

Table of contents

Abstract.....	1
Zusammenfassung	3
General Introduction	5
I. Sphingolipid <i>de novo</i> synthesis and degradation pathway	5
II. Key enzymes involved in ceramide <i>de novo</i> synthesis	8
III. Characterization of mammalian SPT	10
Chapter 1:	27
Hereditary sensory and autonomic neuropathy type I (HSAN I) caused by a novel mutation in SPTLC2	
Chapter 2:	45
Novel HSAN I mutation in Serine Palmitoyltransferase resides at a putative SPTLC2 phosphorylation site that is involved in regulating amino acid substrate specificity	
Chapter 3:	75
Deoxy-sphingolipids are generated in monocytes during PMA stimulated differentiation into macrophages	
General Conclusions	97
Outlook.....	105
Publications	109
Acknowledgements	111
Curriculum Vitae	113