

[REDACTED]

6 October 2016

Dear [REDACTED]

Your email of 1 October 2016

I refer to your request of 1 October 2016 under the Freedom of Information Act 2000. This has been passed to me to deal with. I set out our responses to your questions below:

A. *The total number of genomes currently available within the dataset:* we have sequenced 13971 whole genomes. The figure can be found on our website and is updated frequently.

<https://www.genomicsengland.co.uk/> .

B. *The number of genomes within the dataset that exhibit genetic haemochromatosis (a rare disease which manifests as an autosomal recessive/dominant disorder) by type:* Haemochromatosis is a disorder which is already genetically well characterised. The 100,000 Genomes Project is focusing on disorders where the genetic basis is unknown or only partially understood. For this reason haemochromatosis is not included as a disorder in the project.

We are going to carry out an analysis for a limited number of additional health-related conditions where participants opt in for this. At present haemochromatosis is not on this list because of the difficulty predicting the clinical effects of having the genetic variants in any particular individual, but the list is kept under review and changed as new evidence accumulates.

Because we don't have haemochromatosis as a recruited disorder or an additional finding at this time, we have no information on the frequency of the relevant variants in the database. This analysis may be carried out by researchers in the de-identified database in future but there are no immediate plans to generate this data for any of the project's current activities or aims.

If you have any queries about this letter, please contact me. If you are dissatisfied with the handling of your request, you have the right to ask for an internal review. Internal review requests should be submitted within two months of the date of receipt of the response to your original letter and should be addressed to:

Grant Stapleton
Senior Information Responsible Officer
Genomics England

Email: grant.stapleton@genomicsengland.co.uk

If you are not content with the outcome of your complaint, you may apply directly to the Information Commissioner (ICO) for a decision. Generally, the ICO cannot make a decision unless you have exhausted the complaints procedure provided by the Department. The ICO can be contacted at:

The Information Commissioner's Office
Wycliffe House
Water Lane
Wilmslow
Cheshire
SK9 5AF

Kind regards

Yours sincerely

[Redacted signature]

GENERAL COUNSEL AND COMPANY SECRETARY

[Redacted name]