

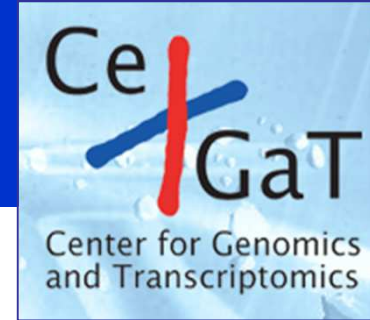


Center for Genomics
and Transcriptomics

March 2012

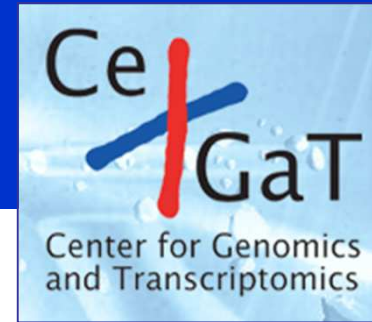


Overview



- **Located in the technology park TTR in Tübingen**
- **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**

Available Technology

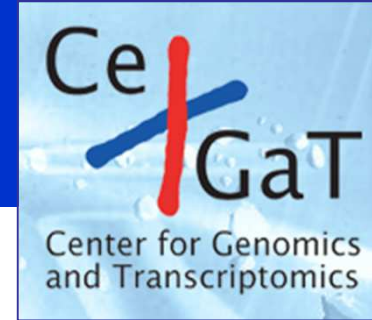


- **2 x SOLiD 4 System**
- **2 x SOLiD 5500xl System**
- **IonTorrent PGM**
 - Targeted Re-Sequencing
 - Genome / Exome Sequencing
 - Transcriptome Sequencing
 - Micro RNA Sequencing
 - Diagnostic Panels

- **96 Capillary Sequencer**
 - Diagnostics
 - Validation of SOLiD results



Business Areas



Molecular Diagnostics

Dementia/ALS
Parkinson
Dystonia
Epilepsy
Hereditary Eye Diseases
Rare Diseases

....

NGS

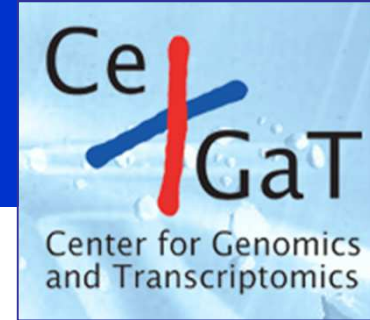
Exome
Transcriptome
Genome
microRNA
Metagenome

Diagnostic Panels

Parkinson
Dementia and ALS
Epilepsy
Hereditary Eye Diseases

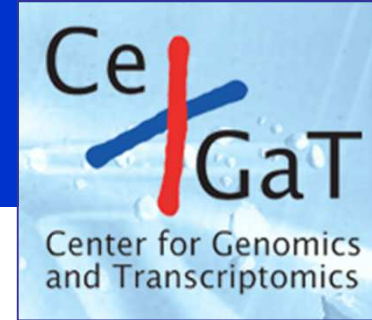
Neuromuscular Diseases

FISH



- **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
 - Rare Diseases
- **Average turn around times of 1 to 4 weeks**
- **International customers ranging from Canada to New Zealand**

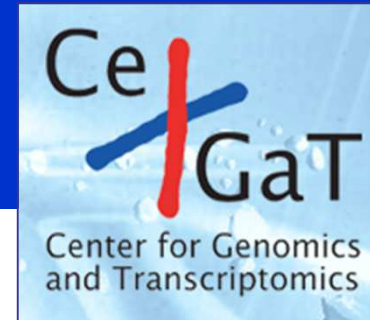
Next-Generation-Sequencing



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



DIAGNOSTIC PANELS



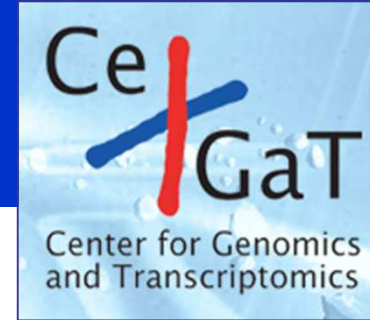
- **Parallel Sequencing of all genes related to a certain disease**
- **Re-Sequencing of variants on Capillary Sequencer**
- **Interpretation of results**

For each Panel the following steps are performed:

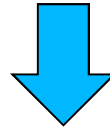
- (1) Enrichment of DNA sample with special CeGaT-Panel and preparation of sequencing in the lab
- (2) Massive parallel sequencing of all genes in the Panel using NGS (SOLID 5500xl or Ion Torrent PGM)
- (3) Bioinformatical evaluation: Potential disease causing variants and regions that are underrepresented are identified
- (4) Post sequencing of variants and underrepresented regions using the gold standard Sanger sequencing as an independent confirmation method,
- (5) Final bioinformatical and clinical evaluation of the complete SNV lists of all genes
- (6) Issuing of medical report.



**DIAGNOSTIC
PANELS**



In approx. 80-90% of clinically diagnosed complex genetic diseases the causative gene variation is not found



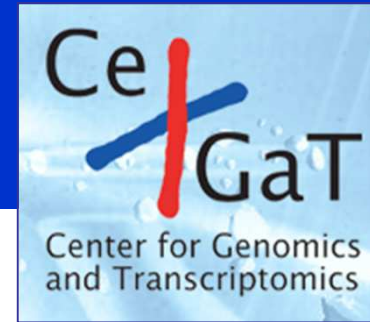
Classical sequencing of gene by gene is
excessively expensive and **very time consuming**

Advantages of using Diagnostic-Panels compared to gene by gene sequencing:

- (1) Much faster
- (2) Considerably less expensive
- (3) The probability of finding the causative gene variation is significantly higher

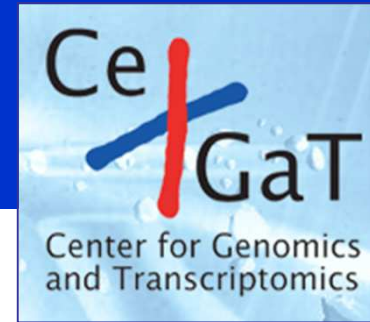
Why clarifying the genetic cause of a Disease?

DIAGNOSTIC PANELS



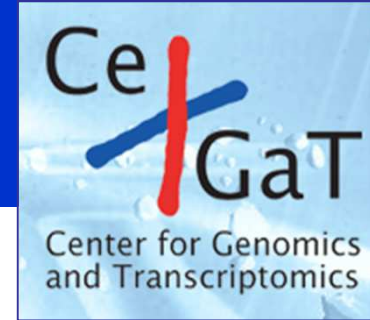
Clarifying the genetic cause in affected families is necessary to:

- (1) Secure a clinical diagnosis
- (2) Be able to offer a targeted examination of other family members
- (3) Make an early therapeutic intervention possible
- (4) Provide a prognostic assessment of the course of the disease
- (5) Provide the basis for new therapeutic methods in the long-term.



➤ **Available Diagnostic-Panels**

- **ALS, FTD, Dementia, Parkinson, Dystonia, Neuroacanthocytosis, and NBIA**
(7 Panels - 280 Genes)
- **Epilepsy & Metabolic Disorders**
(20 Panels - 327 Genes)
- **Hereditary Eye Diseases**
(15 Panels - 206 Genes)
- **Neuromuscular Diseases**
(10 Panels - 500 Genes)
- **Hereditary Hearing Diseases**
(3 Panels - 80 Genes)
- **Pharmacogenomics**
(under construction)



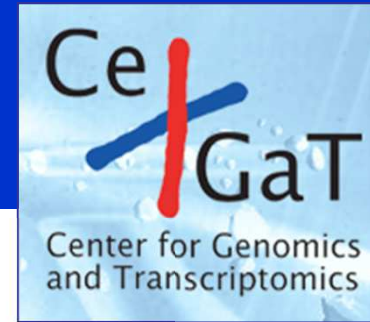
Saskia Biskup, MD, PhD

- 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

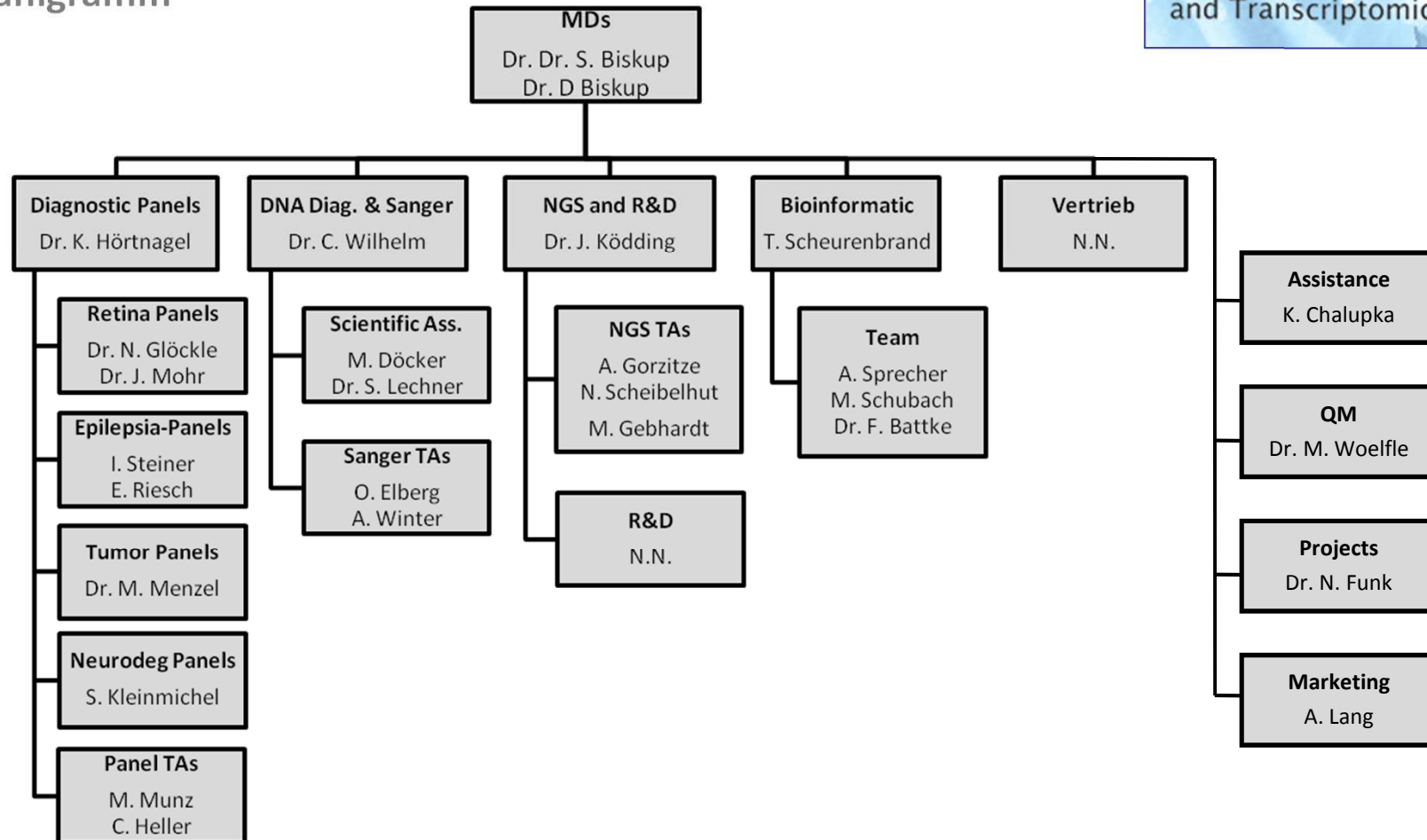
- Research Interest: Parkinson, especially LRRK2



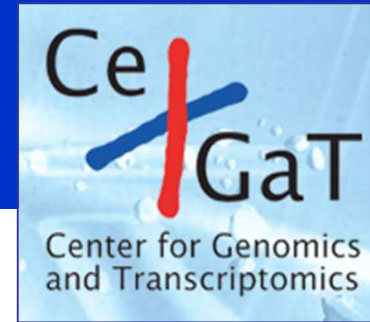
Organization 2012



Organigramm

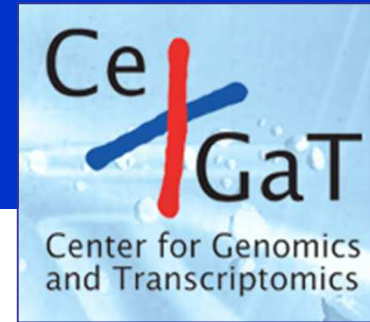


Development of CeGaT



- 2009: Foundation of CeGaT & Start of business
- 2010: Sales EUR 1m and Break-Even
- 2010: Introduction of Diagnostic Panels
- 2011: Sales of more than EUR 2m, profitable
- 2011: Awarded Best German Start-Up
- 2012: B Braun Melsungen AG, a EUR 4.4bn company, acquires 20% interest in CeGaT → long-term strategic partnership

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