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Author: Nurulamin Abu Bakar, Nicol C. Voermans, Thorsten Marquardt, Christian Thiel, Mirian C.H. Janssen, Hana Hansikova, Ellen Crushell, Jolanta Sykut-Cegielska, Francis Bowling, Lars Mørkrid, John Vissing, Eva Morava, Monique van Scherpenzeel, Dirk J. Lefeber

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Intact transferrin and total plasma glycoprofiling for diagnosis and therapy monitoring in phosphoglucomutase-I deficiency

Nurulamin Abu Bakar[Department of Neurology and Translational Metabolic Laboratory, Donders Institute for Brain, Cognition, and Behavior, Radboud University Medical Center, Nijmegen, The Netherlands; email: nurulamin.binabubakar@radboudumc.nl], **Nicol C. Voermans**[Department of Neurology, Radboud University Medical Center, Nijmegen, The Netherlands; email: nicol.voermans@radboudumc.nl], **Thorsten Marquardt**[University Hospital Muenster, Muenster, Germany; email: thorsten.marquardt@ukmuenster.de], **Christian Thiel**[Center for Child and Adolescent Medicine, Kinderheilkunde I, University of Heidelberg, Heidelberg, Germany; email: christian.thiel@med.uni-heidelberg.de], **Mirian C.H. Janssen**[Department of Internal Medicine, Radboud University Medical Center, Nijmegen, The Netherlands; email: mirian.janssen@radboudumc.nl], **Hana Hansikova**[Department of Pediatrics and Adolescent Medicine, First Faculty of Medicine, Charles University in Prague and General University Hospital in Prague, Prague, Czech Republic; email: hana.hansikova@lf1.cuni.cz], **Ellen Crushell**[Academic Centre on Rare Diseases, University College Dublin, Dublin, Republic of Ireland; email: ellen.crushell@cuh.ie], **Jolanta Sykut-Cegielska**[Screening and Metabolic Diagnostics Department, The Institute of Mother and Child, Warsaw, Poland; email: j.cegielska@wp.pl], **Francis Bowling**[Biochemical Diseases, Mater Children's Hospital, South Brisbane, Queensland, Australia; email: francis.bowling@mater.org.au], **Lars Mørkrid**[Institute of Clinical Biochemistry, Faculty of Medicine, University of Oslo and Department of Medical Biochemistry, Oslo University Hospital, Norway; email: lamo2@ous-hf.no], **John Vissing**[Department of Neurology, University of Copenhagen, Denmark; email: john.vissing@regionh.dk], **Eva Morava**[Department of Clinical Genomics, CIM, Mayo Clinic, Rochester, Minnesota, United States of America; email: morava-kozicz.eva@mayo.edu], **Monique van Scherpenzeel**[Translational Metabolic Laboratory, Radboud University Medical Center, Nijmegen, The Netherlands; email: monique.vanscherpenzeel@radboudumc.nl], **Dirk J. Lefeber**[Department of Neurology and Translational Metabolic Laboratory, Donders Institute for Brain, Cognition, and Behavior, Radboud University Medical Center, Nijmegen, The Netherlands; email: dirk.lefeber@radboudumc.nl].

Corresponding author: Dr. Dirk J. Lefeber, Translational Metabolic Laboratory, Department of Neurology, Radboud University Medical Center, Box 9101, 6500 HB Nijmegen, The Netherlands. Tel: +31(0)243614567. email: dirk.lefeber@radboudumc.nl.

Abbreviations: phosphoglucomutase-1, PGM1; glucose-1-phosphate, G1P; congenital disorder of glycosylation, CDG; CDG type-1, CDG-I; CDG type-2, CDG-II; mass spectrometry, MS; transferrin isoelectric focusing, TIEF; apolipoprotein C-III, ApoC-III; isoelectric focusing; IEF; volume/volume, v/v; porous graphitized carbon chip, PGC-chip; confidence intervals, CI; normal glycan index, NGI; lack of complete glycan index, LOCGI; lack of galactose index, LOGI; endoplasmic reticulum, ER.