

CONTENTS

Scope of the thesis

Chapter I	Introduction	11
I-1	Osteogenesis imperfecta: a review with clinical examples. <i>Accepted for publication in Molecular Syndromology</i>	13
Chapter II	Dominant Osteogenesis Imperfecta	41
II-1	Complete <i>COL1A1</i> allele deletions in osteogenesis imperfecta. <i>Genet Med.</i> 2010 Nov;12(11):736-41	43
Chapter III	Recessive Osteogenesis Imperfecta	59
III-1	<i>CRTAP</i> mutations in lethal and severe osteogenesis imperfecta: the importance of combining biochemical and molecular genetic analysis. <i>Eur J Hum Genet.</i> 2009 Dec;17(12):1560-9.	61
III-2	<i>PP1B</i> mutations cause severe osteogenesis imperfecta. <i>Am J Hum Genet.</i> 2009 Oct;85(4):521-7.	77
III-3	Lethal/Severe Osteogenesis Imperfecta in a Large Family: a novel homozygous <i>LEPRE1</i> mutation and bone histological findings. <i>Pediatr Dev Pathol.</i> 2011 May-Jun; 14(3): 228-34.	93
III-4	A novel homozygous 5 bp deletion in <i>FKBP10</i> causes clinically Bruck syndrome in an Indonesian Patient. <i>Submitted</i>	105
Chapter IV	Implications for clinical and laboratory diagnosis of Osteogenesis Imperfecta	117
IV-1	Classification of Osteogenesis Imperfecta revisited. <i>Eur J Med Genet.</i> 2010 Jan-Feb;53(1):1-5	119
IV-2	EMQN Best Practice Guidelines for the Laboratory Diagnosis of Osteogenesis Imperfecta. <i>Eur J Hum Genet.</i> 2011 Aug 10. [Epub ahead of print]	129
Chapter V	Discussion	165
	Summary	169
	Samenvatting voor de geïnteresseerde leek	171
List of Publications		173
Dankwoord		175
Curriculum Vitae		177