Genetic Basis of Dilated Cardiomyopathy in Children

Essay

Submitted for Partial Fulfillment of Master Degree in Pediatrics

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Zakia Badr Sayed Mohamed

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List of Abbreviations

ABCC9	ATP-binding cassette, subfamily C, member 9
ACE	Angiotensin-converting enzyme
ACTC1	Actin, cardiac muscle 1
AD	Autosomal dominant
AFLP	Acute fatty liver of pregnancy
AHA	The American Heart Association
AIDS	Acquired immunodeficiency syndrome
ANKRD1	Ankyrin repeat domain 1
ANP	Atrial natriuretic peptide
ARVC	Arrhythmogenic right ventricular cardiomyopathy
BAG3	BCL2- associated athanogene 3
B-MHC	B-Myosin heavy chain
BNP	B-type natriuretic peptide
CARP	Cardiac arkyrin repeat protein
CASQ2	Calsequestrin 2
CAV3	Caveolin 3
CDDC	Cardiac conduction system disease
CK	Creatine kinase

CLIA	Clinical Laboratories Improvement Act
CM	Cardiomyopathy
CMR	Cardiac magnetic resonance
CPVT	Catechoteminergic polymorphic ventricular tachycardia
CSRP3	Cysteine and glycine –rich protein 3
CTF1	Cardiotrophin 1
CTNT	Cardiac troponine T
CVB3	Coxsackievirus B3
DCM	Dilated cardiomyopathy
DES	Desmin
DMD	Duchenne muscular dystrophy
DSG2	Desmoglein 2
DSP	Desmoplakin
DTMRI	Diffusion tensor magnetic resonance imaging
ECG	Electrocardiogram
EF	Ejection fraction
EMD	Emerein gene
EMD	Emery-Dreifuss muscular dystrophy
EYA4	Eys absent homoiog 4
FDA	Food and Drug Administration

FDC	Familial Dilated Cardiomyopathy
FKRP	Fukutin-related protein
FKTN	Fukutin
FOXD4	Forkhead box D4
FSHMD	Facioscapulohumeral muscular dystrophy
GCSF	Granulocyte colony stimulating factor
GH	Growth hormone
GLA	Galactosidase
GM-CSF	Granulocyte,Monocyte –Colony stimulating factor
HCM	Hypertrophic cardiomyopathy
HELP	Hemolysis, elevated liver enzymes, and low platelet count
HF	Heart failure
HIV	Human immunodeficiency virus
HLA	Human leukocyte antigen
ICD	Implantable cardioverter defibrillator
IDC	Idiopathic dilated cardiomyopathy
IEM	Inborn errors of metabolism
IGF	Insulin like growth factor
IV	Intravenous
JUP	Junction plakoglobin

LAMA	Laminin
LAMP2	Lysosomal-associated membrane protein 2
LAP2α	Lamin associated polypeptide 2α
LBBB	Left bundle branch block
LDB3	LIM domain binding 3
LGMD	Limb girdle muscular dystrophy
LMNA	Lamin A/C
LVADS	Left ventricular assist devices
LVNC	Left ventricular non compaction cardiomyopathy
MLP	Muscle LIM protein
MRI	Magnetic resonance imaging
MYBPC	Myocin binding protein
MYH	Myosin heavy chain
MYL	Myosin light chain
MYLK2	Myosin light chain kinase 2
MYOZ2	Myozenin 2
NEXN	Nexilin
NYHA	New York heart association
PCMR	The pediatric cardiomyopathy registry
PCR	Polymerase chain reaction

PDEI	Phosphodiesterase Enzyme Inhibitors
PKP2	Plakophilin 2
PLN	Phospholamban
PRKAG2	Protein kinase,AMP-activated
PSEN	Presenilin
RB20	Ribonucleic acid binding protein
RBM20	RNA binding motif protein 20
RCM	Restrictive cardiomyopathy
RYR2	Ryanodine receptor 2
SAEGG	Signal averaged electrocardiography
SCD	Sudden cardiac death
SCN5A	Sodium channel , voltage-gated,type V
SDHA	Succinat dehydrogenase complex,sub unit A
SGCD	Sarcoglycan
SND	Sinus node dysfunction
SOLVD	Studies of left ventricular dysfunction
SYNE1	Spectrin repeat containing, nuclear envelope 1
SYNE2	Spectrin repeat containing, nuclear envelope 2
TAZ	Tafazzin
TCAP	Titin-cap
TK	Titin kinase

TMEM43	Transmembran protein 43
TMPO	Thymopoietin
TNNC1	Troponin C type 1
TNNT2	Troponin T Type 2
TNT	Tissue Necrotizing factor
TPM	Tropomyosin
TPM1	Tropomysin 1
TTE	Transthoracic echocardiography
TTN	Titin
TTR	Transthyretin
US	United States
VCL	Vinculin
VF	Ventricular fibrillation
VT	Ventricular tachycardia
WHO	The World Health Organization
XL	X-linked
ZLS	Zimmermann-laband syndrome

Introduction

INTRODUCTION

Cardiomyopathy is an anatomic and pathologic diagnosis associated with muscle dysfunction or electrical dysfunction of the heart. The American Heart Association (AHA) defines cardiomyopathy as a heterogenous group of diseases of the myocardium, usually with inappropriate ventricular hypertrophy or dilatation **(Wexler et al., 2009)**.

Under the new cardiomyopathy classification, it includes dilated cardiomyopathy (DCM), hypertrophic cardiomyopathy (HCM), restrictive cardiomyopathy (RCM), arrhythmogenic right ventricular cardiomyopathy (ARVC), and left ventricular non compaction cardiomyopathy (LVNC). Many of these disorders are genetically inherited, while a small group of these diseases are acquired **(Elliot et al., 2008)**.

DCM is the most common type of cardiomyopathy, accounting for 60% of all primary cardiomyopathies and is a leading cause of heart failure and heart transplants. DCM is a primary heart muscle disease that is characterized by progressive ventricular dilatation and impaired systolic function **(Menon et al., 2008)**.

The annual incidence of dilated cardiomyopathy in children younger than 18yrs was 0.57 per 100,000 per year overall in