PhD Thesis
School of Biomedical Sciences and Pharmacy
Faculty of Health and Medicine
University of Newcastle, NSW, Australia

Title: Clinical use of SNP-microarrays for the detection of genome-wide changes in haematological malignancies with a focus on B-cell neoplasms

Nadine Kaye Berry
BMedSc (*University of Western Sydney*)

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PREFACE

STATEMENT OF ORIGINALITY

I hereby certify that the work embodied in the thesis is my own work, conducted under normal supervision. The thesis contains no material which has been accepted, or is being examined, for the award of any other degree or diploma in any university or other tertiary institution and, to the best of my knowledge and belief, contains no material previously published or written by another person, except where due reference has been made. I give consent to the final version of my thesis being made available worldwide when deposited in the University's Digital Repository, subject to the provisions of the Copyright Act 1968 and any approved embargo.

Nadine Kaye Berry

THESIS BY PUBLICATION: ACKNOWLEDGEMENT OF AUTHORSHIP

I hereby certify that this thesis is in the form of a series of papers. I have included as part of the thesis a written declaration from each co-author, endorsed in writing by the faculty Assistant Dean (Research Training), attesting to my contribution to any jointly authored papers (Appendix 2.0).

Nadine Kaye Berry

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I dedicate this thesis to my late stepfather (step by name, father by love), James (Jim) Brunton. I owe this all to you! I will continue to make my work meaningful because of you. I will miss your joyful face whenever a significant moment was reached and also your inability to hide how proud you were.

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PUBLICATIONS INCLUDED IN THIS THESIS

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Publication 2: Berry, NK, Bain, NL, Enjeti, AK, and Rowlings, P, *Genomic profiling of plasma cell disorders in a clinical setting: integration of microarray and FISH, after CD138 selection of bone marrow.* J Clin Pathol, 2014. **67**(1): p. 66-9.

Publication 3: Berry, NK, Dixon-McIver, A, Scott, RJ, Rowlings, P, and Enjeti, AK, *Detection of complex genomic signatures associated with risk in plasma cell disorders.* Cancer Genet, 2017. **218-219**: p. 1-9.

Publication 4: Berry, NK, Scott, RJ, Sutton, R, Law, T, Trahair, TN, Dalla-Pozza, L, et al., *Enrichment of atypical hyperdiploidy and IKZF1 deletions detected by SNP-microarray in high-risk Australian AIEOP-BFM B-cell acute lymphoblastic leukaemia cohort.* Cancer Genet, 2020. **242**: p. 8-14.

TITLE PAGE FIGURE LEGEND

This image was a result representing chromosome 1, used to describe the complexity of genomic signatures identified in multiple myeloma. It was published in the Cancer Genetics journal. Berry, N. K., A. Dixon-McIver, R. J. Scott, P. Rowlings and A. K. Enjeti (2017). "Detection of complex genomic signatures associated with risk in plasma cell disorders." Cancer Genetics **218–219**: 1-9.

ABBREVIATIONS

ALL	Acute lymphoblastic leukaemia
AML	Acute myeloid leukaemia
B-ALL	B-cell acute lymphoblastic leukaemia
CGH	Comparative genomic hybridisation
CLL	Chronic lymphocytic leukaemia
CMA	Chromosomal microarray
Cn-LOH	Copy number loss of heterozygosity
CNV	Copy number variant
FISH	fluorescent in-situ hybridisation
GWAS	Genome-wide association study
LOH	Loss of heterozygosity
PCD	Plasma cell dyscrasia
PCR	Polymerase chain reaction
MDS	Myelodysplastic syndrome
MGUS	Monoclonal gammopathy of undetermined significance
MLPA	Multiplex Ligation-dependent Probe Amplification
MM	Multiple myeloma
MPS	Massively parallel sequencing
RT-PCR	Reverse transcription polymerase chain reaction
SNP	Single nucleotide polymorphism
T-ALL	T-cell acute lymphoblastic leukaemia