CeGaT offers fast and cost efficient Genetic Screening via Next-Generation Sequencing

28 subpanels for nearly 400 genes are available as screening tool to test for genes involved in diseases like Parkinson, ALS, Dementia, Epilepsy and Hereditary Eye Diseases.

Tübingen, Germany, October 10, 2010: CeGaT today announced that it is offering Sure Select Enrichment and SOLiDTM System sequencing-based genetic screening for hereditary eye diseases, epilepsy, metabolic and neurodegenerative disorders.

CeGaT is partnering with world known experts in each disease to develop and implement panels to screen dozens of genes simultaneously. Detlef Boehm, PhD, Head of Laboratory and Specialist Next-Generation-Sequencing of CeGaT: "Next-Generation Sequencing offers possibilities as fast and cost efficient screening tool that is now available to researchers and clinicians worldwide." CeGaT is focussing on the above mentioned diseases but has already a neuro-muscular panel in the pipeline and is approached by research centres to develop panels for other diseases as well.

"A 100% detection rate and no false negatives led to the fast implementation of the panels in a clinical research setting, said Saskia Biskup, MD PhD, CEO and Founder of CeGaT. "The SOLiDTM System is used for high-throughput screening, but we still rely on Sanger sequencing to verify a positive test result before we sent out a report."

Currently available panels include genes associated with Parkinson, ALS, dementia, non-syndromic and syndromic epilepsy, metabolic disorders and genes associated with hereditary eye diseases.

Epilepsy and metabolic disorders (15 Panels - 231 genes): Epilepsy is a very common neurologic disorder and is more likely to occur in early childhood. This early occurrence makes a genetic cause possible especially if other family members are also affected. Epilepsy can be the only symptom of the disease (primary, non-syndromal epilepsy) but can also present in combination with other symptoms (syndromal epilepsy). To enable a fast and cost efficient screening, CeGaT has developed 15 panels: Generalized / Myoclonic Epilepsy, Febrile Seizures, Absences (27 genes), Epileptic Encephalopathies (12 genes), Epilepsy and X-linked Mental Retardation (12 genes), CDG (Congenital Disorder of Glycosylation) Syndrome (23 genes), Ceroidlipofuscinosisis (8 genes), MPS and Mucolipidosis (13 genes), Zellweger Syndrome (8 genes), Metabolic Disorders with Epilepsy (38 genes), Coenzyme Q Deficiency Syndrome (5 genes), Selected Mitochondrial Disorders (23 nuclear encoded genes), Joubert Syndrome (10 genes), Lissencephaly and Polymicrogyria (18 genes), Microcephaly and Pontocerebellar Hypoplasia (16 genes), Neuro-cardio-facio-cutaneous Syndrome (11 genes), Walker-Warburg Syndrome (7 genes).
Hereditary Eye Diseases (11 panels - 126 genes): Hereditary eye diseases are a very heterogeneous group of diseases that have one thing in common: vision loss. Identifying the genetic cause of the disease has important implications for the prognosis and but also for treatment. Vision loss can occur as the only symptom (non-syndromic) or in combination with other symptoms (syndromic) with deafness being one example. CeGaT has developed 11 Panels to diagnose Retinitis pigmentosa, Achromatopsie, Congenital stationary night blindness, Cone-rod dystrophies, Leber congenital amaurosis, Usher Syndrome, Bardet Biedl Syndrome, Primary ciliary dyskinesia, Refsum disease and Senior Loken syndrome.

Dementia, Parkinson and Amyotrophic Lateral Sclerosis (ALS) (2 Panels - 35 genes): Genetic causes of dementia and movement disorders have been described and although in many instances treatment is not possible, a correct diagnosis is helpful to predict progression and to counsel family members. CeGaT has a main research focus on identifying novel disease causing genes for neurodegenerative disorders in collaboration with the University of Tuebingen, Germany. Genes are the first clue towards understanding of the underlying pathogenic mechanisms and will be the basis for future therapeutic intervention. Early diagnosis of individuals with susceptibility to neurodegeneration will be the only way to start treatment before it is too late. Genetic screening will help to define subgroups of patients to further enable personalized medicine.

All disease panels are offered at sequencing costs of EUR 3,000. Costs for analysis, validation and interpretation depend on the number of genes that are requested. With respect to ongoing dropping sequencing costs, CeGaT is very optimistic that genetic tests will soon be available as routine and affordable screening methods.

CeGaT is a diagnostic sequencing company and official service provider for life technologies located in Tuebingen, Germany. CeGaT was founded in 2009 by Saskia Biskup, MD PhD. CeGaT is specialized in Molecular Diagnostics, Next-Generation-Sequencing, combining technical, bioinformatical and medical know-how.

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