



Universiteit
Leiden
The Netherlands

Genetics of Ollier disease and Maffucci syndrome

Pansuriya, T.C.

Citation

Pansuriya, T. C. (2012, March 15). *Genetics of Ollier disease and Maffucci syndrome*. Retrieved from <https://hdl.handle.net/1887/18591>

Version: Corrected Publisher's Version

License: [Licence agreement concerning inclusion of doctoral thesis in the Institutional Repository of the University of Leiden](#)

Downloaded from: <https://hdl.handle.net/1887/18591>

Note: To cite this publication please use the final published version (if applicable).

Cover Page



Universiteit Leiden



The handle <http://hdl.handle.net/1887/18591> holds various files of this Leiden University dissertation.

Author: Pansuriya, Twinkal C.

Title: Genetics of Ollier disease and Maffucci syndrome

Issue Date: 2012-03-15

Genetics of Ollier disease and Maffucci syndrome

Genetics of Ollier disease and Maffucci syndrome

Proefschrift

ter verkrijging van de graad van Doctor aan de Universiteit Leiden,
op gezag van Rector Magnificus prof. mr. P.F. van der Heijden,
volgens besluit van het College voor Promoties
te verdedigen op donderdag 15 maart 2012
klokke 15.00 uur

door

Twinkal C. Pansuriya

geboren te Amreli, Gujarat, India

in 1985

Promotiecommissie:

Promotor: Prof. Dr. P.C.W. Hogendoorn

Co-promotor: Dr. J.V.M.G. Bovée

Leden: Prof. Dr. M. Breuning
Prof. Dr. A.J.H. Suurmeijer (University Medical Centre Groningen)
Prof. Dr. A.H.M. Taminiau
Dr. K. Szuhai
Dr. J. Oosting

Cover art & Lay-Out by : Four Clowns, Gujarat, India (Mehul Solanki).

Printed by: Wöhrmann print service, Zutphen. The Netherlands

Copyright © T. C. Pansuriya, Leiden, The Netherlands. All rights reserved.

The work presented in this thesis was financially supported by NWO (917-76-315) and EuroBoNet a European Commission-granted European Network of excellence for studying the pathology and genetics of bone tumours. Grant Number: LSHC-CT-2006-018814

Publication of this thesis is financially supported by MRC-Holland and Anna Fonds are highly acknowledged.

To honor my parents
To my beloved Rohan
With love to my sisters, brother-in-law and family

Pansuriya, Twinkal Chandubhai

Genetics of Ollier Disease and Maffucci
syndrome / Twinkal Chandubhai Pansuriya.

Thesis Leiden. - With ref. - With summary in Dutch.

ISBN: 978-90-8570-983-1

SISO UDC

Subject headings: Ollier Disease/Maffucci
Syndrome/genetics

Contents

Chapter 1	General Introduction	9
Chapter 2	Enchondromatosis: Insights on the different subtypes <i>Int J Clin Exp Pathol.</i> 2010, 26;3(6):557-69.	21
Chapter 3	Genome-wide analysis of Ollier disease: Is it all in the genes? <i>Orphanet J Rare Dis.</i> 2011, 14;6:2.	41
Chapter 4	Maffucci Syndrome: A Genome-Wide Analysis Using High Resolution SNP and Expression Arrays on Four Cases. <i>Genes Chromosomes Cancer.</i> 2011, 50(9):673-9.	65
Chapter 5	Analysis of NDST1, EXT1 and HS in central chondrosarcoma and Ollier related tumours.	83
Chapter 6	Somatic mosaic <i>IDH1</i> or <i>IDH2</i> mutations are associated with enchondroma and spindle cell hemangioma in Ollier disease and Maffucci syndrome <i>Nat Genet.</i> 2011, doi:10.1038/ng.1004.	97
Chapter 7	Summary and Concluding Remarks	133
	Nederlandse Samenvatting	141
	Acknowledgements	147
	List of Publications	149
	Curriculum Vitae	151

