Mark schemes

Q1.

(a) **Mark as pairs: 1 and 2 OR 3 and 4**

1. Deletion/translocation;

2. Could mean triplet(s)/codon(s) missing

OR

Could mean amino acid(s) missing (from the polypeptide/SURF1);
Reject could mean an amino acid is not produced

3. Substitution/inversion/addition/duplication/ deletion/translocation;

4. Could result in a (premature) stop triplet/codon;

2 max

(b) Correct answer of 3 (people) = **3 marks**;;;

3.462564706/3.48/3.45 (or any correct rounding down to 1dp) = **2 marks** (answer not to the nearest whole number)

23 = **2 marks** (number of Faroe Islanders with nuclear mutations)

4 = **2 marks** (not factored in that only 80% of mutations are in nuclear DNA)

29 = **1 mark** (number of Faroe Islanders with LS)

3

(c) **Mark as pairs: 1 and 2 OR 3 and 4**

1. Genetic drift;

2. Frequency is higher by chance

OR

High frequency is not due to natural selection;

3. (Only) inbreeding/interbreeding (within a population)

OR

No (inter)breeding with other populations

OR

(Inherited from) common ancestor;
*Accept descriptions of inbreeding **OR** no interbreeding*

Accept reproductively isolated
*Accept genetic bottleneck **OR** founder effect*

4. Low genetic diversity
OR

Small gene pool
OR

Little gene flow
OR

Higher chance of inheriting allele
OR

Frequency of allele higher (in offspring);

2 max

(d) **2 max for mark point 1 to 4 OR 5 to 7**

Yes (no mark)

1. Some people could be heterozygous/carriers;

2. Could prevent (human) suffering/death
OR

Could allow for (informed) decisions about having children;

3. (But only) in families/people with a history of LS
OR

(only) in families/people in the Faroe Islands (where high frequency/1: 1700);

4. Cost of screening might be cheaper than cost of treating LS;

No (no mark)

5. It is rare (globally)
OR

(Only) 1 in 40 000 (globally);

6. Caused by (too) many genes/one of 75 genes
OR

Would need (too) many probes/75 probes;

7. (Too) expensive to produce tests/probes (for more than 75 different genes)

OR

(Too) expensive to screen **all**;

3 max

[10]