

Appendix (1)

Taibah University

College of Applied Medical Science

Medical Laboratory Technology

To Detect Mutations in *ASS1* Gene and Identify Phenotype Variability and Correlate Phenotype with Genotype in Saudi Patients with Citrullinemia Disorder

Study Questionnaire

This questionnaire is designed to obtain personal, medical, and family history information from participants who do or do not have citrullinemia. Thank you.

Patient information:

Name: Gender:

Birth date: ... / ... / City:

Address:

Parent information:

Mobile number: Email:

Marital status:

Single Married Widow divorced

Education:

Read and write Elementary school Intermediate school

High school University graduation Postgraduate.

Job:

Employee Nonemployee Retired

1- Onset of the disease:

2- At first diagnosis, how advanced was the citrullinemia?

- Mild
- Intermediate
- severe

3- Weight: _____

4- Does your weight changes significantly over the years? If yes, write details.

5- Are you committed to a diet? Yes No

6- What is your diet? And how many meals every day?

7- Are you committed to medications? Yes No

8- Names of medications you are taking, and how many times do you take it every day? And in what dose?

9- Do you feel lethargy during the day: yes No

10- Do you have any brain problems or learning difficulties? Name it

11- What are the symptoms that appeared after stopping the treatment or the diet?

12- Have you ever gone to the hospital for hyperammonemia?

Yes No

13- Have you ever needed gastrointestinal tube for lack of appetite?

Yes No

14- Do any of the family members have the disease? If yes write their relative relation to you.

Relative relation	Patient	Carrier

Appendix (2)

Sequences of exons of *ASS1* gene from Ensemble Genome Reference:

Exon 1:

GCTTATAACCTGGGATGGGCACCCCTGCCAGTCCTGCTCTGCCGCTGCCACCGCTGCCCGAGCCC
GGTAAGGAGCCCTCGGCCCTCTCGCTGCCACTCCCTAGCCGAGAGCACCCGCTTCCCGGGCCCA
GAGGAGGAGGCGTAGAAGACGCCCGCCCCGGGGCCCCGCGGAGGCAGAGGGCGGACCCCGATCGC
CACTCGCCGGGACTGGGACCGAGGCAGGAGGGAAGGGGCTCCCGATGCTCAAACGCCTGGCACC
GGATTCCAG

Exon 2:

GACGCTATGTCCAGCAAAGGCTCCGTGGTTCTGGCCTACAGTGGCGGCTGGACACCTCGTGCATC
CTCGTGTGGCTGAAGGAACAAGGCTATGACGTCATTGCCTATCTG

Exon 3:

GCCAACATTGGCCAGAAGGAAGACTTCGAGGAAGCCAGGAAGAAGGCACTGAAGCTTGGGGCCA
AAAAG

Exon 4:

GTGTTCAATTGAGGATGTCAGCAGGGAGTTTGTGGAGGAGTTCATCTGGCCGGCCATCCAGTCCAGC
GCACTGTATGAGGACCGCTACCTCCTGGGCACCTCTCTTGCCAGGCCCTGCATCGCCCCGAAACAA
GTGGAAATCGCCAGCGGGAGGGGGCCAAGTATGTGTCCACGGCGCCACAGGAAAG

Exon 5:

GGGAACGATCAGGTCCGGTTTGAGCTCAGCTGCTACTACTGGCCCCCAGATAAAG

Exon 6:

CCCCCTTCTCACCCCAACACACCACCATTCTGCCATGGAGTGCAGGCTCCGTCCAGCCAGCCCCGG
TGAGGGAGTCTGTGTCCCTCCCCTCCTTCATGGGGCTCCCTCTCACCCCTCACAACAGCATCCTCTCT
GGGGACATGCTGGAGACCCCATGGGCCCTCCAGGAGAGGCCAGCTCTGCAGCTTACAGGCCA
GGGAAGCCCACAGCTCGGCCCTCCCGGCTCTGACCCCTTGTCCTATGTCCAGGTCAATTGCTCCCT
GGAGGATGCCTGAATTCTACAACCGGTTCAAGGGCCGCAATGACCTGATGGAGTACGCAAAG

Exon 7:

CAACACGGGATTCCCATCCCGGTCCTCCCAAGAACCCGTGGAGCATGGATGAGAACCTCATGCA
CATCAG

Exon 8:

CTACGAGGCTGGAATCCTGGAGAACCCCAAG

Exon 9:

CTGCATCAGACGAGAGGCCCATTTTCCAACACGCCCAAGAAAAAATATACATAAAGGTTGATGTG
TTTTCAAATGGGTTTTTGCTTTGCTTCTCTTTTTCTCCCT

Exon 10:

CTTGCTCCTCAAAGTCACTTTTTCTGCCTGACTTGTTCCTCTCCAGCTATTCTACCTCCAGCTG
TGGGGGCGTCGAAGAAAAAGATGAGATCAAGTTGAGGGGAAAAATTGTCTGATTTCTCCAGCCCT
TTGTTTCCCAGGCCTCTGGCAAGCGAGGCTGGTGCTAGGCTGAGGGCTGGGGACCGGGGGATCTG
CCGGACCCACCAGCTGGTGGGGAAATGGACAGAGGAGAGGGGTGCAGATCCCCGCGGGAGGTG
GGCTGTAGGGTGTCCAGGGACTGGTATGTCATCTGCCACCACTTTCTGTCTTTTTTCAGAACCAAG
CGCCTCCAGGTCTCTACACGAAGACCCAGGACCCAGCCAAAGCCCCCAACACCCTGACATTCTCG
AGATCGAGTTCAAAAAAG

Exon 11:

GGGTCCCTGTGAAGGTGACCAACGTCAAGGATGGCACCACCCACCAGACCTCCTTGGAGCTCTTCA
TGTACCTGAACGAAGTCGCGTGAGTGTCTGCAGCCCTGTCCGGCCTCTTGGGAACCGCGTCTCGG
GCGAGCACGAGCCTGGACCGTTGCAGCTAATGGTTGTGGCTGAGGCTCAGGG

Exon 12:

GGGCAAGCATGGCGTGGGCCGTATTGACATCGTGGAGAACCGCTTCATTGGAATGAAGTCCCGAG

Exon 13:

GTATCTACGAGACCCCAGCAGGCACCATCCTTTACCATGCTCATTTAGACATCGAGGCCTTCACCA
TGGACCGGGAAGTGCGCAAAATCAAACAAGGCCTGGGCTTGAAATTTGCTGAGCTGGTGTATACC
G

Exon 14:

GTTTCTGGCACAGCCCTGAGTGTGAATTTGTCCGCCACTGCATCGCCAAGTCCCAGGAGCGAGTGG
AAGGGAAAGTGCAGGTGTCCGTCCTCAAGGGCCAGGTGTACATCCTCGGCCGGGAGTCCCCACTG
TCTCTTACAATGAGGAGCTGGTGAG

Exon 15:

CATGAACGTGCAGGGTGATTATGAGCCAAGTGTATGCCACCGGGTTCATCAACATCAATTCCTCAG

Exon 16:

GCTGAAGGAATATCATCGTCTCCAGAGCAAGGTCACTGCCAAATAGACCCGTGTACAATGAGGAG
CTGGGGCCTCCTCAATTTGCAGATCCCCAAGTACAGGCGCTAATTGTTGTGATAATTTGTAATTGT
GACTTGTCTCCCCGGCTGGCAGCGTAGTGGGGCTGCCAGGCCCCAGCTTTGTTCCCTGGTCCCCCT
GAAGCCTGCAAACGTTGTCATCGAAGGGAAGGTGGGGGGCAGCTGCGGTGGGGAGCTATAAAA
ATGACAATTAAGAGACACTAGTCTTTTATTCTA