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Athena Insight™ Helps Bring Results to a New Level with Additional Scientific Data

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Since its initial launch in 2010, the Athena Diagnostics Insight program continues to build an impressive and noteworthy data pipeline for the reporting and interpretation of Variants of Unknown Significance. Athena Insight was developed by Athena Diagnostics to help take the complexity out of understanding variants of unknown significance, and to provide answers in a simplified report. This analysis can save physicians time and provide them with enhanced test reports containing significant scientific data to help improve patient treatment and care.

Since interpretation is important to patient management, the Athena Insight team accumulates and integrates data to provide physicians key information that characterizes variants as benign, pathogenic, or somewhere in between. To manage the growing library of data, Athena formed a Variant Investigation Team responsible for delivering comprehensive genetic test results through extensive investigation, variant by variant. The team conducts in-depth reviews of both internal and external data as a part of the validation process.

Clinicians have begun taking notice of the Athena Insight program over the past few months. With the increase of recorded data, results comparison and comprehensive cross-reference study, the Variant Investigation Team has actually been able to reclassify some variant cases from causative to benign using their comprehensive variant investigation process.

The Athena Insight program goes beyond simple results. Athena Diagnostics considers follow-up as an important step in its due diligence by continuing to monitor data, identify variants and assist physicians with interpretation to improve the diagnostic accuracy.

To extend the impact of their investigation, the Athena Insight team is also joining forces with the scientific community, helping to use variant investigative data in the clinical setting. At the November 2010 American Society of Human Genetics (ASHG), Athena presented research concentrating on SCN1B sequence

variants, missense variants and predictive age-related mutations. As a leader in genetic testing for epilepsy, Athena is in a unique position to collaborate with researchers to better assess clinical pathogenicity of SCN1B sequence variants. These collaborations are especially critical for variants that are rare and have limited segregation, population studies and co-occurrence available.

Recently, while testing for Hereditary Spastic Paraplegia (HSP), Athena Diagnostics has identified 14 individuals carrying a specific mutation with a predictive age of onset. This research could prove useful in helping to better assess family members risk and potential clinical treatment. Additionally, it may help to determine a mechanism for understanding such a varied age of onset.

In the future, Athena Diagnostics is planning to explore more clinical data than is available at present. In that scenario, almost any variant could be paired and integrated worldwide with clinical information for more precise interpretation. Athena Diagnostics acknowledges that their customers are instrumental in driving the Athena Insight program to new levels of value, finding what variants mean and how they relate to disease. Tests require the rigor of consistency and the diligence of interpreting data to deliver more clinically significant results into a report. On this premise, Athena Diagnostics works in partnership with customers to provide testing that makes a difference.

Athena Diagnostics strives to communicate genetic diagnostic guidance to physicians. The Variant Investigation Team remains steadfast in delivering precise and exhaustive investigation processes that have profound impact upon patient management. Athena's Genetic Counselors and Laboratory Directors are accessible to interpret test results and answer technical questions as they arise.

The Athena Insight Success Story at a Glance

- 1,095 unique variants studied to date
- 1,308 client reports sent
- 75 of 146 genes launched
- Athena Insight profiles include Epilepsy, CH, CADASIL, HSPs, Autism, MR, ALS, Parkinsonism, Migraine, Ataxia, and Mitochondrial Disease
- Of the gene variants so far, the Athena Insight program has been able to conclusively classify 148 variants as notmal (benign polymorphisms) and 28 variants as pathogenic (positive).

Thank you for your response.