

Translating the dynamics of genetics  
into health care practice

Tessel Rigter

---

The study presented in this thesis was performed at the EMGO Institute for Health and Care Research and the Department of Clinical Genetics, section Community Genetics of the VU University Medical Center, Amsterdam, the Netherlands.

The EMGO+ Institute participates in the Netherlands School of Primary Care Research (CaRe), which was re-acknowledged in 2006 by the Royal Netherlands Academy of Arts and Sciences (KNAW). In 2010, the EMGO+ Institute received an excellent review by the international external evaluation committee of all Dutch university research, as organized by the universities in the Netherlands.

The study was funded by the Netherlands Genomics Initiative (NGI) as part of the programme of the Centre for Society and the Life Sciences and the Centre for Systems Biology in the Netherlands (Project no. 70.1.053.2h1), the European Commission as part of the EuroGentest2 Coordination Action 2011 project (Unit2, WP8, Contract no. HEALTH-F4-2010-261469) and Top Institute Pharma (project T6-208) with financial support of the Dutch Health Care Insurance Board (College voor zorgverzekeringen), the Dutch Steering Committee on Orphan Drugs (Stuurgroep Weesgeneesmiddelen), Genzyme Corporation, Shire Corporation, Academic Medical Center (Amsterdam), Erasmus MC University Medical Center (Rotterdam) and Utrecht University Medical Center (Utrecht)

The author received travel grants from the Dittmerfonds of the VU University, Amsterdam, the Netherlands and Simonsfonds at the Erasmus Medical Center, Rotterdam, the Netherlands and prof. Ulf Kristoffersson, Lund University, Lund, Sweden.

Financial support for publication of this thesis was kindly provided by the EMGO+ Institute, An and Ernie Enders and Paul Antkowiak.

Translating the dynamics of genetics into health care practice

Thesis, VU medisch centrum – Vrije Universiteit

ISBN: 978-94-6182-477-6

©Copyright: Tessel Rigter

All rights reserved. No part of this thesis may be reproduced or transmitted in any form or by any means, electronically or mechanically, including photocopy, recording or any other information storage or retrieval system, without permission in writing from the author.

---

# PART I

NEXT GENERATION SEQUENCING  
IN DIAGNOSTICS: RECENT IMPLEMENTATION  
OF A NEW OPPORTUNITY IN CLINICAL GENETICS

# PART II

NEONATAL SCREENING FOR POMPE DISEASE:  
A POTENTIAL OPPORTUNITY FOR PUBLIC HEALTH

# PART III

TESTING FOR MONOGENIC  
SUBTYPES OF COMMON DISORDERS:  
ONGOING IMPLEMENTATION OF NEW GENETIC  
SERVICES IN "MAINSTREAM MEDICINE"

# PART IV

GENERAL DISCUSSION & SUMMARY