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## APPENDICES

## APPENDIX A

### BUFFERS AND REAGENT

#### 1. Lysis Buffer I

Sucrose	109.54	g
1.0 M Tris – HCl (pH 7.5)	10	ml
1.0 M MgCl <sub>2</sub>	5	ml
Triton X – 100 (pure)	10	ml
Distilled water to	1,000	ml

Sterilize the solution by autoclaving and store in a refrigerator (at 4°C).

#### 2. Lysis Buffer II

5.0 M NaCl	15	ml
0.5 M EDTA (pH 8.0)	48	ml
Distilled water to	1,000	ml

Sterilize the solution by autoclaving and store at room temperature.

#### 3. 10% SDS solution

Sodium dodecyl sulfate	10	g
Distilled water to	100	ml

Mix the solution and store at room temperature.

#### 4. 20 mg/ml Proteinase K

Proteinase K	2	mg
Distilled water to	1	ml

Mix the solution and store in a refrigerator (at -20°C).

#### 5. 1.0 M Tris – HCl

Tris base	12.11	g
Dissolve in distilled water and adjusted pH to 7.5 with HCl		
Distilled water to	100	ml

Sterilize the solution by autoclaving and store at room temperature.

**6. 0.5 M EDTA (pH 8.0)**

Disodium ethylenediamine tetraacetate.2H<sub>2</sub>O 186.6 g

Dissolve in distilled water and adjusted pH to 8.0 with NaOH

Distilled water to 1,000 ml

Sterilize the solution by autoclaving and store at room temperature.

**7. 1.0 M MgCl<sub>2</sub> solution**

Magnesium chloride.6H<sub>2</sub>O 20.33 g

Distilled water to 100 ml

Dispense the solution into aliquots and sterilize by autoclaving.

**8. 5 M NaCl solution**

Sodium chloride 29.25 g

Distilled water to 100 ml

Dispense the solution into aliquot and sterilize by autoclaving.

**9. 10X Tris borate buffer (10X TBE buffer)**

Tris – base 100 g

Boric acid 55 g

0.5 M EDTA (pH 8.0) 40 ml

Adjust volume to 1,000 ml with distilled water. The solution was mixed and store at room temperature.

**10. 5.0 M NaOH**

NaCl 8.0 g

KCl 0.2 g

CaCl<sub>2</sub>·2H<sub>2</sub>O 0.132 g

MgCl<sub>2</sub>·6H<sub>2</sub>O 0.1 g

Distilled water 800 ml

Sterilize the solution A and B by autoclaving and adjusted pH to 7.0 with solution B into sterile bottles.

Distilled water to 1,000 ml



## 11. 6X loading dye

Bromphenol blue	0.25	g
Xylene cyanol	0.25	g
Glycerol	50	ml
1M Tris (pH 8.0)	1	ml
Distilled water until	100	ml

Mixed and stored at 4<sup>0</sup>C

12. 10.0 M Ammonium acetate (CH<sub>3</sub>COONH<sub>4</sub>)

Ammonium acetate	77.08	g
Distilled water	80	ml

Adjust volume to 100 ml with distilled water and sterilize by autoclaving.

## 13. 25:24:1 (v/v) Phenol-chloroform-isoamyl alcohol

Phenol	25	volume
Chloroform	24	volume
Isoamyl alcohol	1	volume

Mix the reagent and store in a sterile bottle kept in a refrigerator.

## 14. 1.0% Agarose gel (w/v)

Agarose	1.0	g
1X TBE	100	ml

## 1.5% Agarose gel (w/v)

Agarose	1.5	g
1X TBE	100	ml

## 3.0% Agarose gel (w/v)

Agarose	1.5	g
1X TBE	50	ml

Dissolve by heating in microwave oven and occasional mix until no granules of agarose are visible.

**15. Ethidium bromide**

Ethidium bromide	10	mg
Distilled water	1	ml

Mix the solution and store at 4°C

**16. LB agar**

Agar	1.5	g
Peptone	1.0	g
Yeast extract	0.5	g
NaCl	0.5	g
Distilled water	100	ml

Sterilize the solution by autoclaving and store at 4°C

**17. LB broth**

Peptone	1.0	g
Yeast extract	0.5	g
NaCl	0.5	g
Distilled water	100	ml

Sterilize the solution by autoclaving and store at 4°C

**18. SOC medium**

Peptone	2.0	g
Yeast extract	0.5	g
1M NaCl (1 ml)	0.06	g
1M KCl (0.25 ml)	0.02	g
2M Mg <sup>2+</sup>	1.0	ml
2M Glucose	1.0	ml
Distilled water	97.0	ml

Sterilize the solution by autoclaving and store at 4°C

**19. 0.1 M Sodium formate (HCOONa) buffer**

Sodium formate                      0.01    g

Dissolve in distilled water and adjusted pH to 2.7 with 5.0 M HCl

Distilled water to                      100    ml

**20. 0.5 M Glycine-NaOH buffer**

Glycine                                      37.5    g

Dissolve in distilled water and adjusted pH to 10.3 with 5.0 M NaOH

Distilled water to                      1,000   ml

**21. 1X Phosphate-Buffered (PBS)**

Solution A :

NaCl                                      8.0    g

KCl                                        0.2    g

CaCl<sub>2</sub>·2H<sub>2</sub>O                      0.132   g

MgCl<sub>6</sub>H<sub>2</sub>O                        0.1    g

Distilled water                      800    ml

Solution B :

Na<sub>2</sub>HPO<sub>4</sub>                        1.15   g

KH<sub>2</sub>PO<sub>4</sub>                        0.2    g

Distilled water                      200    ml

Sterilize the solution A and B by autoclaving and adjusted pH to 7.0 with solution B into sterile bottles.

Distilled water to                      1,000   ml

## APPENDIX B

## GLOSSARY

**cDNA (complementary DNA):** DNA synthesized by the enzyme reverse transcriptase using mRNA as template.

**Compound heterozygote:** The presence of two different mutant alleles at a particular gene locus, one on each chromosome of a pair. The human genome contains two copies of each gene, a paternal and a maternal allele. A mutation affecting only one allele is called heterozygous. A homozygous mutation is the presence of the identical mutation on both alleles of a specific gene. However, when both alleles of a gene harbor mutations, but the mutations are different, these mutations are called compound heterozygous. Also called a genetic compound.

**Frameshift mutation:** (also called a framing error) is a genetic mutation caused by indels, ie. insertion or deletion of a number of nucleotides that is not evenly divisible by three from a DNA sequence. Due to the triplet nature of gene expression by codons, the insertion or deletion can disrupt the reading frame, or the grouping of the codons, resulting in a completely different translation from the original. The earlier in the sequence the deletion or insertion occurs, the more altered the protein produced is.

**Genome:** the total set of different DNA molecules of an organelle, cell or organism. The human genome consists of 25 different DNA molecules, the mitochondrial DNA molecule plus the 24 different chromosomal DNA molecules.

**Hemizygous:** Having only one copy of a gene or DNA sequence in diploid cells. Males are hemizygous for most genes on the sex chromosomes. Deletions occurring on one autosome produce hemizyosity in males and in female.

**Heterozygote:** an individual having two different alleles at a particular locus.

**Homozygote:** an individual having two identical alleles at a particular locus. For clinical purposes a person is often described as homozygous AA if they have two normally functioning alleles, or homozygous aa if they have two pathogenic alleles at a locus, regardless whether the alleles are in fact completely identical at the DNA sequence level. Homozygosity for alleles identical by descent is called autozygosity.

**Missense mutation:** a nucleotide substitution that results in an amino acid change.



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