

# **Molecular Diagnosis of Myotonic Dystrophy Type 1 in Egyptian Patients**

*Thesis*

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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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# التشخيص الجزيئى لمرض التوتر العضلى الضمورى فى المرضى المصريين

## رسالة

توطئة للحصول على درجة الماجيستر فى الباثولوجيا الإكلينيكية والكيميائية

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# List of Abbreviations

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<b>3'UTR</b>	The 3' untranslated region
<b>APS</b>	Ammonium persulfate
<b>Bp</b>	Base pair
<b>CK</b>	Creatine kinase
<b>CTG</b>	Cytosine, thymine, guanine
<b>CUG</b>	Cytosine, uracil, guanine
<b>CUG Bp</b>	CUG binding protein
<b>D.D. H<sub>2</sub>O</b>	Double distilled H <sub>2</sub> O
<b>dGTP</b>	Deoxyguanosine-Triphosphate
<b>DM</b>	Dystrophia myotonica
<b>DMPK</b>	Dystrophia myotonica protein kinase
<b>DMWD</b>	Dystrophia myotonica-containing WD repeat
<b>DNA</b>	Deoxyribonucleic acid
<b>dNTPs</b>	Deoxyribo nucleotide triphosphate
<b>ECG</b>	Electrocardiogram
<b>EDTA</b>	Ethylenediaminetetraacetic acid
<b>EMF</b>	Electromotive force
<b>EMG</b>	Electromyography
<b>FCGRT</b>	Fc fragment of IgG, receptor, transporter, alpha
<b>IgG</b>	Immunoglobulin G
<b>Kb</b>	Kilo base
<b>Mb</b>	Mega base

# List of Abbreviations

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

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# Glossary

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<b>3' Untranslated Region</b>	The part of the mRNA which lays between the signal for the termination of translation (the stop codon) and the poly A tail.
<b>Allele</b>	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).
<b>Amplification</b>	Any process by which specific DNA sequences are replicated disproportionately greater than their representation in the parent molecules.
<b>Annealing</b>	A process where the forward and reverse primers anneal to separated DNA template strand.
<b>Autosome</b>	Any chromosome that is not a sex (X or Y) chromosome.
<b>Base pair (bp)</b>	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

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	double helix by the bonds between base pairs.
<b>Chromatin</b>	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.
<b>Chromosome</b>	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.
<b>Cloned DNA</b>	Any DNA fragment that passively replicates in the host organism after it has been joined to a cloning vector.
<b>Cloning</b>	The experimental process of making genetically identical copies.
<b>Codons</b>	In DNA or RNA, a sequence of three nucleotides that codes for a certain amino acid or signals the termination of translation.
<b>Congenital</b>	Any trait present at birth, whether the result of a genetic or non-genetic factor.

# Glossary

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<b>Cytogenetics</b>	The study of chromosomes.
<b>Deletion</b>	A mutation in either RNA or DNA in which one or more nucleotides are removed from the polynucleotide strand.
<b>Denaturation</b>	The separation of double stranded DNA into single stranded DNA by heat or chemical means.
<b>DNA</b>	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.
<b>Domain</b>	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.
<b>Downstream</b>	Describing a location or a sequence of units in the direction in which a process occurs i.e. sequence