Insight Pharma Reports Announces Advances in Clinical Genome Sequencing and Diagnostics

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FOR IMMEDIATE RELEASE

NEEDHAM, MASS. – April 3, 2013 – Cambridge Healthtech Institute's Insight Pharma Reports announces the Advances in Clinical Genome Sequencing and Diagnostic, authored by Kevin Davies, PhD.

This report explores the recent surge in clinical genome sequencing, from the point of view of the sequencing providers, the medical organizations delivering these services, and the start-ups offering a variety of interpretation services, platforms, and business models. Aspects discussed and presented include:

- Progress in clinical genome sequencing
- Organizations leading the way in generating clinical data and its interpretation.
- Determining the causality of documented variants in genetic disease.
- Clinical genome sequencing in oncology.
- Academic and commercial clinical genomics providers.
- The next-gen sequencing landscape.
- Companies providing genome interpretation software.
- Initiatives in setting sequencing standards.
- Interviews with six industry experts, conducted exclusively for this report.
- Results of a custom survey on clinical genome sequencing.
- A list of print and online resources for further investigation into this area.

For many years now, next-generation sequencing (NGS) has been used in clinical research, building on the success of being able to sequence personal genomes affordably, and turning that technology into defining the mutation profile of rare Mendelian diseases and cancer. With

the first report of successful exome sequencing in a patient with a mystery illness, the clinical community has embraced NGS—performed in CLIA- and CAP-certified laboratories—for diagnostic testing.

Nevertheless, at the $1,000–5,000 price point, medical practitioners face a dilemma: It is possible to offer to sequence the exome or complete genome of a patient for roughly the same price as a traditional patented genetic diagnostic test or gene panel. Economics alone would appear to dictate that the practice of medical genetics and clinical diagnostics must evolve radically in the face of the remarkable advances in NGS. That sentiment has been bolstered in recent years with more and more anecdotal stories of the identification of mutations in patients suffering mysterious/undiagnosed (presumably) genetic disorders. Emboldened by these success stories, medical centers, sequencing platform providers, and diagnostics companies are rethinking their strategies for delivering exome and/or whole-genome sequencing services.

In a further irony, the cost of sequencing has fallen so fast that it is now a relatively trivial component of the full array of services required to deliver clinical genome information—what some have (literally or tongue-in-cheek) called the $100,000 or $1 million interpretation. Leaving aside the apples-and-oranges debate of the precise cost of genome interpretation, there is no doubt that 2012 proved to be a stunning year for the maturation of clinical genome testing. Several prominent, public stories relating to the successful end of diagnostic odysseys provide gratifying examples of the potential of this technology. Cases of families receiving a confirmed diagnosis after years of false hope and hundreds of thousands of dollars in medical costs show the benefit of this approach, and beg the question of how long the molecular diagnostics industry will continue in its single-gene/multigene panel approach. (After all, the exome is essentially just one giant gene panel with 22,000 genes.)

Exciting, remarkable progress in clinical genome sequencing is being fueled by steady advances in existing platforms, the arrival of new diagnostic platforms, and improvements in genome analysis software. That said, the adoption of clinical NGS is not trivial, and many questions still remain about setting standards, ensuring analytic and clinical validity of the tests, and reimbursement.

For more information and to purchase this Insight Pharma Report, visit: http://www.InsightPharmaReports.com or contact Kerri Simpson at ksimpson@healthtech.com or 781-972-1347.

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