Professor Henk Blom, PhD, Laboratory Specialist Clinical Genetics, Center for Lysosomal and Metabolic Diseases, Department of Clinical Genetics, Erasmus University Medical Center, Rotterdam, Netherlands; Professor, Biochemistry of Inherited Metabolic Disease

Henk Blom finished his Chemistry study in 1985 and received his PhD in 1988 at the Radboud University Nijmegen. After his post-doc period at the Human Genetics Branch, NIH, USA (William Gahl), he became post-doc in 1990 and later in 1992 staff member of the Clinical Genetics Center Nijmegen at Laboratory of Pediatrics and Neurology, University Hospital Nijmegen, the Netherlands. In 1997 he became Established Investigator of the Netherlands Heart Foundation and in 2003 he was registered as Clinical Biochemical Geneticist. In 2007 he was appointed as vice-head and later head of the Metabolic Unit at the Department Clinical Chemistry, VU University Medical Centre Amsterdam, the Netherlands and in 2009 he became Professor in Biochemistry of Inherited Metabolic Diseases at the VU University Medical Centre Amsterdam. From 2014 to 2017 he was head of the laboratory for Clinical Biochemistry and Metabolism, Department of General Pediatrics, Center for Pediatrics and Adolescent Medicine University Hospital Freiburg, Germany. Since 2018 he has served as Laboratory Specialist, Clinical Genetics, Center for Lysosomal and Metabolic Diseases, Erasmus University Medical Center, Rotterdam, Netherlands.

His research concerns inborn errors of metabolism with special focus on inherited defects of homocysteine, methylation and folate metabolism. His contributions include the association of a disturbed homocysteine metabolism with pregnancy complications, including neural tube defects, cardiovascular disease, thrombosis and stroke in children. He investigated the genetic etiology of thermolabile MTHFR, which resulted in the discovery of the MTHFR 677C>T variant which is the first identified genetic risk factor for neural tube defects. Basic research concerned the effects of homocysteine and its metabolites on development of chicken embryos and endothelial function.
Among inborn errors of metabolism his group described the molecular basis of severe hyperhomocysteinemia. They also explored cystinosis and defects in the methionine methylation pathway, including methionine adenosyltransferase deficiency. They discovered two new genetic defects: one in folate metabolism: dihydrofolate reductase deficiency and one defect in methylation: adenosine kinase deficiency. Prof Henk Blom is coordinator of E-HOD, an international consortium on homocystinurias and methylation disorders. In 2017 the consortium consisted of almost 100 partners. Main achievements are the setup of the E-HOD registry (www.EHOD.registry.com) and website (www.EHOD.com) with information for expert as well as patients and their families. In addition four guideline manuscripts have been published, teaching courses and Patient - Expert Meetings organized.
He supervised as (co)promoter of 31 PhD students and published over 350 papers in international journals resulting in an H-index of 72.