

## T. BECCARI: ELENCO PUBBLICAZIONI 2004-2013

1. Paciotti S, Persichetti E, Klein K, Tasegian A, Duvet S, Hartmann D, Gieselmann V, Beccari T (2014). Accumulation of free oligosaccharides and tissues damage in cytosolic  $\alpha$ -mannosidase (Man2c1)-deficient mice. *J Biol Chem* 289, 9611-22.
2. Parnetti L, Chiasserini D, Persichetti E, Eusebi P, Varghese S, Qureshi MM, Dardis A, Deganuto M, De Carlo C, Castrioto A, Balducci C, Paciotti S, Tambasco N, Bembi B, Bonanni L, Onofrj M, Rossi A, Beccari T, El-Agnaf O, Calabresi P (2014). Cerebrospinal fluid lysosomal enzymes and  $\alpha$ -synuclein in Parkinson's disease. *Mov Disord*
3. Nardicchi V, Ferrini M, Pilolli F, Angeli EB, Perichetti E, Beccari T, Mannucci R, Arcuri C, Donato R, Dorman RV, Goracci G (2014). NGF Induces the Expression of Group IIA Secretory Phospholipase A<sub>2</sub> in PC12 Cells: The Newly Synthesized Enzyme is Addressed to Growing Neurites. *Mol Neurobiol*
4. Van Dijk KD, Persichetti E, Chiasserini D, Eusebi P, Beccari T, Calabresi P, Berendse HW, Parnetti L, van de Berg WD (2013). Changes in endolysosomal enzyme activities in cerebrospinal fluid of patients with Parkinson's disease. *Mov Disord* 28, 747-54.
5. Paciotti S, Persichetti E, Pagliardini S, Deganuto M, Rosano C, Balducci C, Codini M, Filocamo M, Menghini AR, Pagliardini V, Pasqui S, Bembi B, Dardis A, Beccari T (2012). First pilot newborn screening for four lysosomal storage diseases in an Italian region: identification and analysis of a putative causative mutation in the GBA gene. *Clin Chim Acta* 413, 1827-31.
6. Persichetti E, Klein K, Paciotti S, Lecointe K, Balducci C, Franken S, Duvet S, Matzner U, Roberti R, Hartmann D, Gieselmann V, Beccari T (2012). Lysosomal di-N-acetylchitobiase-deficient mouse tissues accumulate Man2GlcNAc" and Man3GlcNAc2. *Biochim Biophys Acta* 1822, 1137-46.
7. De Marchis F, Balducci C, Pompa A, Riise Stensland HM, Guaragno M, Pagiotti R, Menghini AR, Persichetti E, Beccari T, Bellucci M (2011). Human  $\alpha$ -mannosidase produced in transgenic tobacco plants is processed in human  $\alpha$ -mannosidosis cell lines. *Plant Biotechnol J* 9, 1061-73.
8. Schiavoni G, Bennati AM, Castelli M, Fazia MA, Beccari T, Servillo G, Roberti R (2010) Activation of TM7SF2 promoter by SREBP-2 depends on a new sterol regulatory element, a GC-box, and an inverted CCAAT-box. *Biochim Biophys Acta* 1081, 587-92.
9. Tappino B, Chuzhanova NA, Regis S, Dardis A, Corsolini F, Stroppiano M, Tonoli E, Beccari T, Rosano C, Mucha J, Blanco M, Szlago M, Di Rocco M, Cooper DN, Filocamo M (2009) Molecular characterization of 22 novel UDP-N-acetylglucosamine-1-phosphate transferase alpha- and beta- subunit (GNPTAB) gene mutations causing mucolipidosis types IIalpha/beta and IIIalpha/beta in 46 patients. *Hum Mutat* 30, 956-73.
10. Persichetti E, Chuzhanova NA, Dardis A, Tappino B, Pohl S, Thomas NS, Rosano C, Balducci C, Paciotti S, Dominissini S, Montalvo AL, Sibilio M, Parini R, Rigoldi M, Di Rocco M, Parenti G, Orlacchio A, Bembi B, Cooper DN, Filocamo M, Beccari T (2009). Identification and molecular characterization of six novel mutations in the UDP-N-acetylglucosamine-1-phototransferase gamma subunit (GNPTG) gene in patients with mucolipidosis III gamma. *Human Mutat* 30, 978-84.
11. Parnetti L, Balducci C, Pierguidi L, De Carlo C, Peducci M, D'Amore C, Padiglioni C, Mastrocoda S, Persichetti E, Paciotti S, Bellomo G, Tambasco N, Rossi A, Beccari T, Calabresi P (2009). Cerebrospinal fluid beta-glucocerebrosidase activity is reduced in Dementia with Lewy Bodies. *Neurobiol Dis* 34, 484-6.
12. Bennati AM, Schiavoni G, Franken S, Piobbico D, Della Fazia MA, Caruso D, De Fabiani E, Benedetti L, Cusella De Angelis MG, Gieselmann V, Servillo G, Beccari T, Roberti R (2008). Disruption of the gene encoding 3beta-hydroxysterol Delta-reductase (Tm7sf2) in mice does not impair cholesterol biosynthesis. *FEBS J* 275, 5034-47.
13. Riise Stensland HM, Persichetti E, Sorriso C, Hansen GM, Bibi L, Paciotti S, Balducci C, Beccari T (2008). Identification of two novel beta-mannosidosis-associated sequence variants: biochemical analysis of beta-mannosidase (MANBA) missense mutations. *Mol Genet Metab* 94, 476-80.
14. Balducci C, Bibi L, Berg T, Persichetti E, Tiribuzi R, Martino S, Paciotti S, Roberti R, Orlacchio A, Beccari T (2008). Molecular cloning and structural organization of the gene encoding the mouse lysosomal di-N-acetylchitobiase (ctbs). *Gene* 416, 85-91.
15. Balducci C, Pierguidi L, Persichetti E, Parnetti L, Sbaragli M, Tassi C, Orlacchio A, Calabresi P, Beccari T, Rossi A (2007). Lysosomal hydrolases in cerebrospinal fluid from subjects with Parkinson's disease. *Movement Disorders* 22, 1481-1484.

16. Pittis MG, Montalvo ALE, Heikinheimo P, Sbaragli M, Balducci C, Persichetti E; Van Maldergem L, Filocamo M, Bembi B, Beccari T (2007). Functional characterization of four novel MAN2B1 mutations causing juvenile onset alpha-mannosidosis. *Clin Chim Acta* 375, 136-139.
17. Bennati AM, Castelli M, Della Fazia MA, Beccari T, Caruso D, Servillo G, Roberti R (2006). Sterol dependent regulation of human TMSF2 gene expression: role of the encoded 3beta-hydroxysterol delta 14-reductase in human cholesterol biosynthesis. *Biochim Biophys Acta* 1761, 677-685.
18. Tassi C, Angelini A, Beccari T, Capodicasa E (2006). Fluorimetric determination of activity and isoenzyme composition of N-acetyl-beta-D-hexosaminidase in seminal plasma of fertile and infertile patients with secretory azoospermia. *Clin Chem Lab Med* 44, 843-847.
19. Costanzi E, Balducci C, Cacan R, Duvet S, Orlacchio A, Beccari T (2006). Cloning and expression of mouse cytosolic alpha-mannosidase (MAN2C1). *Biochim Biophys Acta* 1760, 1580-1586.
20. Sbaragli M, Bibi L, Pittis MG, Balducci C, Heikinheimo P, Ricci R, Antuzzi D, Parini R, Spaccini L, Bembi B, Beccari T (2005). Identification and characterization of five novel MAN2B1 mutations in Italian patients with alpha-mannosidosis. *Hum Mut* 25, 320-324.
21. Roces DP, Lullmann-Rauch R, Peng J, Balducci C, Andress C, Tollersrud OK, Fogh J, Orlacchio A, Beccari T, Saftig P, von Figura K (2004). Efficacy of enzyme therapy in alpha-mannosidosis mice. A preclinical study. *Hum Mol Genet* 13, 1979-1988.