## Translating the dynamics of genetics into health care practice

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# PUBLICATIONS

*Scientific publications*

Van El CG, **Rigter T**, Reuser AJJ, van der Ploeg AT, Weinreich SS, Cornel MC. Newborn screening for Pompe disease? A qualitative study exploring professional views. BMC Pediatrics 2014, 14:203.

**Rigter T**, Henneman L, Broerse JEW, Shepherd MH, Blanco IG, Kristoffersson U, Cornel MC. Developing a framework for implementation of genetic services: learning from examples of testing for monogenic forms of common diseases. Journal of Community Genetics 2014, June 14. Epub ahead of print. DOI 10.1007/s12687-014-0189-x

Weinreich SS, Bosma AR, Henneman L, **Rigter T**, Spruijt CMJ, Grimbergen AJEMA, Breuning MH, de Koning EJP, Losekoot M, Cornel MC. A decade of molecular genetic testing for MODY: retrospective study of utilisation in the Netherlands. European Journal of Human Genetics 2014, April 16. Epub ahead of print. DOI 10.1038/ejhg.2014.59

**Rigter T**, van Aart CJA, Elting MW, Waisfisz Q, Cornel MC, Henneman L. Informed consent for exome sequencing in diagnostics: exploring first experiences and views of professionals and patients. Clinical Genetics 2014, 85:417-422

**Rigter T**, Henneman L, Kristoffersson U, Hall A, Yntema HG, Borry PM, Tönnies H, Waisfisz Q, Elting MW, Dondorp WJ, Cornel MC. Reflecting on earlier experiences with unsolicited findings: Points to consider for next generation sequencing and informed consent in diagnostics. Human Mutation 2013, 34(10): 1322-1328

Cornel MC, **Rigter T**, Weinreich SS, Burgard P, Hoffman GF, Lindner M, Loeber JG, Rupp K, Taruscio D, Vittozzi L. A framework to start the debate on neonatal screening policies in the EU: and expert opinion document. European Journal of Human Genetics 2014, 22:12-17.

Burgard P, Rupp K, Lindner M, Haege G, **Rigter T**, Weinreich SS, Loeber JG, Taruscio D, Vittozzi L, Cornel MC, Hoffmann GF. Newborn screening programmes in Europe; arguments and efforts regarding harmonization. Part 2 - From Screening laboratory results to treatment, follow-up and quality assurance Journal of Inherited Metabolic Disease 2012, 35(4): 613-25

Loeber JG, Burgard P, Cornel MC, **Rigter T**, Weinreich SS, Rupp K, Hoffmann GF, Vittozzi L. Newborn screening programmes in Europe; arguments and efforts regarding harmonization. Part 1 - From blood spot to screening result. Journal of Inherited Metabolic Disease 2012, 35(4): 603-11

**Rigter T**, Weinreich SS, van EL CG, de Vries JM, van Gelder CM, Güngör D, Reuser AJ, Hagemans ML, Cornel MC, van der Ploeg AT. Severely impaired health status at diagnosis of Pompe disease: A cross-sectional analysis to explore the potential utility of neonatal screening. Molecular Genetics and Metabolism 2012, 107(3):448-55

Weinreich SS, **Rigter T**, van El CG, Dondorp WJ, Kostense P, van der Ploeg AT, Reuser AJJ, Cornel MC, Hagemans MLC. Public support for neonatal screening for Pompe disease, a broad-phenotype condition. Orphanet Journal of Rare Diseases 2012, 7:15

*Published abstracts and presentations*

**Rigter T**, Bosma AR, Weinreich SS, Henneam L, de Koning EJP, Breuning MH, Losekoot M, Cornel MC. Towards best practice guidelines for genetic testing for Maturity-Onset Diabetes of the Young. **Oral presentation.** November 28-29 2013 Oosterbeek, the Netherlands. Annual Dutch Diabetes Research Meeting.

**Rigter T**, Bosma AR, Weinreich SS, Henneam L, de Koning EJP, Breuning MH, Losekoot M, Cornel MC. Towards best practice guidelines for genetic testing for Maturity-Onset Diabetes of the Young. **Oral presentation.** November 28 2013 Utrecht, the Netherlands. Autumn meeting Dutch Association of Community Genetics and Public Health Genomics (NACGG).

**Rigter T,** Henneman L, Kristoffersson U, Hall A, Yntema HG, Borry PM, Tönnies H, Waisfisz Q, Elting MW, Dondorp WJ, Cornel MC. Reflecting on earlier experiences with unsolicited findings: Points to consider for next generation sequencing and informed consent in diagnostics. **Poster and oral presentation.** Received Poster Award for a presentation judged outstanding. June 8-11 2013 Paris, France. European Society of Human Genetics

Weinreich S.S., Bosma AR, Henneman L, **Rigter T**, Breuning MH, de Koning EJP, Losekoot M, Cornel MC. Genetic testing for MODY: retrospective study of utilization in the Netherlands. **Poster presentation.** June 8-11 2013 Paris, France. European Society of Human Genetics.

**Rigter T.** Reflecting on earlier experiences with unsolicited findings: Points to consider for next generation sequencing and informed consent in diagnostics. **Video highlight** website Human Mutation: www.youtube.com/watch?v=gh4qc0Ucy3Y .

**Rigter T**. European initiatives for unified informed consent. **Oral presentation.** January 13-14 2013 Tel Aviv, Israel. E-Rare Workshop: Ethical aspects of exome and whole genome sequencing studies (WES/WGS) in rare diseases.

**Rigter T**. Exploring needs for the informed consent procedure for genome sequencing in diagnostics. **Oral presentation** (presented by Henneman L due to unforeseen circumstances). March 7-8 2013 Prague, Czech Republic. 3rd International Scientific Symposium EuroGentest.

**Rigter T**, van Aart CJA, Elting MW, Waisfisz Q, Cornel MC, Henneman L. Exome sequencing in diagnostics: exploring needs for the informed consent procedure. **Oral presentation.** September 21-22 2012 Arnhem, the Netherlands. Symposium Dutch Society of Human Genetics (NVHG).

**Rigter T**, Henneman L, Kristoffersson U, Cornel MC. Developing best practice guidelines for provision of clinical genetic service: Examples of testing for monogenic subtypes. **Poster presentation**. June 23-26 2012 Nurnberg, Germany. European Society of Human Genetics.

Van El CG, **Rigter T**, Weinreich SS, Reuser AJJ, van der Ploeg AT, Cornel MC. Newborn screening for Pompe disease? Exploring professional views. **Poster presentation**. May 28-31 2011 Amsterdam, the Netherlands. European Society of Human Genetics.

Weinreich SS, Hagemans MLC, **Rigter T**, van El CG, Cornel MC, van der Ploeg AT, Reuser AJJ. Should newborn screening be introduced for Pompe disease? Preliminary report on valuation of benefits and risks by the general public in the Netherlands. **Poster presentation**. June 12-15 2010 Gothenburg, Sweden. European Society of Human Genetics.

**Rigter T**, Weinreich SS, van El CG, Cornel MC, Reuser AJJ, van der Ploeg AT, Hagemans MLC. Health and functional status of patients symptomatically diagnosed with Pompe disease. A case for earlier diagnosis through newborn screening? **Poster presentation**. June 12-15 2010 Gothenburg, Sweden. European Society of Human Genetics.