

**For immediate release**

## **Gene Linked to Early Onset Stroke Discovered by Local Researchers**

Middlefield, Ohio, March 29, 2011 – Researchers at DDC Clinic ~ Center for Special Needs Children in Middlefield, Ohio have discovered a gene connected with early onset of stroke.

The gene identified by DDC Clinic was found through a new condition never before described in medical literature. People with this disorder suffered from cerebral vasculopathy – a disease of the blood vessels in the brain – and early onset of stroke. The findings will appear in this week's edition of the prestigious journal *Proceedings of the National Academy of Sciences (PNAS)*.

The first patient with this condition was referred to DDC Clinic four years ago. The family had three affected children who had been evaluated for years by several major medical centers without a diagnosis. Early signs of the disease include relatively smaller size at birth, low muscle tone and irritability during infancy, along with poor growth and short stature during their childhood. These children have poor blood circulation in their hands and feet and frequently develop frostbite during the winter. They also have low-pitch hoarse voices, joint problems and abnormal laboratory findings.

Dr. Heng Wang, a physician scientist at DDC Clinic, believed that this might be a new condition. While working to assist these children, DDC Clinic received referrals from physicians of Cincinnati Children's Hospital for six more children with similar clinical features. Some of these children displayed more severe symptoms with a history of early onset of stroke.

Genome-wide genetic mapping studies performed by the DDC Clinic research team suggested that all children were affected by the same disorder and that a mutation, or variant, in the SAMHD1 gene was responsible for this novel condition. SAMHD1 was originally identified in human dendritic cells and its roles in immune function and inflammation are further highlighted by this study as each person also showed signs of an immune system disorder.

"The disease consistently attacks vessels of certain organs while sparing others. The specificity in targeting cerebral vessels in this condition is astonishing", said Dr. Wang, Medical Director of DDC Clinic and senior author of the study. The researchers have named the condition as SAMS association, which stands for the acronym of **S**tenosis, **A**neurysm, **M**oyamoya and **S**troke, the most striking characteristics of the cerebral vascular disorders associated with this condition. The first three letters of the gene have been included in the name to indicate its genetic connection.

"We are very excited about our findings and are ready to move to the next step in further understanding the exact molecular mechanism of the disease and its treatment," said Dr. Baozhong Xin, a research scientist of DDC Clinic and lead author on the study. The research team notes that although the disease was found in Amish communities and might be rare in the general population, the study is an important contribution to the body of knowledge about the SAMHD1 gene, and will provide some important clues in understanding common health issues such as strokes.

A total of eighteen patients from Ohio, Kentucky and Tennessee have been confirmed. Dr. Wang visited each family to discuss the research findings soon after the gene mutation was identified. Last year, a previously healthy young man with this condition suffered of a catastrophic stroke but recovered because the family had been made aware of the disease and knew to seek immediate medical attention.

“We hope that our current study and future translational research will provide some effective treatment for this condition.” Dr. Wang said. He believes that the work will open the door to a completely new field of study for many similar cerebral vascular diseases as well.

This is the third new genetic condition DDC Clinic has described since the clinic began providing medical services in 2002. Last year, DDC Clinic published a new condition called TMCO1 defect syndrome and the associated gene in *PNAS*. The clinic also played an essential role in identifying a condition called ganglioside GM3 synthase deficiency, which was published in the journal *Nature Genetics* in 2004.

President and founding board member, JoAnn Brace, commented about these accomplishments “it is amazing how much has been achieved in such a short period of time by this clinic. We are proud of the world-class research and quality of care achieved by our staff. This could not have been accomplished without the help of our visionary supporters.”

The study was supported in part by The Elisabeth Severance Prentiss Foundation, The Reinberger Foundation, the Leonard Krieger Fund of the Cleveland Foundation and many individual supporters. Researchers from Cleveland Clinic, Clinic for Special Children of Pennsylvania, Cleveland State University, Cincinnati Children’s Hospital and Akron Children’s Hospital also contributed to the study.

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**About DDC Clinic** The mission of DDC Clinic is to enhance the quality of life for people with special needs caused by rare genetic disorders. Since it began providing medical services in 2002, DDC Clinic’s achievements have been widely recognized. The clinic received the 2006 American Academy of Pediatrics Special Achievement Award and the 2007 Johnson & Johnson Community Health Care Crystal Award. DDC Clinic was also the recipient of the 2007 Northern Ohio Live Award of Achievement in Health and Medicine and the Excellence Award in Community Service and Medicine from The United States Pan Asian Chamber of Commerce.

For more information on DDC Clinic call 440-632-1668 or visit the website at [www.ddcclinic.org](http://www.ddcclinic.org).

Contact

Patti Gallagher DDC Clinic ~ Center for Special Needs Children  
14567 Madison Road, Middlefield Ohio 44062  
440.632.1668  
[patti@ddcclinic.org](mailto:patti@ddcclinic.org)