pharm-analyt – BIOMARKER SUCCESS STORY

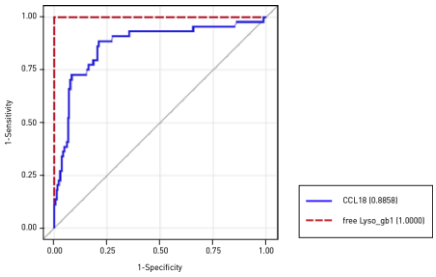
BACKGROUND

Continuous individualization in health care continues to urge for suitable biomarkers. Triggered by specific clinical demand, pharm-analyt started its R&D work in the area of rare diseases. After intensive, targeted investigation, highly specific and sensitive biomarkers could be discovered and validated for several Lysosomal Storage Diseases.

SMALL MOLECULE MARKERS

Our R&D breakthroughs have shown that endogenous Small Molecules can have highly specific and sensitive marker-characteristics (see graph below).

ROC Curve of Lyso Gb1 (Gaucher)



LSD – Lysosomal Storage Diseases

Validated with several hundred clinical samples, these markers are being used for diagnostic and therapeutic purposes.

Examples for LSD Markers

Fabry: Pioneers using lyso-Gb3 in plasma and DBS, Publications: 8, several posters

Gaucher: pharm-analyt discovered / patented lyso-Gb1 in plasma
(patent: US 14/124,375 or EP12 728 976.7)
Publications: 1, several posters
Patent was sold to Centogene AG (2016)

NP-C (Niemann-Pick-Typ C): pharm-analyt discovered / patented lyso-Sphingomyelin and 509 in plasma
(US 14/358,669 or EP 12 790 426.6)
Patent was sold to Centogene AG (2016)
Publications: 3, 1 poster

MLD (Metochromatic Leukodystrophy):
pharm-analyt discovered / patented lyso-Sulfatide in plasma (US 14/651, 450 or EP 13 815 378.8)
Patent was sold to Centogene AG (2016)

Current Marker Discovery Projects

Pompe: One prospect molecule recently found, currently in validation process. Scouting for more patient samples.

Krabbe: Differentiation to Gaucher by HPLC-MS/MS separation of Glucose- and Galactose-Sphingolipid at pharm-analyt possible.

MPS: Currently working on differentiation of the various types by HPLC-MS/MS (different sulfation, different mol. weight)

Other Indications

SEPSIS: We have just submitted an European Patent for a biomarker with similar potency like Procalcitonin but with another focus.

STROKE: Ongoing work

COPD: Ongoing work

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Publications in Peer Reviewed Journals

FABRY

Johnson B, Mascher H, Mascher D, Legnini E, Hung CY, Dajnoki A, Chien YH, Maródi L, Hwu WL, Bodamer OA.
Ann Lab Med. 2013, 33, 274-8
Analysis of **Lyso-Globotriaosylsphingosine** in Dried Blood Spots

Markus Niemann, Arndt Rolfs, Anne Giese, Hermann Mascher, Frank Breunig, Georg Ertl, Christoph Wanner, Frank Weidemann
Publisher: J Inherit Metab Dis Report, 2013, 7, 99-102
Lyso-GB3 Indicates that the Alpha-Galactosidase A Mutation D313Y is not Clinically Relevant for **Fabry Disease**

Jan Lukas, Anne-Katrin Giese, Arseni Markoff, Ulrike Grittner, Ed Kolodny, Hermann Mascher, Karl J. Lackner, Wolfgang Meyer, Phillip Wree, Viatcheslav Saviouk, Arndt Rolfs
PLOS Genetics, August 2013, Vol. 9, Issue 8, 1-10
Functional Characterisation of Alpha-Galactosidase A Mutations as a Basis for a New Classification System in **Fabry Disease**

Arndt Rolfs, Hermann Mascher, several authors
Stroke 2013, 44(2):340-9, Published online Jan. 10, 2013
Acute Cerebrovascular Disease in the Young: the **Stroke in Young Fabry Patients** (sifap) Study

Chien YH, Bodamer OA, Chiang SC, Mascher H, Hung C, Hwu WL.
J Inherit Metab Dis., 2012, Oct.
Lyso-globotriaosylsphingosine (lyso-Gb(3)) levels in neonates and adults with the **Fabry Disease** later-onset GLA IVS4+919G>A mutation

Jan Lukas, Joan Torras, Itziar Navarro, Anne-Katrin Giese, Tobias Böttcher, Hermann Mascher, Karl J. Lackner, Guenter Fauler, Eduard Paschke, Josep M. Cruzado, Ales Dudesek, Matthias Wittstock, Wolfgang Meyer, Arndt Rolfs
Clinical Kidney Journal, 2012, 5, 395-400
Broad spectrum of **Fabry Disease** manifestation in an extended Spanish family with a new deletion in the GLA gene

Markus Niemann, Arndt Rolfs, Anne Giese, Hermann Mascher, Frank Breunig, Georg Ertl, Christoph Wanner, Frank Weidemann
Journal of Inherited Metabolic Disease Report, 2012, July
Lyso-Gb3 Indicates that the Alpha-Galactosidase A Mutation D313Y is not Clinically Relevant for **Fabry Disease**

C. Tanislav, M. Kaps, A. Rolfs, T. Böttcher, K. Lackner, E. Paschke, H. Mascher, M. Laue, F. Blaes,
Eur J Neurol, 2011, 18, 631-636
Frequency of **Fabry Disease** in patients with small-fibre neuropathy of unknown aetiology: a pilot study

GAUCHER

A. Rolfs, A-K Giese, U. Grittner, D. Mascher, D. Elstein, A. Zimran, T. Böttcher, J. Lukas, R. Hübner, U. Gölnitz, A. Röhle, A. Dudesek, W. Meyer, M. Wittstock, H. Mascher
Publisher: PLOS ONE, 2013, 8(11)
Glucosylsphingosine Is a Highly Sensitive and Specific Biomarker for Primary Diagnostic and Follow-Up Monitoring in Gaucher Disease in a Non-Jewish, Caucasian Cohort of **Gaucher Disease** Patients

NP-C

Michaela Trilck, Franziska Peter, Chaonan Zheng, Marcus Frank, Kostantin Dobrenis, Hermann Mascher, Arndt Rolfs, Moritz J. Frech
Brain Research 2017, 1657, 52 – 61
Diversity of glycosphingolipid GM2 and cholesterol accumulation in **NPC1** patient-specific iPSC-derived neurons

Anne-Katrin Giese, Hermann Mascher, Ulrike Grittner, Sabrina Eichler, Guido Kramp, Jan Lukas, Danielle te Vruchte, Nada Al Eisa, Mario Cortina-Borja, Forbes D Porter, Frances M Platt, Arndt Rolfs
Publisher: Orphanet Journal of Rare Diseases, 2015, 10:78
A novel, highly sensitive and specific biomarker for **Niemann-Pick Type C1 Disease**

Marine Hovakimyan, Oliver Stachs, Maria Reichard, Jan Lukas, Moritz Johannes Frech,
Rudolf Guthoff, Martin Witt, Hermann Mascher, Arndt Rolfs, Andreas Wree
CORNEA, 2011, 30, 796-803
Morphological Alterations of the **Cornea** in the Mouse Model of **Niemann-Pick Disease Type C1**