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Prediction of "BRCAness" in breast cancer by array comparative genomic hybridization

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List of Abbreviations

ALDH1	aldehyde dehydrogenase 1 family, member A1
APC	adenomatous polyposis coli
ATM	ataxia telangiectasia mutated
BAC	bacterical artificial chromosome
BARD1	BRCA1 associated RING domain 1
BCS	breast conserving surgery
BER	base excision repair
BIC	breast cancer information core
bp	base pair
BRCA	breast cancer, early onset
BRCT	BRCA1 C terminus (domain)
CHEK2	checkpoint kinase 2
CHK1	checkpoint kinase 1
CGH	comparative genomic hybridization
CI	confidence interval
CISH	chromogenic in situ hybridization
CNA	copy number alteration
CSC	cancer stem cell
CTRM	c-terminal RAD51 binding motive
DCIS	ductal carcinoma in situ
DGGE	denaturing gradient gel electrophoresis
DHPLC	denaturing high-performance liquid chromatography
DNA	deoxyribonucleic acid
DOL	degree of labeling
DSB	double-strand break
DSS1	deleted in split hand/foot protein 1
EDTA	ethylenediaminetetraacetic acid
EGFR	epidermal growth factor receptor
ER	estrogen receptor
ERBB2	human epidermal growth factor receptor 2
ESR1	estrogen receptor 1
FAMA	fluorescent-assisted mismatch analysis
F-CSGE	fluorescent conformational sensitive gel eletrophoresis
FE test	Fisher's exact test
FFPE	Formalin-fixed, paraffin embedded
GAPDH	glyceraldehyde-3-phosphate dehydrogenase
GE	gene expression
GEP	gene expression profiling
GEO	gene expression omnibus
HBOC	hereditary breast and ovarian cancer
HD	helix-rich domain
HDPB	high-dose platinum-based
H&E	hematoxylin and eosin
HR	homologous recombination
HRD	homologous recombination deficient/deficiency
IDC	invasive ductal carcinoma
IGF	insulin-like growth factor
IHC	immunohistochemistry
ILC	invasive lobular carcinoma
IVS	intervening sequence
kbp	kilo base pairs
KRT	keratin
LC	lobular carcinoma
LCIS	lobular carcinoma in situ
LOD	logarithm of odds
LOH	loss of heterozygosity
LOOCV	leave-one-out cross-validation
LUMC	Leiden University Medical Center
Mbp	mega base pairs
MLPA	multiplex ligation dependent probe amplification
MRI	magnetic resonance imaging
MYC	v-myc myelocytomatosis viral oncogene homolog

NCBI	National Center for Biotechnology Information
NGS	next generation sequencing
NHEJ	non-homologous end-joining
NKI	Netherlands Cancer Institute
NLS	nuclear localization signals
NPP	negative predictive power
OB	oligonucleotide binding
PALB2	partner and localizer of BRCA2
PAM	prediciton analysis of microarrays
PARP	poly ADP ribose polymerase
PBM	prophylactic bilateral mastectomy
PBSO	prophylatic bilateral salpingo-oophorectomy
PCR	polymerase chain reaction
PPP	positive predictive power
PR	progesteron receptor
PTEN	phosphatase and tensin homolog
PTT	protein truncation test
RAD51	RAD51 homolog
RAP80	receptor-associated protein 80
RING	really interesting new gene
RNA	ribonucleic acid
RT-PCR	realtime PCR
SC	shrunken centroids
SCD	SQ-cluster domain
SCP	single-stranded conformational polymorphism
SDS	sodium dodecyl sulfate
SNP	single nucleotide polymorphism
SSB	single-strand break
SSC	saline-sodium citrate
STK11	serine/threonine kinase 11
STR	short tandem repeat
TBE	tris/borate/EDTA
TDGS	two-dimensional gene scanning
TP53	tumor protein 53
TNM	tumor/node/metestasis
ULS	universal linkage system
UV	unclassified variant
VEGF	vascular endothelial growth factor

Curriculum Vitae

Simon Joosse was born on August 6, 1983 in IJmuiden (Velsen), the Netherlands. He started studying medical biology at the faculty of Applied Sciences at Inholland University in Alkmaar in 2000. His graduation report on the subject "Gene inactivation for cell immortality" was carried out under the supervision of Dr. R.L. Beijersbergen at the department of Molecular Carcinogenesis at the Netherlands Cancer Institute, Amsterdam, in 2004. After graduation, he worked at the Netherlands Cancer Institute as a technician for two years and performed the Master Course on Experimental Oncology in 2006, organized by the Oncology school of Amsterdam. In 2007, Simon started working as a PhD student at the department of Experimental Therapy in the Netherlands Cancer Institute, under the supervision of Dr. P.M. Nederlof in the group of Dr. L.J. van 't Veer. In 2009, he moved to Hamburg, Germany, where he started working as scientist for the Department of Tumor Biology, University Medial Center Hamburg-Eppendorf, while simultaneously finishing his PhD studies performed at the Netherlands Cancer Institute. The results of this research are described in this thesis.

List of Publications

1. **Joosse SA**, Hannemann J, Spötter J, Bauche A, Andreas A, Müller V, Pantel K. *Changes in keratin expression during metastatic progression of breast cancer: impact on the detection of circulating tumor cells*. Clin Cancer Res. 2012; [Online publication ahead of print]

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