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To whom it may concern

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Summary statement on the project "3Gb-TEST"
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written by Clemens R. Müller-Reible on behalf of the Scientific Advisory Board

The 3Gb-TEST project has set out to prepare the genetics community, the medical professionals and the general public for the introduction of Whole Genome Sequencing (WGS) into routine genetic diagnostics.
The technical development of WGS has matured to a level where accuracy, turn-around-times and cost make it a realistic alternative to more conventional methods of genetic analysis. WGS bares the potential of not only replacing targeted gene sequencing but also genetic screening methods such as high-resolution karyotyping by array-based assays. It is now conceivable that a single WGS test at the beginning of life may deliver the entire genetic information of an individual which is relevant for her/his present and future health status.
The introduction of such a powerful analytic technique poses a number of questions which – ideally – should be addressed before its wide application. For the geneticists, important issues – among others – are best practice guidelines, quality control and the reliability of data interpretation. For the medical community, the clinical utility of WGS is the foremost question together with an assessment of its cost effectiveness. For health politics and the general public, a serious health technology assessment and programs of education in genetics are required.

The 3Gb-TEST project aims and deliverables have addressed many of these questions. The project partners have organized five meetings/courses in Leiden (2), Prague, Toulouse and Athens and have presented at the ESHG conferences in Milan (2014) and Glasgow (2015) as well as at regional meetings in Paris, Lund and Berlin thus reaching well over 1.200 professionals from various areas of medicine. The written deliverables include (in addition to the meeting reports) a policy document on WGS (D4.1), a handbook of proposals for the actual implementation of WGS (D5.3), guidelines for the issues and requirements of external quality assessment in WGS (D6.2), a White Paper on WGS for the medical/scientific community (D8.3) and a technology roadmap for a tactical introduction of WGS (D8.4). In these papers, the consortium has addressed not only technical issues but also economic aspects as well as societal and ethical issues and has produced a compendium of proposals and recommendations for practical solutions.

At the final project meeting in Leiden in August 2015, the presentation of a pilot project of WGS impressively demonstrated the full power of WGS in detecting DNA sequence variants and copy number variations at the same time. This study demonstrated that WGS is already a feasible approach as of 2015. The timing of the 3Gb-TEST project thus couldn't have been any better as it has significantly helped to prepare the ground for this technology and to set a framework of points-to-consider for a prudent application of WGS.

It was a privilege and a learning experience to follow the project's achievements which will be relevant in many aspects for the "next generation" of genetic diagnostics.

Prof. Dr. Clemens R. Müller-Reible