



Universiteit  
Leiden  
The Netherlands

## The genetic etiology of familial breast cancer: Assessing the role of rare genetic variation using next generation sequencing

Hilbers, F.S.M.

### Citation

Hilbers, F. S. M. (2020, July 7). *The genetic etiology of familial breast cancer: Assessing the role of rare genetic variation using next generation sequencing*. Retrieved from <https://hdl.handle.net/1887/123226>

Version: Publisher's Version

License: [Licence agreement concerning inclusion of doctoral thesis in the Institutional Repository of the University of Leiden](#)

Downloaded from: <https://hdl.handle.net/1887/123226>

**Note:** To cite this publication please use the final published version (if applicable).

Cover Page



Universiteit Leiden



The handle <http://hdl.handle.net/1887/123226> holds various files of this Leiden University dissertation.

**Author:** Hilbers, F.S.M.

**Title:** The genetic etiology of familial breast cancer: Assessing the role of rare genetic variation using next generation sequencing

**Issue Date:** 2020-07-07

# **The genetic etiology of familial breast cancer**

Assessing the role of rare genetic variation using next generation sequencing

Florentine Hilbers

Cover & layout: Florentine Hilbers  
Printing: Print Service Ede - [www.proefschriftenprinten.nl](http://www.proefschriftenprinten.nl)

The work presented in this thesis was financially supported by the Dutch Cancer Society.

© 2020 Florentine Hilbers  
ISBN: 978-90-830704-2-1

# **The genetic etiology of familial breast cancer**

Assessing the role of rare genetic variation using next generation sequencing

Proefschrift

ter verkrijging van  
de graad van Doctor aan de Universiteit Leiden,  
op gezag van Rector Magnificus prof.mr. C.J.J.M. Stolkers,  
volgens besluit van het College voor Promoties  
te verdedigen op dinsdag 7 juli 2020  
klokke 13:45 uur

door  
Florentine Susanna Maria Hilbers  
Geboren te Wilnis in 1986

**Promotoren**

Prof. dr. P. Devilee

Prof. dr. C.J. van Asperen

**Leden promotiecommissie**

Prof. dr. H. van Attikum

Prof. dr. J. Morreau

Dr. A.R. Mensenkamp, Radboud Uiverstitair Medisch Centrum

Dr. M.A. Rookus, Nederlands Kanker Instituut

## Table of Contents

<b>Chapter 1</b>	General Introduction	7
<b>Chapter 2</b>	Exome sequencing of non-BRCA1/2 hereditary breast cancer: no genetic evidence for a subgroup defined by aCGH profiling <i>PLoS ONE</i> 2013; 8, e55734	27
<b>Chapter 3</b>	Rare variants in XRCC2 as breast cancer susceptibility alleles <i>J Med Genet.</i> 2012 Oct; 49(10): 618-20	45
<b>Chapter 4</b>	Functional analysis of missense variants in the putative breast cancer susceptibility gene XRCC2 <i>Hum Mutat.</i> 2016 Sep; 37(9): 914-25	55
<b>Chapter 5</b>	Clustering of known low and moderate risk alleles rather than a novel recessive high risk gene in non-BRCA1/2 sib trios affected with breast cancer <i>Int J Cancer. In Press</i>	87
<b>Chapter 6</b>	The impact of next generation sequencing on the analysis of breast cancer susceptibility: a role for extremely rare genetic variation? <i>Clin Genet.</i> 2013 Nov; 84(5): 407-14	115
<b>Chapter 7</b>	Summary and discussion	131
<b>Appendix</b>	Nederlandse Samenvatting	147
	Dankwoord	157
	Curriculum vitae	158
	Publicatielijst	159

