

GeneDx now offers molecular testing for Cardio-facio-cutaneous (CFC) syndrome and Costello syndrome, and a new test protocol for Noonan syndrome

(April 2, 2006) Gaithersburg, MD—GeneDx, Inc., a leader in specialized genetic testing for rare genetic disorders, announced at the annual ACMG clinical genetics meeting in March a new look to its website at www.genedx.com and the launch of several new tests. In addition to a more efficient and cost-effective protocol for PTPN11 analysis in Noonan and LEOPARD syndromes, GeneDx has expanded its extensive menu with tests for two related disorders, Cardio-facio-cutaneous (CFC) syndrome and Costello syndrome. GeneDx now provides complete sequence analysis of the three major genes involved in these disorders with overlapping clinical features. BRAF gene testing in CFC syndrome will identify disease-causing mutations in 40%–78% of patients, while HRAS gene testing in Costello syndrome is expected to reveal mutations in 82%–92% of patients. The availability of molecular diagnostic testing for CFC, Costello and Noonan syndromes will aid physicians in differentiating among these syndromes and related conditions such as Kabuki syndrome, help in developing a targeted evaluation and management plan, and provide information for more accurate family counseling.

CFC, Costello and Noonan syndromes are complex developmental disorders including cardiovascular defects, developmental delay, mental retardation, facial dysmorphism, and a wide spectrum of other associated anomalies. Patients with Costello syndrome have a tumor predisposition and 10%–15% develop malignant tumors, such as rhabdomyosarcoma, neuroblastoma, ganglioneuroblastoma, and transitional carcinoma of the bladder. Postnatal growth deficiency, short stature and congenital heart defects, the most frequent of which are pulmonic stenosis, atrial septal defects and hypertrophic cardiomyopathy, are common manifestations of CFC syndrome. In Noonan syndrome, the cognitive deficits and

ectodermal problems are usually milder than in the other two disorders.

In early 2006, several research studies revealed that the clinical similarities between CFC, Costello and Noonan syndromes derive from a common molecular basis as mutations in each causative gene result in activation or alteration of the ancient Ras/MAP kinase pathway that controls cell proliferation and differentiation. The vast majority of mutations in BRAF (CFC syndrome), HRAS (Costello syndrome) and PTPN11 (Noonan syndrome) are de novo missense mutations, which can be detected with DNA sequence analysis as performed at GeneDx. Based on a reported clustering of mutations, GeneDx offers cost-effective two-tier testing in these genes. Tier 1 testing includes sequence analysis of hot spot regions. If no mutation is found, Tier 2 sequence analysis of the remaining coding exons of these genes is available.

Additional information on GeneDx, Inc., the entire test menu offered and molecular testing in CFC, Costello and Noonan syndromes can be found on the improved website at: www.genedx.com or contact:

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GeneDx introduces gene deletion/duplication testing for genetic disorders

(April 2, 2006) Gaithersburg, MD—GeneDx Inc., a provider of specialized genetic testing for rare genetic disorders, now offers a landmark test, CopyDxSM, for the diagnosis of deletion syndromes. CopyDxSM is a new service to determine the number of copies of a gene in a patient's genomic DNA based on the innovative use of quantitative PCR-based technology. Partial or complete gene deletions or duplications are a common cause for many birth defects and disorders resulting in mental retardation, such as X-linked hydrocephalus, steroid sulfatase deficiency, Kallman syndrome, Smith-Magenis syndrome, and Alagille syndrome, to name a few. CopyDxSM can

rapidly and reliably detect partial or whole gene deletions or duplications.

“This copy number analysis has a broad range of applications and will provide improved diagnosis for genetic disorders,” says Sherri Bale, President and Clinical Director of GeneDx. CopyDxSM allows the detection of gene deletions or duplications in X-linked disorders in affected males and carrier testing for at-risk female relatives. It will be a valuable diagnostic tool in autosomal genetic disorders where sequence analysis fails to disclose pathogenic mutations and haploinsufficiency is suspected. This technology can also be used to determine the specific genes deleted in patients with an autosomal contiguous gene deletion syndrome.

CopyDxSM analysis overcomes the limited resolution of conventional cytogenetic testing. Yuriy Shevchenko, Director of Research & Development at GeneDx, explains, “The high resolution of CopyDxSM analysis is achieved by probing very short genomic sequence segments (100 bp) and determining whether one or several copies are present in the genome.” Therefore, this method can identify small deletions on the gene level, which would not be detectable using FISH, and thus complements or replaces FISH and quantitative microarray analysis in single and contiguous deletion syndromes.

GeneDx diagnostic services, which include more than 100 tests for rare and ultra rare disorders based predominantly on DNA sequencing, now also encompass copy number analysis for detection of gene deletions/duplications. In addition, GeneDx welcomes inquiries into customized CopyDxSM service for copy number analysis of your gene(s) of interest.

Additional information on GeneDx, Inc., the entire test menu offered, and CopyDxSM analysis can be found on the new and improved website: www.genedx.com or contact:

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Ambry Genetics announces 508 FIRST™ cystic fibrosis test

\$59.00 assay detects the most common cystic fibrosis mutation

(March 14, 2006) Aliso Viejo, CA—Ambry Genetics has introduced 508 FIRST™, an inexpensive genetic test to tell quickly whether a patient with symptoms of cystic fibrosis (CF) has a genetic mutation known as DeltaF508. DeltaF508 causes up to half the cases of classic CF. Aside from DeltaF508, most CF mutations are rare.

Positive results are reported in less than one week for the small cost of \$59.00.

Samples from patients that prove negative for DeltaF508 will be automatically routed to The Ambry Test: CF AMPLIFIED™ for the most comprehensive analysis available. With a detection rate of approximately 99%, no other test can identify more mutations than The Ambry Test: CF AMPLIFIED™.

“508 FIRST™ is the optimum starting point for complete gene analysis of any cystic fibrosis patient,” said Charles Dunlop, president of Ambry Genetics. “508 FIRST™ provides a quick and affordable screen for the most likely positive result, with the security and confidence of full gene analysis in a single test. The certainty that comes with precise genetic results can reduce the cost of further tests, and help doctors provide the most effective care,” Dunlop said.

Because Ambry Genetics treats both phases of 508 FIRST™ as a single test, the process of arriving at a definitive diagnosis of CF is covered more easily by insurance.

Ambry Genetics is a worldwide leader in specialized genetic testing. The company’s advancements in testing for mutations related to cystic fibrosis, chronic and hereditary pancreatitis, alpha-1-antitrypsin deficiency, and other severe pulmonary diseases provide precise, cost-effective methods to aid in disease definition and early, more accurate diagnosis.

For more information, visit Ambry Genetics’ website: www.ambrygen.com or contact:

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EraGen Biosciences establishing strategic alliances within Wisconsin to address global health concerns

Innovative diagnostic tests in development throughout the state

(April 10, 2006) Madison, WI—EraGen Biosciences Inc., a venture-backed biotechnology company focused on automating molecular diagnostics, today announced the development of an innovative MultiCode diagnostic panel to address a range of clinical and research applications for respiratory diseases to include SARS and Avian Flu.

Because of its advantages over competing methods, MultiCode tests received the recent Frost & Sullivan Award for Product Innovation of the Year. “We are very confident in MultiCode products. Our successful collaborations with respected global clinical laboratories and business partners are proof that MultiCode is a very accurate and rapid means of detection,” said Irene Hrusovsky, MD, President and CEO, EraGen Biosciences. “This, plus the recent closing of our \$12 Million round of financing, allows us to further commercialize our company’s patented, expanded genetic alphabet.”

The MultiCode tests are currently being validated at the University of Wisconsin Hospital and Clinics. These innovative multi-infectious agent panels will minimize the need for traditional culture methods and reduce diagnostic time from weeks to hours. This is a critical factor since many treatments need to be administered within a day or two of onset.

“Collaborative efforts have been ongoing for a few years now with Wisconsin-based infectious disease world leaders,” said Jim Prudent, Ph.D., CSO of EraGen Biosciences, “Our state is in many ways a center of excellence when

it comes to viral influenza medical research and surveillance.”

Charles Hoslet, managing director of UW-Madison’s Office of Corporate Relations, which facilitates stronger relationships between the university and the business community, said EraGen’s initiative “is yet another great example of the collaborations underway that are putting Wisconsin at the forefront of finding innovative responses to global health care challenges.”

Hoslet pointed to the recent announcement of joint private and public support for the new Wisconsin Institutes for Discovery on the UW-Madison campus as “the best possible evidence that academic research, discovery, and commercial applications for a better world are truly coming together here in the Madison area.”

James E. Gern, M.D., Professor of Pediatrics, University of Wisconsin-Madison will be a sponsored speaker at the EraGen workshop at the 22nd Annual Clinical Virology Symposium later this month.

“We have found MultiCode technology to provide a superior means of scanning for respiratory diseases,” said Dr. Gern. “New respiratory viruses are continuing to be discovered, including new serotypes of coronavirus. The emergence of these new viruses provides a rationale for better antiviral testing.”

Other important on-going EraGen collaborations within the state include the BloodCenter of Wisconsin and Promega Corporation.

“BloodCenter of Wisconsin considers EraGen a key partner who enables us in our quest to remain at the cutting-edge of bioscience,” said Brian Curtis, Technical Director, Platelet & Neutrophil Immunology Laboratory, BloodCenter of Wisconsin.

“Promega recently introduced the Plexor technology which provides life science researchers new capability in real-time PCR,” said Doug Storts, Promega R&D Director. Plexor, which was just featured by Promega at the American Association of Cancer Research, was developed in collaboration with EraGen.

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About BloodCenter of Wisconsin

BloodCenter of Wisconsin is a multifaceted, world-renowned organization focused on blood collection, diagnostic testing, treatment, and research. The diagnostic laboratories develop and perform highly specialized tests on patient samples from throughout the United States and the world. They include matching potential organ donors and recipients, detecting risk factors for cancer and heart disease, and identifying various bleeding and clotting disorders. Customers rely on BloodCenter of Wisconsin's diagnostic laboratories for expert assistance to solve complicated clinical cases.

The organization's mission is to save patients' lives by providing blood products, medical expertise and scientific discovery. www.bloodcenter.com

About Promega

Promega Corporation is a leader in providing innovative solutions and technical support to the life sciences industry. The company's 1,450 products enable scientists worldwide to advance their knowledge in genomics, proteomics, cellular analysis, molecular diagnostics and human identification. www.promega.com

About University of Wisconsin

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About EraGen Biosciences

Based in Madison, Wisconsin, EraGen Biosciences is driving innovative molecular diagnostics to market faster. The company develops and commercializes high performance automated molecular diagnostics products to serve the growing markets for personalized medicine and rapid response needs for homeland security. www.eragen.com

The "New Products" page is designed to offer you news and information from businesses serving the genetics community. We welcome your submissions. All submissions are subject to review by the Editor. For more information, contact Sherry Reed, National Accounts Manager, Lippincott Williams & Wilkins, 351 West Camden Street, Baltimore, MD 21202; phone 410-528-8553; fax 410-528-4452; e-mail: sherry.reed@wolterskluwer.com.