International Stroke Genetics Consortium Update

Miriam R. Raffeld, BA; Stephanie Debette, MD, PhD; Daniel Woo, MD, MS

Created in April 2007, the International Stroke Genetics Consortium (ISGC) has grown into a scientific community’s global organizing force for stroke genetics, open to all who can contribute to its mission. Stroke is a leading cause of death worldwide,1 with 15 million people suffering a stroke per year.1 Intracerebral hemorrhage, which makes up 15% of all stroke in the United States,2 results in a 50% rate of severe disability or death.2,3 Although the major cerebrovascular risk factors of hypertension, cardiac disease, diabetes mellitus, and the influence of hyperlipidemia have been well established, a substantial portion of stroke is not explained by these risk factors alone and biological susceptibility to known risk factors remains to be discovered. The ISGC has been at the forefront of genetic discovery to understand the mechanisms behind stroke. Formed with the principal value that collaboration and coordinated effort will produce the highest quality results in the most efficient manner (Table), the ISGC has developed other major stroke collaborations, including the METASTROKE consortium and the SiGN Consortium, and partnered with the CHARGE consortium. The ISGC has identified 8 loci associated with ischemic stroke, 6 loci associated with intracerebral hemorrhage, 5 loci associated with intracranial aneurysm, and 1 locus associated with cervical artery dissection in collaboration with the CADISP consortium. Each of these findings was genome-wide significant with replication in an independent sample.

Collaboration is the cornerstone of the ISGC because we continue to increase the number of samples available for research to uncover the genetics underlying stroke. The ISGC has grown substantially during the years, and now encompasses >250 members from 6 continents, spanning >50 countries. With the enormous growth of the ISGC, we are excited to announce the ISGC’s Platform for Accelerating Genetic Discovery in Cerebrovascular Disease, which was awarded funding earlier this year. This platform will enable investigators around the world to access ISGC data, both summary and standardizing phenotype data to create harmonious data sets, and identification of ischemic and hemorrhagic stroke susceptibility loci. See www.strokegenetics.org for current workshops, all publications, and news from the ISGC.

Education is another important aspect of the ISGC. The ISGC holds biannual workshops with our most recent workshop in Barcelona, Spain and we are looking forward to our 19th workshop on April, 2016 in Cambridge, MA. The 20th workshop will be held in Milan, Italy on November 3 and 4, 2016. The workshops allow for collaborators to come together and discuss cutting-edge topics in stroke genetics and ongoing ISGC projects. In addition, every year the ISGC awards a travel scholarship to an ISGC community to attend the workshop. This year we were pleased to award the scholarship to Stacie Demel, DO, PhD from the University of Cincinnati.

We are excited for another productive year that includes the launch of the Platform for Accelerating Genetic Discovery in Cerebrovascular Disease and the upcoming workshop on April 28th and 29th at the Broad Institute of MIT and Harvard, hosted jointly with Massachusetts General Hospital and Harvard Medical School. The ISGC welcomes all with the skills and commitment to contribute to our efforts. Those interested in becoming a member or who seek more information about the consortium should join us.

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**Table. ISGC Founding Principles**

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<tr>
<th>Principle</th>
<th>Description</th>
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<td>Cerebrovascular disease is a complex disorder influenced by variation in many genetic and nongenetic exposures, each of which contributes only a small influence to disease risk. Therefore, large (larger than any single center can assemble on its own) well-characterized sample sets will be necessary to discover these exposures.</td>
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<td>ISGC principles of collaboration.</td>
<td>The ISGC is open to all who can contribute.</td>
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<td>All contributions are fairly recognized in publications.</td>
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<td>We work together in a spirit of cooperation and open communication to promote the best science in the present and the best science in the future.</td>
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ISGC indicates International Stroke Genetics Consortium.

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