HUMAN LEUKOCYTE ANTIGEN (HLA) AND AUTOANTIBODIES ASSOCIATION WITH JUVENILE SYSTEMIC LUPUS ERYTHEMATOSUS

Thesis
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IN

RHEUMATOLOGY AND REHABILITATION

by

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DEDICATION

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To My Parents

I dedicate this humble work to my mother and my father, who were and will always be my tower of support. I also dedicate every success and every achievement I accomplish to them, as their endless love, care and guidance were the real inspiration for every good work I do.

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ABSTRACT

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A person's ability to manufacture pathogenic immunoglobulin in SLE and sustain its production depends on intrinsic abnormalities of B and T lymphocytes, and those in turn depend on inheriting an appropriate number of susceptibility genes, lacking protective genes, and encountering an environmental stimulus that sets the whole process into action. The aim of this work is to elucidate the role of HLA genes and autoantibodies in the clinical presentation of SLE and thus to evaluate the usefulness of such genetic and autoimmune markers in predicting disease course and outcome. Fifty three Egyptian patients with childhood onset systemic lupus erythematosus were examined for clinical manifestations, detection of ANA, anti-DNA, anti-Ro (SS-A), anti-La (SS-B), anti-Sm, anti-RNP and anti-cardiolipin antibodies and for determination of HLA-DR alleles. This study showed an association of certain MHC antigens and autoantibodies with the development of certain disease manifestations.

Key Words

Juvenile systemic lupus erythematosus in Egyptians

Autoantibodies and HLA in childhood onset systemic lupus erythematosus

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LIST OF ABBREVIATIONS

Ab	Antibody
aCL	Anticardiolipin
Ag	Antigen
ANA	Antinuclear antibody
aPL	Antiphospholipid
APS	Antiphospholipid syndrome
ARA	American Rheumatism Association
C.C.J.	Costochondral joint
CDR	Complementary determining region
CHB	Congenital heart block
CREST	Calcinosis – Raynaud's phenomenon – esophageal dysmotility – sclerodactyly – telangiectasias
CSF	Cerebrospinal fluid
CTscan	Computer tomographic scan
CVA	Cerebrovascular accidents
DLE	Discoid lupus erythematosus
DNA	Deoxyribonucleic acid
dsDNA	Double stranded DNA
EBV	Epstein-Barr virus
ECG	Electrocardiograph
E coli	Escherichia coli
EEG	Electroencephalograph
ELISA	Enzyme linked immunosorbent assay
ENA	Extractable nuclear antigen
FANA	Fluorescent antinuclear antibody
FW	Frame work
HLA	Human leukocyte antigen
IC	Immune complex
Id	Idiotype
Ig	Immunoglobulin
JRA	Juvenile rheumatoid arthritis
LAC	Lupus anticoagulant

LIST OF ABBREVIATIONS

LCA	Lymphocytotoxic antibody
mAb	Monoclonal antibody
M.C.Ps.	Metacarpophalangeal joints
MCTD	Mixed connective tissue disease
M.T.Ps.	Metatarsophalangeal joints
MHC	Major histocompatibility complex
MLC	Mixed lymphocyte cultures
MRI	Magnetic resonance imaging
NLS	Neonatal lupus syndrome
NOR-90	90 kilodalton protein of the chromosomal nucleolar organizing region
nRNP	Nuclear ribonucleoprotein
NSAIDs	Nonsteroidal antiinflammatory drugs
PAPS	Primary antiphospholipid syndrome
PBS	Physiologic buffer solution
PCNA	Proliferating cell nuclear antigen
PCR	Polymerase chain reaction
P.I.Ps.	Proximal interphalangeal joints
PSS	Progressive systemic sclerosis
PTT	Partial thromboplastin time
R.O.M.	Range of motion
RA	Rheumatoid arthritis
rbc	Red blood cell
RF	Rheumatoid factor
RFLP	Restriction fragment length polymorphism
RIA	Radioimmunoassay
RNA	Ribonucleic acid
RNA P II	RNA polymerase II
RNP	Ribonucleoprotein
RP1	RNA polymerase 1
RR	Relative risk
S.C.J.	Sternoclavicular joint
ScL	Scleroderma

LIST OF ABBREVIATIONS

SCLE	Subacute cutaneous lupus erythematosus
scRNP	Small cytoplasmic ribonucleoprotein
SD	Standard deviation
SLAM	Systemic Lupus Activity Measure
SLE	Systemic lupus erythematosus
Sm	Smith
snRNP	Small nuclear ribonucleoprotein
SPARC	Secreted protein acidic rich in cysteine (also called osteonectin, BM-40)
SPECT	Single photon emission computed tomography
SS	Sjogren's syndrome
ssDNA	Single stranded DNA
SSO	Sequence specific oligonucleotides
TAP 1,2	Transporters of antigenic peptides 1 and 2
TcR	T cell receptor
Th cells	T helper cells
T.I.Ps.	Terminal interphalangeal joints
TM	Transverse myelitis
T.M.Js.	Tempromandibular joints
TNF	Tumor necrosis factor
U_1RNP	Uridine 1 ribonucleoprotein
V_H	Variable region of heavy chain
V_L	Variable region of light chain
wbc	White blood cell
WHO	World health organization

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