Q1.

Leigh syndrome (LS) is a rare, recessive, inherited condition.

LS is caused by a mutation in any one of more than 75 different genes coding for proteins involved in oxidative phosphorylation.

In 80% of people with LS, these mutations occur in nuclear DNA. In 20% of people with LS, these mutations occur in mitochondrial DNA (mtDNA).

15% of the nuclear DNA mutations that cause LS occur in the *SURF1* gene. A mutated *SURF1* gene codes for a shorter polypeptide than a non-mutated *SURF1* gene.

(a) Name **one** type of *SURF1* gene mutation and explain how this mutation could lead to production of a shorter polypeptide.

Type of mutation _____

Explanation _____

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(2)

Globally, the frequency of LS is 1 in 40 000

In the Faroe Islands, which are 18 isolated islands, the frequency of LS is 1 in 1700

(b) The population of the Faroe Islands is 49 053

Estimate the number of people in the Faroe Islands with LS caused by a mutation in the *SURF1* gene.

Use information in this question.

Give your answer to the nearest whole number.

Show your working.

Answer _____ people

(3)

(c) The frequency of LS is higher in the Faroe Islands than globally.

Suggest and explain **one** reason why.

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(2)

(d) LS usually causes death within the first three years of life.

Using all the information in this question, evaluate whether all people should be genetically screened for LS.

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(3)

(Total 10 marks)