

VAN SCHAFTINGEN, Emile

Born in 1953 in Wasmès (Belgium)

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Degrees

- 1978 M.D., Catholic University of Louvain Medical School
- 1985 Agrégé de l'Enseignement Supérieur, (corresponding to a Ph.D. degree) Catholic University of Louvain

Current position

Emeritus Professor of Biochemistry, Faculty of Medicine, Université Catholique de Louvain.
Director, de Duve Institute,

Awards and honours

- Merck Sharp & Dohme Award for Belgium (1987)
- Minkowski Award of the European Association for the Study of Diabetes (1992)
- Member of the Royal Academy of Medicine of Belgium (1998)
- Van Gysel Award for Biomedical Research (2003)
- Chaire Francqui FUNDP (2011-2012)

Main Scientific achievements

Regulation of intermediary metabolism · Discovery of fructose 2,6-bisphosphate (with L. Hue and H.G. Hers), of its pathways of synthesis and degradation, and of its major function as a glycolytic regulator. This is presented in all major biochemistry textbooks. · Discovery of the regulatory protein of glucokinase (GCKR) and its central role in the regulation of hepatic glycolysis.

Metabolite and protein repair · Discovery of 8 metabolite repair enzymes (representing about half of these enzymes known to date). · Discovery of the first example of a disease caused by a deficiency in metabolite repair · Discovery of protein deglycation (by fructosamine-3-kinase), a new protein repair mechanism.

Metabolic diseases and enzymology · Identification of the molecular defect in 10 new metabolic diseases (in collaboration with J. Jaeken and G. Matthijs) including the most common congenital disorder of glycosylation (CDG1a, PMM2 deficiency). · Discovery of ≈ 21 enzymes catalyzing 'new' enzymatic reactions.

Track records

- ≈ 240 Peer-Reviewed publications listed in PUBMED
- Scopus citations: ≈13127 Google Scholar citations: ≈19260

Selected publications in the last 15 years

Hart CE, Race V, Achouri Y, Wiame E, Sharrard M, Olpin SE, Watkinson J, Bonham JR, Jaeken J, Matthijs G, Van Schaftingen E. Phosphoserine aminotransferase deficiency: a novel disorder of the serine biosynthesis pathway. *Am J Hum Genet.*80(5):931-7 (2007)

Wiame E, Tyteca D, Pierrot N, Collard F, Amyere M, Noel G, Desmedt J, Nassogne MC, Vikkula M, Octave JN, Vincent MF, Courtoy PJ, Boltshauser E, van Schaftingen E. Molecular identification of aspartate N-acetyltransferase and its mutation in hypoacetylaspartia. *Biochem J.* 425(1):127-36 (2009)

Veiga-da-Cunha, M., Chevalier, N., Stroobant, V., Vertommen, D. & Van Schaftingen, E. Metabolite proofreading in carnosine and homocarnosine synthesis: molecular identification of PM20D2 as betaalanyl-lysine dipeptidase. *J Biol Chem* 289, 19726-19736 (2014)

Marbaix, A.Y., Noel, G., Detroux, A.M., Vertommen, D., Van Schaftingen, E. & Linster, C.L. Extremely conserved ATP- or ADP-dependent enzymatic system for nicotinamide nucleotide repair. *J Biol Chem* 286, 41246-41252 (2011)

Linster, C.L., Noel, G., Stroobant, V., Vertommen, D., Vincent, M.F., Bommer, G.T., Veiga-da-Cunha, M. & Van Schaftingen, E. Ethylmalonyl-CoA decarboxylase, a new enzyme involved in metabolite proofreading. *J Biol Chem* 286, 42992-43003 (2011)

Linster, C.L., Van Schaftingen, E. & Hanson, A.D. Metabolite damage and its repair or preemption. *Nature chemical biology* 9, 72-80 (2013)

Barbier T, Collard F, Zúñiga-Ripa A, Moriyón I, Godard T, Becker J, Wittmann C, Van Schaftingen E,* Letesson JJ*. Erythritol feeds the pentose phosphate pathway via three new isomerases leading to Derythrose-4-phosphate in *Brucella*. *Proc Natl Acad Sci U S A.* 111:17815-20 (2014)
* equal contribution as last authors

Rzem R, Achouri Y, Marbaix E, Schakman O, Wiame E, Marie S, Gailly P, Vincent MF, Veiga-da-Cunha M, Van Schaftingen E. A mouse model of L-2-hydroxyglutaric aciduria, a disorder of metabolite repair. *PLoS One.* 10(3):e0119540 (2015)

Collard, F., Baldin, F., Gerin, I., Bolsee, J., Noel, G., Graff, J., Veiga-da-Cunha, M., Stroobant, V., Vertommen, D., Houddane, A., Rider, M.H., Linster, C.L., Van Schaftingen, E.* & Bommer, G.T.* A conserved phosphatase destroys toxic glycolytic side products in mammals and yeast. *Nature chemical biology* 12, 601-607 (2016).
* equal contribution as last authors

Peracchi, A., Veiga-da-Cunha, M., Kuhara, T., Ellens, K.W., Paczia, N., Stroobant, V., Marlaire, S., Bommer, G.T., Sun, J., Hueber, K., Linster, C.L., Cooper, A.J. & Van Schaftingen, E. NIT1 is a metabolite repair enzyme that hydrolyzes deaminated glutathione. 114:E3233-E3242 (2017)

Veiga-da-Cunha M, Chevalier N, Stephenne X, Defour JP, Paczia N, Ferster A, Achouri Y, Dewulf JP, Linster CL, Bommer GT, Van Schaftingen E. Failure to eliminate a phosphorylated glucose analog leads to neutropenia in patients with G6PT and G6PC3 deficiency. *Proc Natl Acad Sci U S A.* 116(4):1241-1250. (2019)

Wortmann SB, Van Hove JLK, Derks TGJ, Chevalier N, Knight V, Koller A, Oussoren E, Mayr JA, van Spronsen FJ, Lagler FB, Gaughan S, Van Schaftingen E, Veiga-da-Cunha M. Treating neutropenia and neutrophil dysfunction in glycogen storage disease type Ib with an SGLT2 inhibitor. *Blood.* 136(9):1033-1043 (2020)