## **Dedicated to**

# "<u>Maa and Pitai</u>"

### Declaration

I hereby declare that the thesis entitled "**Studies on immune related genes with genomic instability in cancer.**" is an authentic work carried out by me under the supervision of Prof. Shashi Baruah, Department of molecular Biology and Biotechnology, Tezpur University, Assam-784028. No part of this has been presented for any other degree or diploma earlier.

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#### **CERTIFICATE OF SUPERVISOR**

This is to certify that the thesis entitled **"Studies on immune related genes with genomic instability in cancer."** submitted to the School of Sciences, Tezpur University in requirement of partial fulfilment for the award of the degree of Doctor of Philosophy in Molecular Biology and Biotechnology is a record of research work carried out by **Ms. Bhaswatee Das** under my supervision and guidance.

All help received by her from various sources have been duly acknowledged. No part of this thesis has been submitted elsewhere for award of any other degree.

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### **LIST OF ABBREVIATIONS**

G4	G-quadruplexes
HLA	Human leukocyte antigen
NK	Natural killer cell
IL-10	Interleukin 10
TGF-β	Transforming growth factor-beta
CTLA-4	Cytotoxic T-lymphocyte-associated protein 4
PD-1	Programmed Cell Death Protein 1
PD-L1	Programmed Cell Death Ligand 1
Tregs	Regulatory T cells
ТАМ	Tumor associated Macrophages
MDSC	Myeloid derived suppressor cells
CSF1	Colony stimulating factor 1
CCL2	Chemokine (C-C motif) ligand 2
CCL22	Chemokine (C-C motif) ligand 22
CCL5	Chemokine (C-C motif) ligand 5
CXCL5	C-X-C motif chemokine 5
CXCL8	C-X-C motif chemokine 8
KRAS	Ki-ras2 Kirsten rat sarcoma viral oncogene homolog
BRAF	B-Raf
CASP8	Cysteine-aspartic acid protease
CNA	Copy Number Alteration
TG	Thyroglobulin
PIK3CA	Phosphatidylinositol-4,5-Bisphosphate 3-Kinase Catalytic Subunit
	Alpha
PIK3/AKT	Phosphoinositide-3-kinase-protein kinase B/Akt
PI3K	Phosphatidylinositol-4,5-bisphosphate 3-kinase
МНС	Major Histocompatibility complex
HLA-A	Human leukocyte antigens A

HLA-B	Human leukocyte antigens B
HLA-C	Human leukocyte antigens C
HLA-DR	Human leukocyte antigens DR
HLA-DP	Human leukocyte antigens DP
HLA-DQ	Human leukocyte antigens DQ
TNF	Tumor necrosis factor
C3	Complement component 3
C4	Complement component 4
C5	Complement component 5
HLA-DRB1	Human leukocyte antigens DRB1
CIITA	CIITA class II major histocompatibility complex transactivator
NSCLC	Non-small cell lung carcinoma
TNF-α	Tumor necrosis factor alpha
IFN-γ	Interferon-gamma
HLA-G	Human leukocyte antigens G
HLA-E	Human leukocyte antigens E
IL-10	Interleukin 10
TGF-β	Transforming growth factor-beta
IDH1	Iocitrate Dehydrogenase 1
IDH2	Isocitrate Dehydrogenase 2
STING	Stimulator of Interferon Genes
B2M	beta-2 microglobulin
TME	Tumor microenvironment
TIME	Tumor immune microenvironment
CAR-T	Chimeric Antigen Receptor T-cell
TCGA	The Cancer Genome Atlas
ICGC	International Cancer Genome Consortium
ACC	Adrenocortical carcinoma
BLCA	Bladder Urothelial Carcinoma
BRCA	Breast invasive carcinoma
CHOL	Cholangiocarcinoma

COAD	Colorectal adenocarcinoma
CESC	Cervical squamous cell carcinoma and endocervical adenocarcinoma
НСС	Hepatocellular carcinoma
HNSC	Head and Neck Squamous cell Carcinoma
KICH	Kidney Chromophobe
KIRC	Kidney renal clear cell carcinoma
KIRP	Kidney renal papillary cell carcinoma
LUAD	Lung adenocarcinoma
LUSC	Lung squamous cell carcinoma
OV	Ovarian carcinoma
PRAD	Prostate adenocarcinoma
ESCA	Esophageal adenocarcinoma
THCA	Thyroid carcinoma
UCEC	Uterine Corpus Endometrial Carcinoma
GBM	Glioblastoma multiforme
LGG	Brain Lower Grade Glioma
PCPG	Pheochromocytoma and Paraganglioma
DLBC	Diffuse large B cell lymphoma
LAML	Acute Myeloid Leukemia
SARCOMA	Sarcoma
GISTIC	Genomic Identification of Significant Targets in Cancer
DEG	Differentially expressed genes
GEPIA	Gene Expression Profiling Interactive Analysis
COSMIC	COmmon Software Measurement International Consortium
TIMER	Tumor Immune Estimation Resource
TCR	T-cell receptor
BCR	B-cell receptor
PQRS	putative G4
M1	M1 macrophages
ACE2	Angiotensin Converting Enzyme 2
IDO1	Indoleamine 2,3-Dioxygenase 1

IDO2	Indoleamine 2,3-dioxygenase 2
CD8+	Cytotoxic T-cells
CD4+	Helper T-cells
B cell	Plasma cells
M0	Undifferentiated macrophages
M1	M1 macrophages
M2	M2 macrophages
DC	Dendritic cells
MAC	Membrane attack complex
KNG1	Kininogen 1
F2	Coagulation factor II, thrombin
FGA	Fibrinogen alpha chain
FGB	Fibrinogen beta chain
FGG	Fibrinogen gamma chain
SERPIND1	Serpin family D member 1
VTN	Vitronectin
CFHR5	Complement Factor H Related 5
C3P1	Complement Component 3 Precursor Pseudogene
CFH	Complement Factor H
CFHR3	Complement Factor H Related 3
F13A1	Coagulation Factor XIII, A Polypeptide
BDKRB2	Bradykinin Receptor B2
C6	Complement C6
C7	Complement C7
CR1	Complement C3b/C4b Receptor 1
C4BPB	Complement Component 4 Binding Protein Beta
RFXANK	regulatory factor X-associated protein-containing ankyrin repeat
RFXAP	Regulatory factor X-associated protein
NFYA	Nuclear transcription factor Y subunit alpha
NFYB	Nuclear transcription factor Y subunit beta
NFYC	Nuclear transcription factor Y subunit gamma

CREB	cAMP-response element binding protein
RFX5	Regulatory factor X5
Th1	Type 1 T helper cells
C3b	C3b is the larger of two elements formed by the cleavage
	of complement component 3
C5a	Complement C5a Anaphylatoxin
C5aR	Complement component 5a receptor 1