae and some degree of vertebral collapse.

Dr. McKusick said that a majority of the patients showed some degree of mental retardation because the defective enzyme is involved in feeding the brain. Sixteen of the 46 Hopkins cases have normal intelligence, however, and one of them has two college degrees in a highly technical field and is an employee of a leading chemical firm.

A low-protein dietary treatment has been attempted on one mentally retarded six-year-old boy. The diet is especially low in methionine. Dr. McKusick said the diet should be started as soon as possible after the birth of a child found to have homocystinuria. A family with one affected child should be on the lookout for another.

The Hopkins study will be reported in the Journal of the American Medical Association on Aug. 30. Collaborating in the report are Dr. R. Neil Schimke, now of Kansas City, Mo., and Drs. Thomas Huang and Abou D. Pollack, both of Baltimore city hospitals.

• Science News Letter, 88:131 August 28, 1965

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Genes, Psychosis Linked

► EVERYBODY is potentially a schizophrenic, Dr. John L. Fuller, geneticist and associate director of the Jackson Laboratory in Bar Harbor, Maine, believes.

"We are getting away from the idea that a single bad gene produces a disease," he said. Dr. Fuller explained that the vulnerability to schizophrenia, the most common mental disease, is genetically conditioned.

In a lecture to doctors and professors attending a short course in medical genetics, Dr. Fuller referred to the recent book, "Genetics and the Social Behavior of the Dog," which he wrote in collaboration with Dr. Paul J. Scott.

Both scientists did their work at the Behavioral and Genetics Laboratories at Hamilton Station, part of Jackson Laboratory.

Dogs forced to listen to the continual sound of a doorbell go into convulsions. This is an example of vulnerability.

Different breeds of dogs, like different types of people, behave differently. In isolation experiments the wire-haired terriers were affected less seriously than beagles, which are less aggressive.

"Our temperament is not all determined by how our parents treat us," Dr. Fuller told SCIENCE SERVICE in an interview. "There are children as well as dogs who are born with

a high drive and stand up for themselves. On the other hand, a student can go to pieces under pressure."

Some geneticists believe that genetic factors in the mentally retarded have not been emphasized enough in the past few years, Dr. Fuller pointed out. Environment, instead, is being stressed.

However, there are children in culturally deprived homes who are bright mentally. This is a sign of genetic factors at work, Dr. Fuller said.

The short course in medical genetics was conducted by the Jackson Laboratory and Johns Hopkins University with financial sponsorship of the National Foundation-March of Dimes.

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PUBLIC HEALTH

Black Cat Gains Fame As Hearing Loss Patient

► HANGOVER is a black cat with a hearing loss in the inner ear, where the process of converting sound energy into nerve impulses begins.

Because of this hearing loss Dr. F. Blair Simmons, assistant professor of surgery at



Stanford University

HANGOVER—A black cat named Hangover, under the care of Dr. F. Blair Simmons, assistant professor of surgery at Stanford Medical School, has a hearing loss in the inner ear. He is being studied by Dr. Simmons with the hope that the findings will be applicable to human beings with ear disorders.

the Stanford Medical School in Palo Alto, Calif., has been observing the cat since 1961.

Dr. Simmons, who specializes in disorders of the ear, nose and throat, hopes his research will show what kind of auditory information will allow Hangover to hear. The information may possibly be applied to humans.

Since Hangover took up residence at the Stanford Medical Center in 1961, he has been fed the finest cat foods. Every precaution is taken to be sure that he does not become afflicted with ailments common to cats.

Dr. Simmons is studying Hangover because he has not been able to get enough persons with impaired hearing to volunteer for research on deafness. The ultimate prevention of most deafness depends, to a large extent, on having the bones of the inner ear available for investigation.

The research problem is that the inner ear cannot be seen or directly examined during life because of its inaccessible location within the temporal bones, the hardest bones within the head, which surround the inner ear bones and their nerve structures.

Stanford Medical Center is participating in a program sponsored by the Deafness Research Foundation, which encourages persons to bequeath their earbones to bone banks after death. The bones are used in research toward aiding other deaf persons with potentially repairable hearing defects.

They are of the greatest value when submitted in conjunction with the results of hearing tests and other available pertinent medical records. When enough of them become available, the hearing tests on Hangover will no longer be required.

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