

8 June 2023

## Genetic Haemochromatosis

**Please may I make the following request under Freedom of Information Act?**

**For the most recent financial year, please could you confirm:**

- a. The total number of HFE genetic tests for genetic haemochromatosis undertaken by Belfast HSCT**

4267

This number includes 863 samples that were received and processed by the department but not tested due to either not enough clinical details to meet criteria or that a result was already available for the patient.

- b. For the tests completed in (a) the number of results classified by result:**

3404

- |  |     |
|--|-----|
| • C282y homozygous                     | 575 |
| • C282y heterozygous                   | 865 |
| • Compound heterozygous (C282Y + H63D) | 453 |
| • H63D homozygous                      | 131 |
| • H63D heterozygous                    | 578 |
| • Failed test or invalid sample        | 0   |
| • None of the above                    |     |

30 of the above samples had further testing and confirmation of result by PCR Sanger sequence.

The 802 remaining samples did not have either the C282Y or H63D genotype in any of the combinations detailed and are considered to have a normal genotype for these variants.

- c. The number of non-HFE genetic tests for genetic haemochromatosis (for examples, for types 2,3,4) undertaken by Belfast HSCT, by type**

Belfast Trust do not provide a service for this test.