CONTENTS

Chapter 1 General introduction 9

Chapter 2 Genetic counselling and testing in dementia and related disorders
   2.1 Genetics of dementia: update and guidelines for the clinician 25
   2.2 The effect of predictive testing in adult onset neurodegenerative diseases on social and personal life 51

Chapter 3 Prevalence and phenotypes of inherited dementia
   3.1 The clinical and pathological phenotype of C9orf72 hexanucleotide repeat expansions 67
   3.2 Mutation frequency of PRKAR1B and the major familial dementia genes in a Dutch early onset dementia cohort 95

Chapter 4 Identification of new genetic causes of cognitive decline
   4.1 A novel CCM2 variant in a family with non-progressive cognitive complaints and cerebral microbleeds 111
   4.2 Rare genetic variant in SORL1 may increase penetrance of Alzheimer’s disease in a family with several generations of APOE-ε4 homozygosity 125

Chapter 5 Summary 145

Chapter 6 General discussion 151

Addendum
   Nederlandse samenvatting (Dutch summary) 163
   List of publications 169
   Dankwoord (Acknowledgements) 171
   About the author 174