

Contents

Chapter 1	General introduction and aims of this thesis	7
Chapter 2	Megalencephalic leukoencephalopathy with subcortical cysts: defect in chloride currents and cell volume regulation	31
Chapter 3	Mutant GlialCAM causes leukoencephalopathy with subcortical cysts, benign familiar macrocephaly, and macrocephaly with retardation and autism	57
Chapter 4	The astrocytic MLC1 protein is differentially expressed during development in humans compared to mice	83
Chapter 5	Summary, Discussion and Future challenges	99
	Samenvatting, discussie en uitdagingen voor de toekomst	115
	List of abbreviations	133
	List of publications	135
	About the author	136
	Acknowledgements	137