

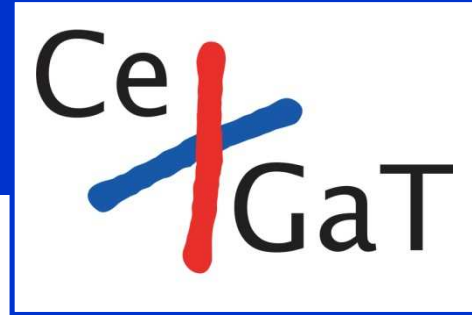
Ce  GaT

**DIAGNOSTIC
PANELS**

Center for Genomics
and Transcriptomics

March 2011

Overview



- **Located in the technology park TTR in Tübingen**
- **6 separated rooms (270 sqm in total)**
- **Fully equipped state-of-the-art laboratory**
- **QM / Accreditation Q1 2011; first audit highly successful**
- **Molecular Diagnostics for all known disease associated genes**
- **Next-Generation-Sequencing including consultation and full bioinformatic analyses**
- **Diagnostics-Panels**
- **Service Provider for Applied Biosystems**

Technology



➤ SOLiD 4 System

- Targeted Re-Sequencing
- Genome / Exome Sequencing
- Transcriptome Sequencing
- Epigenome Sequencing
- Micro RNA Sequencing
- Diagnostic Panels

Upgrade to SOLiD 5500xl already ordered



➤ 96 Capillary Sequencer

- Diagnostics
- Validation of SOLiD results



Services – Molecular Diagnostics



➤ Molecular Diagnostics

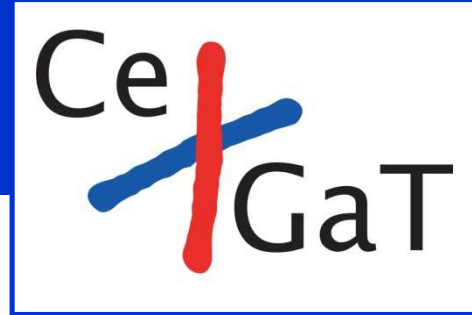
- Sequencing of single genes
- Interpretation of results; report takes into consideration state-of-the-art knowledge
- Offered for all known disease-causing variants in human genome
- Specialization in
 - Dementia / ALS
 - Parkinson
 - Dystonia
 - Epilepsy
 - Hereditary Eye Diseases
- Average turn around times of 1 to 2 weeks
- International customers ranging from Canada to New Zealand



➤ **Next-Generation-Sequencing**

- **Service Provider for Universities, Researchers, and Companies**
- **Genome, Exome, Transcriptome, Epigenome, microRNA, and Re-Sequencing**
- **Project Consultation**
- **Full Bioinformatic services**

Services – Diagnostic Panels



➤ Diagnostic Panels

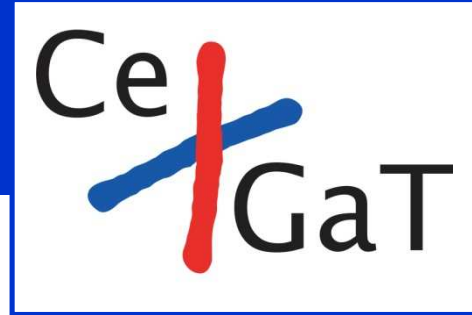
- Sequencing of all genes related to a certain disease simultaneously on the SOLiD 4 platform
- Re-Sequencing of variants on Capillary Sequencer
- Interpretation of results

➤ Diagnostic Panels introduced May 2010:

- 15 different Epilepsy Panels (265 Genes in total)
- 15 different Retina Panels (196 Genes in total)
- Dementia and ALS Panel (20 Genes)
- Parkinson Panel (16 Genes)

**DIAGNOSTIC
PANELS**

Services – Diagnostic Panels II



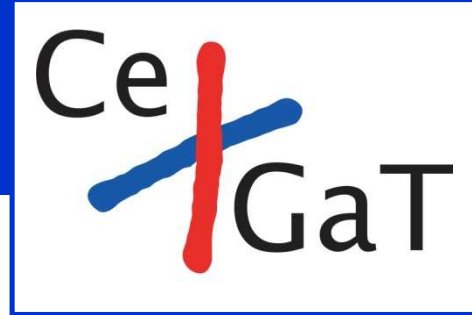
Epilepsy and metabolic disorders (15 Panels - 265 genes):

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), Ceroidlipofuscinosis (8 genes), MPS and Mucopolysaccharidosis (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).

**DIAGNOSTIC
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Services – Diagnostic Panels III



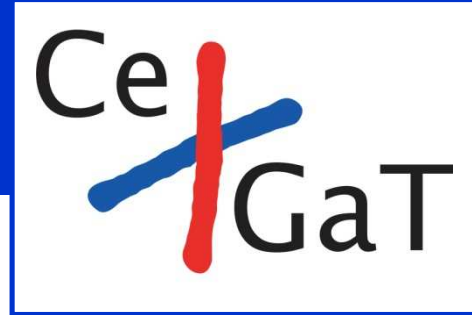
Hereditary Eye Diseases (15 panels – 196 genes):

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. In total more than 180 genes have been described to cause non-syndromic and syndromic vision loss. CeGaT has developed 11 Panels:

Usher syndrome (10 genes), Autosomal dominant retinitis pigmentosa (26 genes), Autosomal rezessive retinitis pigmentosa (28 genes), Achromatopsia (3 genes), Bardet Biedl syndrome (14 genes), Congenital stationary night blindness (11 genes), Joubert syndrome (9 genes), Leber congenital amaurosis (8 genes), Primary ciliary dyskinesia (6 genes), Refsum disease (5 genes), Senior Loken syndrome (6 genes), Stargardt disease and macular dystrophies (11 genes), Cone rod dystrophies (25 genes), Flecked retina disorders (6 genes)

**DIAGNOSTIC
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Services – Diagnostic Panels IV



Parkinson, Dementia and 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. CeGaT has developed two panels:

Parkinson (16 genes), Demetia and ALS (19 genes)

**DIAGNOSTIC
PANELS**

CEO & President



Dr. med. Dr. rer.nat. Saskia Biskup

- Born in Frankfurt 1971
- MD, University of Würzburg, 1999
- MD PhD, University of Würzburg, 2002
- Bioinformatics at the University of Heidelberg 2002
- „Assistenzärztin“ Institute of Human Genetics, TU München, 2002 - 2005
- Post Doc at Johns Hopkins University in Baltimore, MD, USA, 2005-2008
- „Assistenzärztin“ Institute of Human Genetics & IZKF Junior Group (Parkinson) University of Tübingen, 2008 - 2010
- „Fachärztin“ Human Genetics, 2009; Praxis für Humangenetik Tübingen
- Research Interest: Parkinson, especially LRRK2



Head of Laboratory



Dr. rer.nat. Detlef Böhm

- Born in Heinebach / Melsungen 1965
- Biology University of Göttingen, 1999
- PhD, University of Göttingen, 2001
- Institute of Human Genetics, Göttingen, 2001-2005
- Internal Medicine, University Clinic, Freiburg, 2006
- Head of Laboratory “Praxis für Humangenetik”, Freiburg, 2006-2009
- Head of Laboratory CeGaT GmbH since 2009
- Pioneer in establishing new technologies in human genetics



Kontakt



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