

Case Study

Queen Mary University of London-Coeliac Genetics Research Group

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About the study

The Coeliac Genetics Research Group, lead by Professor van Heel, has to date, identified eight gene regions implicated in predisposing an individual to coeliac disease. Interestingly, of a total of nine coeliac gene regions, eight contain likely candidate genes involved in immune system function. In collaboration with colleagues around the world, the Coeliac Genetics Research Group will be collecting and analyzing thousands of DNA samples in search for more genetic risk factors associated with coeliac disease. The aim of the study is to identify and map the majority of inherited genetic risk factors associated with coeliac disease and to understand how these variants are influencing biological function. Based on the previous findings, this study may also advance the understanding of other common immune mediated diseases.

Main challenges

A main challenge of the study was to find a way to access potential study participants that reside around the globe. In order to maximize the number of samples collected, they required an easy, non-invasive and reliable method to collect samples via the post. The key was to find a solution that would maximize donor compliance and minimize both logistical challenges and cost. In addition, the samples must provide high yields of high quality DNA for use on sensitive applications such as the Illumina Golden Gate assay.

...100% concordance with blood vs. Oragene•DNA on the Illumina Golden Gate Assay.

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Collection methods considered

The collection methods considered for the study included collection of venous blood samples versus Oragene•DNA.

Why Oragene•DNA

Oragene•DNA was selected as the preferred sample collection method due to its ease of use and non-invasive approach. The self-collection kits also enabled collection of as much DNA as possible from as many participants as possible because the samples could easily be shipped to donors all around the world. In addition, the Oragene•DNA kits decreased costs by eliminating the need for clinical staff or professionals to collect blood samples. Donors could easily provide a sample and send it back to the researchers by regular mail.

Results

The DNA collected using the Oragene•DNA kits is of high quality and high quantity (typically 100 µg DNA). When compared to DNA from blood, the concordance rates between genotypes obtained by Oragene•DNA are very high (n=4, 99.85% across 1025 SNPs) and resulted in identical genotype cloud positions using the Illumina Golden Gate assay.

