Immunological Profile of Patients with Skeletal Dysplasia and Disproportionate Short Stature

Thesis
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LIST OF ABBREVIATIONS

Abbrev.	*Full term*
ACP5	Acid phosphatase 5, tartrate resistant gene
ADA	Adenosine deaminase
AD	Autosomal dominant
AIRE	Autoimmune regulator
AK2	Adenylate kinase
ALPS	Autoimmune lymphoproliferative syndrome
APECED	Autoimmune polyendocrinopathy-candidiasis-ectodermal dystrophy
APLAID	Autoinflammation and PLCG2-associated antibody deficiency and immune dysregulation
AR	Autosomal recessive
CANDLE	Chronic atypical neutrophilic dermatosis with lipodystrophy and elevated temperature
CASP	Caspase
CBC	Complete blood count
CD	Cluster of differentiation
CGD	Chronic granulomatous disease
cj	coding joint
CHH	Cartilage-Hair Hypoplasia
CHARGE	Coloboma, heart defects, atresia choanae, growth retardation, genital abnormalities, and ear abnormalities
cm	Centimeter
CTRL	Control
CORO1A	Coronin 1A deficiency
CVID	Common variable immunodeficiency
DBS	Dried blood spot
DCLRE1C	DNA cross-link repair enzyme 1C
DHR	Dihydrorhodamine
DKC	Dyskeratosis Congenita
DNA	Deoxyribonucleic acid
EDTA	Ethylene diamine tetra-acetic acid

Abbrev.	*Full term*
ESID	European Society of Immunodeficiency Disorders
EVER	Epidermodysplasia veruciformis
FASL	Fas ligand
FILS	Facial dysmorphism, immunodeficiency, livedo and short stature
FITC	Fluorescein isothiocyanate
FMF	Familial Mediterranean Fever
FOXP3	Forkhead box protein 3
gDNA	genomic DNA
G6PDH	Glucose-6-phosphate dehydrogenase
Hb	Hemoglobin
HIV	Human immunodeficiency virus
ID	Immunodeficiency
IDR	Immunodeficiency-related
IFN	Interferon
Ig(s)	Immunoglobulin(s)
IPEX	Immunodysregulation polyendocrinopathy enteropathy X-linked
ITP	Idiopathic thrombocytopenic purpura
IUIS	International Union of Immunological Societies
IVIG	Intravenous immunoglobulin
JAK3	Janus kinase 3
LIG4	Ligase
LAD	Leukocyte adhesion deficiency
Max.	Maximum
MHC	Major histocompatibility complex
Min.	Minimum
ml	Milliliters
MoAbs	Monoclonal antibodies
MTHFD	Methylenetetrahydrofolate dehydrogenase
MW	Molecular weight
NBS	New born screening
NCBI	National Center for Biotechnology Information
NHEJ1	Nonhomologous end-joining protein 1

Abbrev.	*Full term*
NK	Natural Killer
NRAS	Neuroblastoma RAS viral oncogene homolog
NRC	National Research Centre
NS	Non-significant
PBS	Phosphate buffered saline
PCR	Polymerase chain reaction
PE	Phycoerythrin
PHA	Phytohaemagglutinin
PID EC	Primary Immunodeficiency Expert Committee
PIDs	Primary immunodeficiencies
PLAID	PLCG2 associated antibody deficiency and immune dysregulation
PNP	Purine nucleoside phosphorylase
PRKDC	DNA-dependent protein kinase Cernunnos
PTPRC	Protein tyrosine phosphatase receptor type C
RAG	Recombination-activating gene
RMRP	Mitochondrial RNA-processing endoribonuclease gene
RNase P	Ribonuclease P
RNU4ATAC	small nuclear RNA, U4, AT-AC form gene
rpm	Revolutions per minute
rs	Correlation coefficient
S	Significant
SAC	Staphylococcus aureus Cowan antigen
SCID	Severe combined immunodeficiency
SD	Standard deviation
SEMD	Spondyloepimetaphyseal Dysplasia
SIOD	Schimke Immunoosseous Dysplasia
sj	Signal Joint
SLE	Systemic lupus erythematosus
SMARCAL1	SWI/SNF related, matrix associated, actin dependent regulator of chromatin, subfamily A-like1 gene
SMD	Spondylometaphyseal dysplasia
SPENCDI	Spondyloenchondrodysplasia with Immune Dysregulation

Abbrev.	*Full term*
SPSS	Statistical Package for Special Sciences
SPUR	Serious, Persistent, Unusual (pathogens or site) and Recurrent
ST	Standard
TCL	T cell lymphopenia
TCR	T cell receptor
TCR-δ	T cell receptor delta
TREC	T-cell receptor excision circle
TSH	Thyroid stimulating hormone
VDJ	variable, diversity and joining
VODI	Hepatic veno-occlusive disease with immunodeficiency
WAS	Wiscott Adrich Syndrome
WB	Whole blood
WBCs	White blood cells
WHIM	Warts, Hypogammaglobulinemia, Immuno-deficiency and Myelokathexis syndrome
WHO	World Health Organization
αβ-TCR	alpha/beta T cell receptor
δREC	Delta recombination element
δREC–ψJα TREC	Delta-Rec and psi-Joining segment-alpha Tcell receptor excision circle

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INTRODUCTION