Scans of the genome of patients with schizophrenia have revealed rare spontaneous copy number mutations that account for at least 10 percent of the non-familial cases of the disease. Researchers describe specific genetic mutations present in individuals who have schizophrenia, but not present in their biological parents who do not have the disease. These individuals were eight times more likely to have these mutations than unaffected individuals. This new data, reported in the May 30 on-line issue of Nature Genetics, will help researchers account for the persistence of schizophrenia in the population despite low birth rates among people with the disease. Researchers at Columbia University Medical Center scanned the genome of 1,077 people which included 152 individuals with schizophrenia, 159 individuals without schizophrenia, and both of their biological parents for copy number mutations. They found mutations, either a gain or loss of genes, in 15 individuals diagnosed with schizophrenia that were not present in the chromosomes of either biological unaffected parent. Only two of such mutations were found in those without schizophrenia. Study subjects were from the European-origin Afrikaner population in South Africa, a genetically homogenous population that is ideal for genetic evaluation.
“We now know the cause of around 10 percent of the cases of sporadic schizophrenia,” said Maria Karayiorgou, M.D., professor of psychiatry, Columbia University Medical Center, the senior author on the study. “Schizophrenia is not as much of a ‘big black box’ as it used to be. The identification of these genes lets us know what brain development pathways are involved in disease onset, so that in the future we can look at better ways of treating this devastating disease.”

Schizophrenia affects approximately 1 percent of the population worldwide. About 40 percent of the disease is thought to be inherited, with the other 60 percent sporadically showing up in people whose family history does not include the disease.

One of the new or de novo mutations researchers found in more than one affected individual in this study was a deletion of a region of chromosome 22. Dr. Karayiorgou had previously provided evidence that loss of genes in this region, 22q11.2, was responsible for introducing “new” or sporadic cases of schizophrenia in the population. This confirms 22q11.2 as the only known recurrent such mutation linked to schizophrenia.

“We have already demonstrated 22q11.2 to be involved in sporadic schizophrenia and we have made considerable progress in understanding the underlying biological mechanisms,” said Dr. Gogos. “Now, we have a new set of mutations that we can investigate. The more information we have about the biological basis for this disease, the more information we can provide to those who suffer from it and their families.”

“Such abnormal deletions or duplications of genetic material are increasingly being implicated in schizophrenia and autism,” explains National Institute of Mental Health Director Thomas R. Insel, M.D. “Now we have a dramatic demonstration that genetic vulnerabilities for these illnesses may stem from both hereditary and non-hereditary processes. This line of research holds promise for improved treatments – and perhaps someday even prevention – of developmental brain disorders.”

Karayiorgou and co-senior author Joseph A. Gogos, M.D., Ph.D., associate professor of physiology and neuroscience at Columbia University Medical Center, agree that the goal is for psychiatrists to be able to inform patients that they have a mutation that is causing their disease and ultimately to be able to tailor treatments to individual patients based on their specific mutation. This tailored treatment is a ways off, according to Dr. Karayiorgou, but she says patients and their families are relieved to know that there is a biological cause of their illness.

The researchers plan to extend their screen for additional de novo mutations by using increased resolution scans to study additional families. They also plan to scrutinize further genes affected by the identified mutations through human genetics and animal model approaches.

This entry was posted on Friday, May 30th, 2008 at 8:22 pm and is filed under Genetics News, Mental Diseases News, Neuroscience Research News. You can follow any responses to this entry through the RSS 2.0 feed. You can leave a response, or trackback from your own site.

- Spontaneous Mutations Rife in Nonfamilial Schizophrenia
- Common Mechanisms in Autism
- Genes Associated with Increased Gout Risk
- New Genes Linked to Lung Cancer
- Isocitrate Dehydrogenase gene IDH1 Mutations Isolated in Brain Tumors
Related Gene Mutations Responsible for Ten Percent of...
Schizophrenia Articles

- **Spontaneous Mutations Rife in Nonfamilial Schizophrenia** People with schizophrenia from families with no history of the illness were found to harbor eight times more spontaneous mutations
- **New Genes Linked to Lung Cancer** Working as part of a multi-institutional collaboration, scientists at Washington University School of Medicine in St. Louis have assembled the
- **HSN2 Mutations Lose Pain and Heat Perception** Hereditary sensory and autonomic neuropathy type II, abbreviated to HSANII, is a poorly understood genetic disorder wherein affected patients lose
- **Common Mechanisms in Autism** Many of the seemingly disparate mutations recently discovered in autism may share common underlying mechanisms, say researchers supported in part
- **Gene Mutation Linked to Walking** What are the genes implicated in upright walking of humans? The discovery of four families in which some members only
- **Research in Genetically Predisposed Obesity** Individuals who have a genetic mutation associated with high body mass index (BMI) may be able to offset their increased
- **Isocitrate Dehydrogenase gene IDH1 Mutations Isolated in Brain Tumors** HHMI investigators have detected a multitude of broken, missing, and overactive genes in pancreatic and brain tumors, in the most

**Images**

**Recent Comments**

- Valerie Marshall on [Therapy for Hypophosphatasia a Congenital Form of Rickets](http://www.molecularstation.com/science-news/2008/05/therapy-for-hypophosphatasia-a-congenital-form-of-rickets/)
- lekmut dhiemmex p. on [Living Cells as Nanotechnology Factories](http://www.molecularstation.com/science-news/2008/05/living-cells-as-nanotechnology-factories/)
- Doug - Colon Cancer Association on [Enzyme Involved in Cell Replication Could be Cancer Treatment Key](http://www.molecularstation.com/science-news/2008/05/enzyme-involved-in-cell-replication-could-be-cancer-treatment-key/)
- doctorb on [HPV Vaccine Decreases Negative Pap Results](http://www.molecularstation.com/science-news/2008/05/hpv-vaccine-decreases-negative-pap-results/)
- Louis_MD on [HPV Vaccine Decreases Negative Pap Results](http://www.molecularstation.com/science-news/2008/05/hpv-vaccine-decreases-negative-pap-results/)

**Tags**

alzheimer's, antibiotics, bacteria, brain, cancer, cell, cells, chemotherapy, climate, diabetes, disease, dna, drug, drugs, environment, evolution, extinction, gene, genes, genetic, genome, health, heart, heart failure, HIV, immune, immune cells, infection, inflammation, lung cancer, mouse, mutations, neurons, nutrition, obesity, physical activity, prostate, protein, proteins, stem cells, stress, tumor, vaccine, virus, water

**Search...**

**Archives**

- January 2009
- November 2008
- October 2008

http://www.molecularstation.com/science-news/2008/05/gene-mutations-responsible-for-ten-percent-of-schizophrenia/
Categories

- Alzheimer Disease Research (7)
- Anthropology Research (3)
- Antibiotic Research News (2)
- Archeology News (4)
- Astronomy Research (1)
- Bacteriology Research News (28)
- Behavior Research (9)
- Bioinformatics News (11)
- Biology News (6)
- Biotechnology Research (1)
- Cancer Research News (55)
- Cardivascular Research (31)
- Cell Biology News (46)
- Chemistry News (9)
- Child Care Research (5)
- Computer Technology News (1)
- Dermatology Research (5)
- Diabetes Research News (8)
- DNA Research News (9)
- Drug Discovery Research News (14)
- Embryology News (5)
- Embryology Research News (5)
- Endocrinology Research (2)
- Environmental Research (53)
- Enzyme Research (1)
- Epigenetics Research News (3)
- Evolutionary Research (12)
- Fertility Research (3)
- Gastrointestinal Research (2)
- Genetics News (50)
- Health News (30)
- HIV Research (3)
- Immunology Research (25)
- Infectious Diseases Research (6)
- Inflammatory Bowel Disease Research (3)
- Mammalian Research (5)
- Marine Research (2)
- Medical Research News (7)
Gene Mutations Responsible for Ten Percent of Schizophrenia

- Mental Diseases News (3)
- Microbiology Research (6)
- Molecular Biology News (12)
- Molecular Imaging News (3)
- Nanoelectronics Research (1)
- Nanotechnology News (5)
- Neuroscience Research News (60)
- Nutrition Research (17)
- Obesity News (13)
- Obstetrics Research (1)
- Ophthalmology News (5)
- Osteo Research (1)
- Palentology Research (2)
- Physical Activity News (8)
- Plant Biology News (3)
- Plastic Surgery Research (5)
- Pregnancy News (5)
- Primary Care News (2)
- Prostate Cancer Research News (3)
- Protein Research News (23)
- Protein Structure News (7)
- Psychiatric Research (6)
- Rehabilitation Research (1)
- Renal Research (1)
- Respiratory Research (7)
- RNA Research News (2)
- RNAi Research News (2)
- Social Research (4)
- Software Research (1)
- Stem Cell Research News (17)
- Stress Research (1)
- Student Learning Research (7)
- Tissue Engineering Research News (1)
- Vascular Research (1)
- Virology News (20)
- Zoology Research (2)

- Alzheimer Disease Research (7)
- Anthropology Research (3)
- Antibiotic Research News (2)
- Archeology News (4)
- Astronomy Research (1)
- Bacteriology Research News (28)
- Behavior Research (9)
- Bioinformatics News (11)
- Biology News (6)
Recent Entries

- Findings in Early TB Infection
- New Genetic Evidence for First Americans
- Reduced Colorectal Cancer Risk With Hormone Therapy
- Childhood Trauma Connection and Risk for Chronic Fatigue Syndrome
- Dual Role Gene Plays Part in Breast Cancers with Poor Prognosis
- Insight Into Aggressive Childhood Cancer
- New Genetic Markers for Ulcerative Colitis
- Novel Glioblastoma Mouse Model
- New Way To Fuse Cells
- Aquaculture Growth Continuing

Copyright © Science News