Many thanks for your considered letter on behalf of the Science and Technology Committee and your helpful comments relating to the National Genomic Medicine Service.

Building on the world leading 100,000 Genomes Project, the UK will enhance its international leadership as one of the first countries in the world to establish a fully integrated Genomic Medicine Service (GMS) from October 2018. Through the GMS, NHS England will provide comprehensive and equitable access to the latest in genomic testing, including Whole Genome Sequencing (WGS), across the whole country.

To deliver the vision that the Chief Medical Officer for England set out in her annual report ‘Generation Genome’, the new GMS will be supported by a network of National Genomic Laboratory Hubs (GLHs) that will deliver an integrated system for genomic testing, working to clear common standards and protocols, supported by, for the first time, a comprehensive national directory of genomic tests for specified cancers and rare diseases that encompasses the entire testing repertoire from WGS to tests for single genes, molecular markers and other functional genomic tests.

The national directory of genomic tests will define situations in which whole genome sequencing has sufficient evidence to be used as a first-line test, alongside situations in which an alternative test should be used, either instead of or before, a whole genome sequence is appropriate for use. I am reassured that Thermo Fisher Scientific are working with Genomics England to pilot innovative new tools for cancer diagnostics.

As the costs for WGS continue to fall, it will increasingly become the most cost-effective first-line diagnostic test of choice for many different clinical conditions,
particularly in the overall assessment of cancers. However, in the immediate term, many existing tests will remain the most appropriate approach – this is reflected in the directory of tests for 2018/19 with only 24 of the 397 rare disease indications and 3 cancer indications to use WGS.

In December 2017 the Life Science Sector Deal announced the sequencing of an additional 50,000 genomes from cancer patients which will be delivered in parallel to the new Genomic Medicine Service through Genomics England and the NHS. This programme will define additional situations in which WGS has sufficient evidence to be used as a first-line test for cancers.

On the issue of creating a potential monopoly among genomic test providers, the national genomic test directory will set out the targets that should be tested for, and the expected methodology for testing but it will not prescribe the technology platforms by which the testing is delivered.

The service specification for the GLHs set out requirements for turnaround times for genomic testing of between 3 and 21 days, depending on the specific clinical indication, as defined by NHS England and in line with for example other NHS waiting time standards. These represent the maximum turnaround times that should be achieved across the whole of the NHS in England.

The new national testing service will be clinically led and NHS England will establish working groups with clinical and scientific leads from the GLHs. These groups will work collaboratively on a number of issues including turnaround times, to ensure the service continues to deliver genomic testing to nationally agreed standards and protocols whilst supporting and developing clinical pathways of care across their geographical areas. I understand that when you met with Lord O’Shaughnessy last month a potential summit on Genomics was proposed. Perhaps there would be some value in exploring the issue of turnaround times of Genomic testing for cancer within just such a forum.

Thermo Fisher have also written to NHS England to convey their concerns and NHS England have offered to meet with them directly to understand and discuss the issues raised. NHS England have also confirmed they are happy to copy their response to the Science and Technology Select Committee.