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Institute for Systems Biology Uses Complete Genomics' Human Genome Sequencing Service to Verify Gene Responsible For Miller Syndrome; Results Published in *Science*

MOUNTAIN VIEW, Calif. – March 10, 2010 – Complete Genomics Inc., a third-generation human genome sequencing company, today announced that the Institute for Systems Biology (ISB) employed Complete Genomics' human genome sequencing service to sequence a family quartet to determine the depth of genetic information possible in analyzing a full family's sequence, and to verify the gene responsible for Miller syndrome, a rare craniofacial disorder. Results from this collaboration will be published later today in the journal *Science*; the manuscript is titled "Analysis of Genetic Inheritance in a Family Quartet by Whole Genome Sequencing."

"We are convinced that this new kind of analysis, family sequencing, will be a remarkably powerful scientific and medical tool in the future. ISB was delighted to work closely with the group at Complete Genomics to generate the data that enabled us to complete this study," said David Galas, professor and senior vice president of ISB.

ISB used Complete Genomics' service to sequence the genomes of a four-member nuclear family in which the two children suffer from Miller syndrome and ciliary dyskinesia, a lung disorder similar to cystic fibrosis, but neither parent is affected. The goal of this study was to identify rare genetic variations that could be responsible for Miller syndrome, and to estimate the intergenerational mutation rate.

Complete Genomics sequenced the four genomes to a depth of 51x to 88x, and 85-92% of the bases in each genome were called. A comparative analysis of the four genomes yielded four genes consistent with recessive inheritance of rare variations. One of these



genes, *DHODH*, was concurrently identified as a cause of Miller syndrome [Nat Genet. 2010 Jan;42(1):13-4]. Mutations in a second gene, *DNAH5*, have previously been shown to cause primary ciliary dyskinesia.

“When we established Complete Genomics, our goal was to provide large-scale complete human genome sequencing as a service that would enable our customers to make medically relevant discoveries. We are delighted that ISB is already making breakthroughs of that caliber from its first study using our service,” said Dr. Clifford Reid, chairman, president and CEO of Complete Genomics. “This is the type of positive disruptive influence that we want our technology to have on medical research.”

By providing a turnkey, outsourced complete human genome sequencing service, Complete Genomics provides researchers with research-ready data directly from DNA samples.

Customers send their DNA samples to Complete Genomics, which handles all the genome sequencing, data management and analysis. In return, customers receive their assembled sequences and variant reports including a functional annotation report. This annotation report describes the changes to gene products that each variation had caused, as well as annotations of the detected variations against public databases (dbSNP in particular), thus enabling comparison with other data.

About Complete Genomics

Founded in 2006, Complete Genomics is a California company that has developed a novel approach to sequencing human DNA that is revolutionizing the human genome sequencing industry. Complete Genomics combines its proprietary third-generation DNA sequencing technology with its high-performance computing capabilities to deliver low-cost, high-quality genomic data on an unprecedented scale. The company is currently building the world’s largest human genome sequencing center. This development will allow academic and biopharmaceutical researchers, for the first time, to conduct large-scale complete human genome studies that will help identify the genetic underpinnings of complex diseases and drug responses. For additional information about the company, please visit <http://www.completegenomics.com>.

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