



Asuragen and Collaborators Report Results Using PCR-only Technologies for the Comprehensive Molecular Assessment of the Fragile X Gene

Austin, Texas – July 26, 2010. Asuragen, Inc. announced today the results from two collaborative studies, one with the University of California Davis M.I.N.D. Institute and another with Rush University Medical Center, that demonstrate comprehensive molecular profiling of the Fragile X Mental Retardation (*FMR1*) gene using advanced PCR-based methods. Findings from the first study, titled “An Information-Rich CGG Repeat Primed PCR That Detects the Full Range of Fragile X Expanded Alleles and Minimizes the Need for Southern Blot Analysis,” were published online by the *Journal of Molecular Diagnostics* and will appear in print in the September 2010 issue. The results of the second study, titled “Applications of Novel PCR Technologies that Provide Enhanced Molecular Characterization of the Fragile X Gene” were recently presented at the 12th International Fragile X Conference in Detroit, Michigan by Andrew Hadd, Ph.D., Senior Scientist at Asuragen. Both studies significantly demonstrate Asuragen’s breakthrough PCR technologies by offering sensitive, specific, and robust detection of fragile X expanded alleles, and more informative genotyping data than existing methods.

Fragile X Syndrome (FXS) is one of several disorders linked to the expansion of CGG repeat sequences in the 5’ untranslated region of *FMR1*. Expansion to >200 triplet repeats often results in FXS, which affects about 1 in 5,000 individuals and is a leading cause of autism and the most common form of inherited intellectual disability. Approximately 1 in 130 to 260 people present 55-200 CGG repeats and are fragile X carriers. Carriers are at risk to develop fragile X-associated tremor/ataxia syndrome (FXTAS) or primary ovarian insufficiency (FXPOI). Although fragile X disorders impact more than 1 million people in the US alone, the vast majority of individuals at risk are unaware of their fragile X status. Current testing methods include Southern blot analysis, and are low throughput, time-consuming, and provide limited molecular information about the fragile X gene. The PCR strategies developed by Asuragen address each of these limitations.

“Based upon the results with more than 170 unique samples, including 75 full mutations, it is clear that the CGG repeat primed PCR represents a substantial improvement over current approaches for *FMR1* molecular assessments,” commented Flora Tassone, Ph.D., a senior author in the JMD study and biochemist at the UC Davis M.I.N.D. Institute. “This 3-primer assay design represents an important extension of work previously published by Dr. Paul Hagerman and me at the M.I.N.D. Institute. Combined with the novel, long read PCR capabilities of Asuragen’s technology, the repeat primed assay provides confidence for the detection of expanded alleles, irrespective of the number of triplet repeats. Furthermore, the assay definitively resolves zygosity in female samples and can rule out samples that would otherwise be needlessly analyzed by Southern blot. Lastly, the PCR can reveal both the number and sequence context of interrupting AGG elements that may impact the risk of CGG expansion in the next generation. No other technology can provide such a rich set of relevant molecular data from a single PCR reaction.”

In addition, Asuragen and Elizabeth Berry-Kravis, M.D., Ph.D., Professor of Pediatrics, Biochemistry, and Neurological Sciences at Rush University Medical Center, reported results from an independent study of 41 clinical samples using a prototype high resolution methylation PCR assay that can assess *FMR1* methylation concordant with both Southern blot analysis and patient phenotype. “Asuragen’s arsenal of fragile X PCR technologies is a leap forward,” said Dr. Berry-Kravis. “Our study included the full range of clinical phenotypes, from those with classic fragile X to those with milder symptoms, including patients with unmethylated full mutations. The combination of the CGG repeat primed PCR and the newly developed methylation PCR assay was able to correctly identify the number of repeats, even for low

abundance mosaic alleles, and accurately determined the methylation fraction in each allele. In addition, the capability to deduce the number of consecutive CGG triplet repeats in each patient sample may provide much needed insight into allele stability from parent to child, which is an area that has long been a blind spot for fragile X analysis. Taken together, Asuragen *FMR1* reagents offer the potential for a high throughput, PCR-only workflow for sample testing, substantially reducing the requirement for the laborious Southern blot.”

The CGG repeat primed PCR reagents described in both studies are available as a Research Use Only (RUO*) kit manufactured by Asuragen. “AmplideX™ *FMR1* products reflect our 22-year heritage of developing cutting edge molecular biology-based technologies, starting at our predecessor company Ambion,” commented Matt Winkler, Ph.D., CEO of Asuragen. “Our growing number of molecular diagnostic products exemplifies our efforts to provide laboratories with improved sensitivity, workflow and information content to positively impact patient care.”

*For Research Use Only. Not For Use in Diagnostic Procedures.

About Asuragen

Asuragen is a fully integrated diagnostic development company and pharmaceutical services provider. The Company’s diagnostic product portfolio consists of the first-ever validated microRNA diagnostic assay for pancreatic cancer, quantitative RNA tests for leukemia gene translocations, and the Signature® Oncology and Genetic Testing products. Asuragen is empowered with a high level of scientific expertise and assay development capabilities, CLIA and GLP testing services, and an established cGMP manufacturing facility, which allow it to span the spectrum of discovery, testing, production and commercialization. For more information, visit www.asuragen.com.

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