SeqOne Genomics and the French Thrombotic MicroAngiopathies National reference center (CNR-MAT) pioneer the use of Oxford Nanopore sequencing technology to improve patient outcomes in kidney disease while reducing turnaround times

Montpellier, February 9th, 2023: SeqOne today announced the results of a long-standing research collaboration with Pr. Laurent Mesnard, Co-Director of the French National center for Thrombotic MicroAngiopathies (CNR-MAT), aimed at improving the diagnosis and management of patients suspected of suffering from thrombotic microangiopathies and atypical Hemolytic syndrome (aHUS). The program involved using Oxford Nanopore's revolutionary technology to sequence patients at the bedside, dramatically reducing turnaround times to obtaining actionable insights. Patients were sequenced using Oxford Nanopore's adaptive sampling option that makes it possible to target the parts of the genome of interest for thrombotic microangiopathies **(**TMAs) and modify the read-depth to explore in detail the regions that contain complex mutations that are hard to detect using standard methods. This Oxford Nanopore-based analysis was compared with traditional short read sequencing approaches in order to establish a benchmark against current standards of care and hence quantify benefits of nanopore sequencing. So far, the program, has analyzed 15 patients demonstrating the potential of the approach and highlighting the following benefits when compared with short read sequencing;

- Significantly faster turnaround times of under three days from sample collection to clinical report versus 3-4 week turnaround using outsourced short read sequencing
- Reliable detection of complex variants such as those affecting alternative complement pathways that are often duplicated
- Detection of certain types of variants, notably CFH:CFHR1 hybrid gene mutations that cannot be detected using traditional short-read technology
- A clear indication of those patients who will respond to one of the drugs that have proven effective in treating this disease; vitamin B12 in Cobalamin III deficiency or Eculizumab in case of complement alternate pathway dysregulation associated aHUS.

TMAs, such as aHUS, are rare diseases primarily affecting the kidney. They are hard to diagnose using traditional methods as they present as a number of nonspecific symptoms, such as severe hypertension, acute kidney injury/failure in the context of mechanical anemia and thrombocytopenia. TMAs lesions can be associated with many diseases and some genetic disorders linked to aHUS. Molecular diagnoses, in particular in genes associated with alternative complement pathway regulation, make it possible to identify patients that suffer from this condition as well as informing on possible therapeutic options for TMA/aHUS.

“It is clear that a cost effective, rapid technology that can sequence long fragments of DNA, such as those provided by Oxford Nanopore and a robust interpretation solution such as SeqOne are necessary to extract the full potential of third generation sequencing technology in nephrology and aHUS” said Pr. Laurent Mesnard, the PI heading the study.

In order to support the program, SeqOne implemented an Oxford Nanopore compatible long-read support capable of managing adaptive sampling data. SeqOne is a CE-IVD compliant platform that simplifies the analysis and interpretation of the data enabling the rapid interpretation of patient files which in turn maximizes efficiencies and enables management of higher caseloads.

“SeqOne is convinced both of the clinical potential of Oxford Nanopore’s technology as well as the benefits of bedside sequencing enabled by Oxford Nanopore’s hyper-cost-effective sequencing solutions” said Nicolas Philippe, CEO of SeqOne. “We are investing heavily to provide the best interpretation solution for the data provided in support of this exciting initiative”.

“We are delighted to be part of this research project and to see the potential of nanopore sequencing in nephrology and TMA/aHUS. The technology offers the chance to improve outcomes with significantly faster turnaround times and the ability to identify complex variants.” Commented Dr Emma Stanton, VP Clinical, Oxford Nanopore Technologies.

On the basis of this successful program, SeqOne and CNR-MAT now plan to extend its investigation of the use of nanopore sequencing technology to qualify further patients eligible to actionable (Vitamin B12, Eculizumab) therapy as well as to investigate the impact of other variant types in patient prognosis during TMA/aHUS.

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About SeqOne Genomics

SeqOne Genomics offers high-performance genomic analysis solutions for healthcare providers treating patients suffering from cancer, rare and hereditary diseases as well as pharmaceutical companies developing new therapies. The solution supports both short read and long-read nanopore sequencing and leverages advanced machine learning coupled with the company's proprietary genomics operating system to dramatically reduce turnaround times and costs while delivering comprehensive and actionable insights for personalized medicine. The company has won numerous awards including the iLab award and the ARC cancer foundation's Hélène Stark prize. Investors include Elaia, IRDI Capital Investissement, Merieux Equity Partners, Omnes and Software Club.

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About the French Thrombotic Microangiopathies National reference centre (CNR-MAT)

The Thrombotic Microangiopathies National reference centre (CNR-MAT) is a research network focused on the study of thrombotic microangiopathies (MAT) and associated rare diseases. It was recognized 2006 as a national reference center for France under the stewardship of Paul Coppo (Hematology department, Saint-Antoine Hospital and Sorbonne-University) in Paris. The missions of the CNR-MAT are to promote clinical and basic research in the field of thrombotic microangiopathies through innovative medical approaches, as well as to inform practitioners, patients and all actors involved in the management of these diseases. The CNR-MAT coordinates the healthcare establishments treating patients suffering from these pathologies in France.

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About Oxford Nanopore Technologies

Oxford Nanopore Technologies' goal is to bring the widest benefits to society through enabling the analysis of anything, by anyone, anywhere. The company has developed a new generation of nanopore-based sensing technology for real-time, high-performance, accessible and scalable analysis of DNA and RNA. The technology is used in more than 120 countries to understand the biology of humans and diseases such as cancer, plants, animals, bacteria, viruses and whole environments. Oxford Nanopore Technologies products are intended for molecular biology applications and are not intended for diagnostic purposes. www.nanopore.com

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Forward-looking statements

This announcement contains certain forward-looking statements. Phrases such as "potential", "expect", "intend", "believe we can", "working to", "anticipate", "when validated", and similar expressions of a future or forward-looking nature should also be considered forward-looking statements. Forward-looking statements address our expected future business, and by definition address matters that are, to different degrees, uncertain and may involve factors beyond our control.