

Cover Page



Universiteit Leiden



The following handle holds various files of this Leiden University dissertation:
<http://hdl.handle.net/1887/81575>

Author: Tuin, K. van der

Title: Joining forces in endocrine cancer genetics: molecular testing, surveillance and treatment decision making in clinical practice

Issue Date: 2019-12-12

JOINING FORCES IN ENDOCRINE CANCER GENETICS

**Molecular testing, surveillance and
treatment decision making
in clinical practice**

Karin van der Tuin

ISBN: 978-94-6182-982-5

Cover design: K. van der Tuin and Off Page

Layout and printing: Off Page, Amsterdam

The research presented in this thesis was performed at the Departments of Clinical Genetics and Pathology of Leiden University Medical Center, and was (partly) financially supported by the Children Cancer-free Foundation (KiKa, project number 265) and the Archer® International Research Challenge Grant

© 2019 Karin van der Tuin

All rights reserved. No part of this thesis may be reprinted, reproduced or utilized in any form or by any electronic, mechanical, or other means, now known or hereafter invented, including photocopying and recording of any information storage or retrieval system, without prior written permission of the author.

JOINING FORCES IN ENDOCRINE CANCER GENETICS
Molecular testing, surveillance and treatment decision making
in clinical practice

Proefschrift

ter verkrijging van
de graad van Doctor aan de Universiteit Leiden,
op gezag van Rector Magnificus prof.mr. C.J.J.M. Stolker,
volgens besluit van het College voor Promoties
te verdedigen op donderdag 12 december 2019
klokke 16.15 uur

door

Karin van der Tuin

geboren te Groningen
in 1987

Promotores

Prof. Dr. H. Morreau

Prof. Dr. T.P. Links, Universitair Medisch Centrum Groningen

Prof. Dr. F.J. Hes, Universitair Ziekenhuis Brussel, België

Leden promotiecommissie

Prof. Dr. A.M. Pereira

Prof. Dr. J.W.A. Smit, Radboud Universitair Medisch Centrum, Nijmegen

Prof. Dr. F.J. van Kemenade, Erasmus Medisch Centrum, Rotterdam

Dr. M.C.J. Jongmans, Universitair Medisch Centrum Utrecht

TABLE OF CONTENTS

Chapter 1	General Introduction	7
Part I	The role of molecular testing in endocrine cancer diagnostics and treatment decision making	27
Chapter 2	Clinical and Molecular Characteristics May Alter Treatment Strategies of Thyroid Malignancies in DICER1-syndrome	29
Chapter 3	Targetable Gene Fusions Identified in Radioactive Iodine-Refractory Thyroid Carcinoma	53
Part II	Identification of genetic predisposition in pediatric non-medullary thyroid carcinoma	65
Chapter 4	Germline Mutations in Predisposition Genes in Pediatric Non-Medullary Thyroid Cancer	67
Part III	Genetic counseling in endocrine tumor predisposition syndromes	81
Chapter 5	<i>CDC73</i> -Related Disorders: Clinical Manifestations and Case Detection in Primary Hyperparathyroidism	83
Chapter 6	Clinical Aspects of <i>SDHA</i> -Related Pheochromocytoma and Paraganglioma: A Nationwide Study	99
Chapter 7	A 93-year-old <i>MEN2a</i> Mutation Carrier Without Medullary Thyroid Carcinoma: A Case Report and Overview of the Literature	115
Part IV	General discussion	123
Chapter 8	Discussion and Future Perspectives	125
Chapter 9	Nederlandse Samenvatting	143
Appendix	List of Publications	156
	Authors and Affiliations	157
	Genetic Glossary	160
	List of Abbreviations	165
	About the Author	167
	PhD Portfolio	168
	Dankwoord	171

