Molecular Diagnosis of Myotonic Dystrophy Type 1 in Egyptian Patients

Thesis

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Master Degree in Clinical and Chemical Pathology

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TO MY FATHER

TO MY MOTHER

TO MY HUSBAND

AND TO ALL OF MY DEAR FAMILY

بسم الله الرحمن الرحيم

وما أوتيتم من العلم إلا قليلا

صدق الله العظيم

(الإسراء:٨٥)

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List of Abbreviations

3'UTR	The 3' untranslated region
APS	Ammonium persulfate
Bp	Base pair
CK	Creatine kinase
CTG	Cytosine, thymine, guanine
CUG	Cytosine, uracil, guanine
CUG Bp	CUG binding protein
D.D. H2O	Double distilled H2O
dGTP	Deoxyguanosine-Triphosphate
DM	Dystrophia myotonica
DMPK	Dystrophia myotonica protein kinase
DMWD	Dystrophia myotonica-containing WD repeat
DNA	Deoxyribonucleic acid
dNTPs	Deoxyribo nucleotide triphosphate
ECG	Electrocardiogram
EDTA	Ethylenediaminetetraacetic acid
EMF	Electromotive force
EMG	Electromyography
FCGRT	Fc fragment of IgG, receptor, transporter, alpha
IgG	Immunoglobin G
Kb	Kilo base
Mb	Mega base

List of Abbreviations

MBNL	Muscle blind like protein
mRNA	Messenger ribonucleic acid
PAGE	Polyacrylamide gel electrophoresis
PCR	Polymerase chain reaction
RNA	Ribonucleic acid
SDS	Sodium dodecyl sulphate
SIX 5	SIX homeobox 5
TAE buffer	Tris-acetate-EDTA
Taq	Thermus aquaticus
TBE	Tri-borate EDTA
TE Buffer	Tris-EDTA buffer
TEMED	(N, N, N, N-Tetramethylethylenediamine)
UV	Ultraviolet
ZNF9	Zinc finger 9

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Glossary

3' Untranslated Region	The part of the mRNA which lays between the signal for the termination of translation (the stop codon) and the poly A tail.
Allele	Alternative form of a genetic locus; a single allele for each locus is inherited from each parent (e.g., at a locus for eye color the allele might result in blue or brown eyes). In an individual, one form of the allele (the dominant one) may be expressed more than another form (the recessive one).
Amplification	Any process by which specific DNA sequences are replicated disproportionately greater than their representation in the parent molecules.
Annealing	A process where the forward and reverse primers anneal to separated DNA template strand.
Autosome	Any chromosome that is not a sex (X or Y) chromosome.
Base pair (bp)	The fundamental unit of a double stranded DNA molecule, (more strictly-a nucleotide pair). Two nitrogenous bases (adenine and thymine or guanine and cytosine) held together by weak bonds. Two strands of DNA are held together in the shape of a

Glossary

	double helix by the bonds between base pairs.
Chromatin	Is the complex combination of DNA, RNA, and protein that makes up chromosomes. It is found inside the nuclei of eukaryotic cells, and within the nucleoid in prokaryotic cells.
Chromosome	The self-replicating genetic structure of cells containing the cellular DNA that bears in its nucleotide sequence the linear array of genes. In prokaryotes, chromosomal DNA is circular, and the entire genome is carried on one chromosome.
Cloned DNA	Any DNA fragment that passively replicates in the host organism after it has been joined to a cloning vector.
Cloning	The experimental process of making genetically identical copies.
Codons	In DNA or RNA, a sequence of three nucleotides that codes for a certain amino acid or signals the termination of translation.
Congenital	Any trait present at birth, whether the result of a genetic or non-genetic factor.

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Cytogenetics	The study of chromosomes.
Deletion	A mutation in either RNA or DNA in which one or more nucleotides are removed from the polynucleotide strand.
Denaturation	The separation of double stranded DNA into single stranded DNA by heat or chemical means.
DNA	The molecule that encodes genetic information. DNA is a double stranded molecule held together by weak bonds between base pairs of nucleotides. The four nucleotides in DNA contain the bases: adenine (A), guanine (G), cytosine (C), and thymine (T). In nature, base pairs form only between A and T and between G and C; thus the base sequence of each single strand can be deduced from that of its partner.
Domain	A discrete portion of a protein with its own function. The combination of domains in a single protein determines its overall function. A folded region of a polypeptide chain that varies in size from about 40 to 400 amino acid residues.
Downstream	Describing a location or a sequence of units in the direction in which a process occurs i.e. sequence