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**Overview of Publications on Population-based Genetic Carrier Screening**  
**Research Group Community Genetics**  
**VU University Medical Center & Academic Medical Center, Amsterdam, the Netherlands**

**Cystic fibrosis carrier screening**

Henneman L, Bramsen I, Van der Ploeg HM, Adèr HJ, Van der Horst HE, Gille JJP, Ten Kate LP. Participation in preconceptional carrier couple screening: characteristics, attitudes, and knowledge of both partners. *JMG* 2001;38:695-703.

Henneman L, Bramsen I, Van Kempen L, Van Acker MB, Pals G, Van der Horst HE, Adèr HJ, Van der Ploeg HM, Ten Kate LP. Offering preconceptional cystic fibrosis carrier couple screening in the absence of established preconceptional care services. *Community Genetics* 2003;6:5-13.

Henneman L, Bramsen I, Van der Ploeg HM, Ten Kate LP. Preconceptional cystic fibrosis carrier couple screening: impact, understanding and satisfaction. *Genetic Testing* 2002;6:195-202.

Henneman L, Kooij L, Bouman K, Ten Kate LP. Personal experiences of cystic fibrosis (CF) carrier couples prospectively identified in CF families. *American Journal of Medical Genetics* 2002;110:324-331.

Henneman L, Bramsen I, Van Os ThAM, Reuling IEW, Heyerman HGM, Van der Laag J, Van der Ploeg HM, Ten Kate LP. Attitudes towards reproductive issues and carrier testing among adult patients and parents of children with cystic fibrosis (CF). *Prenatal Diagnosis* 2001;21:1-9.

Henneman L, Ten Kate LP. Preconceptional couple screening for cystic fibrosis carrier status: couples prefer full disclosure of test-results. *Journal of Medical Genetics* 2002;39:e26.

Henneman L, Poppelaars FAM, Kate LP ten. Wenselijkheid en haalbaarheid van preconceptionele screening op dragerschap voor cystische fibrose. *Nederlands Tijdschrift voor Geneeskunde*. 2004; 148: 618-622. [in Dutch]

Lakeman P, Gille JJ, Dankert-Roelse JE, Heijerman HG, Munck A, Iron A, Grasemann H, Schuster A, Cornel MC, Ten Kate LP. CFTR mutations in Turkish and North African cystic fibrosis patients in Europe: implications for screening. *Genet Test* 2008;12:25-35.

Poppelaars FAM, Henneman L, Adèr HJ, Cornel MC, Hermens RPMG, Van der Wal G, Ten Kate LP. Preconceptional cystic fibrosis carrier screening: attitudes and intentions of the target population. *Genetic Testing* 2004;8:80-86.

Poppelaars FAM, Van der Wal G, Braspenning JCC, Cornel MC, Henneman L, Langendam MW, Ten Kate LP. Possibilities and barriers in the implementation of a preconceptional screening programme for cystic fibrosis carriers: a focus group study. *Public Health* 2003;117:396-403.

Poppelaars FAM, Henneman L, Adèr HJ, Cornel MC, Hermens RPMG, Van der Wal G, Ten Kate LP. How should preconceptional cystic fibrosis carrier screening be provided? Opinions of potential providers and the target population. *Community Genet* 2003;6:157-165.



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## Hemoglobinopathies carrier screening

Jans SMPJ, Jonge A de, Henneman L, Cornel MC, Lagro-Janssen ALM. Attitudes of general practitioners and midwives towards ethnicity-based haemoglobinopathy carrier screening. *Eur J Hum Genet* 2012;20:1112-1117.

Jans SMPJ, Annegarn A, Legdeur MA, Henneman L, Lakeman P. Screenen op dragerschap van erfelijke aandoeningen tijdens het kindwens spreekuur. Pilot project in Amsterdam. *Tijdschrift voor Verloskundigen* 2010;46-48.

Lakeman P, Henneman L, Bezemer PD, Cornel MC, Ten Kate LP. Developing and optimizing a decisional instrument using self-reported ancestry for carrier screening in a multi-ethnic society. *Genet Med* 2006;8:502-9.

Lakeman P, Plass AM, Henneman L, Bezemer PD, Cornel MC, Ten Kate LP. Three-month follow-up of Western and non-Western participants in a study on preconceptional ancestry-based carrier couple screening for cystic fibrosis and hemoglobinopathies in the Netherlands. *Genet Med* 2008;10:820-30.

Lakeman P, Plass AM, Henneman L, Bezemer PD, Cornel MC, Ten Kate LP. Preconceptional ancestry-based carrier couple screening for cystic fibrosis and haemoglobinopathies: what determines the intention to participate or not and actual participation? *Eur J Hum Genet* 2009;17:999-1009.

Weinreich SS, de Lange-de Klerk ES, Rijmen F, Cornel MC, de Kinderen M, Plass AM. Raising awareness of carrier testing for hereditary haemoglobinopathies in high-risk ethnic groups in the Netherlands: a pilot study among the general public and primary care providers. *BMC Public Health*. 2009;9:338.

## Carrier screening in a Dutch founder population

Mathijssen IB, Henneman L, Van Eeten-Nijman JMC, Lakeman P, Ottenheim CPE, Redeker EJW, Ottenhof W, Meijers-Heijboer H, Van Maarle MC. Targeted carrier screening for four recessive disorders: High detection rate within a founder population. *Eur J Med Genet*. 2015;58:123-128

## Carrier screening in the Jewish population

Holtkamp KCA, Van Maarle MC, Schouten M, Dondorp WJ, Lakeman P, Henneman L. Do people from the Jewish community prefer ancestry-based or pan-ethnic expanded carrier screening? *Eur J Hum Genet*. 2016;24:171-7.

## Consanguinity

Kelmami W, Teeuw ME, Bochdanovits Z, Ouburg S, Jonker MA, Alkuraya F, Hashem M, Kayserili H, van Haeringen A, Sheridan E, Masri A, Cobben JM, Rizzu P, Kostense PJ, Dommering CJ, Henneman L, Bouhamed-Chaabouni H, Heutink P, Ten Kate LP, Cornel MC. Determining the genome-wide kinship coefficient seems unhelpful in distinguishing consanguineous couples with a high versus low risk for adverse reproductive outcome. *BMC Med Genet*. 2015; 16: 50.



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Teeuw ME, Henneman L, Bochdanovits Z, Heutink P, Kuik DJ, Cornel MC, Ten Kate LP. Do consanguineous parents of a child affected by an autosomal recessive disease have more DNA identical-by-descent than similarly-related parents with healthy offspring? Design of a case-control study. *BMC Medical Genetics* 2010;11:113.

Teeuw ME, Loukili G, Bartels EA, Ten Kate LP, Cornel MC, Henneman L. Consanguineous marriage and reproductive risk: attitudes and understanding of ethnic groups practising consanguinity in Western society. *Eur J Hum Genet.* 2014; 22: 452-57.

Teeuw ME, Waisfisz Q, Zwijnenburg PJG, Sistermans EA, Weiss M, Henneman L, Ten Kate LP, Cornel MC, Meijers-Heijboer EJ. First steps in exploring prospective exome sequencing of consanguineous couples. *European Journal of Medical Genetics* 2014;57:613-6.

Teeuw ME, Borry P, Ten Kate LP, Preconception care for consanguineous couples in the Netherlands, in "Cousin marriages: Between tradition, genetic risk and cultural change". Edited by A Shaw and A Raz. Berghahn Books: Oxford and New York. 2015 ISBN 978-1-78238-492-2.

Ten Kate LP, Teeuw ME, Henneman L, Cornel MC. Consanguinity and Endogamy in the Netherlands: Demographic and Medical Genetic Aspects. *Human Heredity* 2014;77:161-166.

Ten Kate LP, Teeuw M, Henneman L, Cornel MC. Autosomal recessive disease in children of consanguineous parents: inferences from the proportion of compound heterozygotes. *J Community Genet.* 2010;1:37-40.

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## Health care professionals attitudes and knowledge

Baars MJH, Henneman L, Kate LP ten. Preconceptional Cystic Fibrosis Carrier Screening: Opinions of General Practitioners, Gynecologists and Pediatricians in The Netherlands. *Genet Testing* 2004;8:431-436.

Baars MJH, Henneman L, Kate LP ten. Deficiency of knowledge of genetics and genetic tests among general practitioners, gynaecologists and paediatricians; A global problem. *Genet Med* 2005;7:605-610.

Jans SMPJ, Jonge A de, Henneman L, Cornel MC, Lagro-Janssen ALM. Attitudes of general practitioners and midwives towards ethnicity-based haemoglobinopathy carrier screening. *Eur J Hum Genet* 2012;20:1112-1117.

Jans SMPJ, Henneman L, de Jonge A, van El CG, van Tuyl L, Cornel MC, Lagro-Janssen ALM. "A morass of considerations": Exploring attitudes towards primary care ethnicity-based haemoglobinopathy screening. *Fam Pract* 2013;30:604-10.

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Teeuw ME, Hagelaar A, Ten Kate LP, Cornel MC, Henneman L. Challenges in the care for consanguineous couples: an exploratory interview study among general practitioners and midwives. *BMC Fam Pract* 2012;13:105.

## Screening methods

Ten Kate LP, Verheij JB, Wildhagen MF, Hilderink HB, Kooij L, Verzijl JG, Habbema JD. Comparison of single-entry and double-entry two-step couple screening for cystic fibrosis carriers. *Hum Hered* 1996;46:20-5.

## Policy

Achterbergh R, Lakeman P, Stermerding D, Moors EH, Cornel MC. Implementation of preconceptional carrier screening for cystic fibrosis and haemoglobinopathies: a sociotechnical analysis. *Health Policy* 2007;83:277-86.

Bolhuis PA, Henneman L. Screening bij kinderwens komt eraan. *Medisch Contact* 2014;39:1884-6. [in Dutch]

Borry P, Henneman L, Lakeman P, Ten Kate LP, Cornel MC, Howard H. Preconceptional genetic carrier testing and the commercial offer directly-to-consumers. *Human Reprod* 2011;26:972-977.

Cornel MC, Lakeman P, Dondorp W. [Preconception carrier screening should not be delayed] Stel preconceptionele dragerschapsscreening niet uit. *Ned Tijdschr Geneesk*. 2011;155:A3205. [in Dutch]

Grosse SD, Rogowski WH, Ross LF, Cornel MC, Dondorp WJ, Khoury MJ. Population Screening for Genetic Disorders in the 21st Century: Evidence, Economics, and Ethics. *Public Health Genomics* 2010;13:106-115.

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Henneman L, Cornel MC. Genetic Carrier Testing. *Encyclopedia of Life Sciences*. John Wiley & Sons, Ltd: Chichester.

Jans SM, van El CG, Houwaart ES, Westerman MJ, Janssens RJ, Lagro-Janssen AL, Plass AM, Cornel MC. A case study of haemoglobinopathy screening in the Netherlands: witnessing the past, lessons for the future. *Ethn Health*. 2012;17:217-39.

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Van der Laar J, Ten Kate LP. Preconceptional screening for carriership of cystic fibrosis; testing against the criteria of the Health Council for genetic screening [in Dutch]. *Ned Tijdschr Geneesk* 1996;140:487-91.

Weijers-Poppelaars FAM, Henneman L, Cornel MC, Kate LP ten. Preconceptionele screening op dragerschap voor cystische fibrose: Implementatiemogelijkheden en barrières. *Huisarts en Wetenschap* 2005;48:395-98. [in Dutch]

Weijers-Poppelaars FAM, Wildhagen MF, Henneman L, Cornel MC, Ten Kate LP. Preconception cystic fibrosis carrier screening: costs and consequences. *Genet Testing* 2005;9:158-66.



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## Thesis

L. Henneman. Preconceptional cystic fibrosis carrier screening - desirability and feasibility in the Netherlands. Promotoren: L.P. ten Kate, H.M. van der Ploeg. VU University 2002.

F.A.M. Weijers-Poppelaars. Preconceptional cystic fibrosis carrier screening - opportunities for implementation. Promotoren: L.P. ten Kate, G. van der Wal; Co-promotor: L. Henneman. VU University 2004.

P. Lakeman. Preconceptional screening for carriers of haemoglobinopathies and/or cystic fibrosis, dependent on ethnic background. Feasibility of a combined offer in the Dutch Society. Promotoren: L.P. ten Kate, M.C. Cornel; Co-promotor: L. Henneman. VU University 2008.

S.M.J.P. Jans. Screening for anaemia and haemoglobinopathy before and during pregnancy. Promotoren: A.L.M. Lagro-Janssen, M.C. Cornel; Co-promotoren: A. de Jonge, L. Henneman. Radboud University 2012.