Plain English Summary of Results

There are several forms of diabetes including Type 1 Diabetes and Type 2 Diabetes. It is not always easy for a doctor to be certain what type of diabetes a person has. It is important that the correct type is confirmed as the patient treatment and care pathways are very different. This study aimed to evaluate the performance of a test which provides a genetic risk profile for Type 1 Diabetes (Type 1 Diabetes (T1D) SNP Array EV4489A/B). The genetic component of Type 1 Diabetes is well known and remains unchanged throughout life offering a means to identify individuals who are at a higher genetic risk for this condition helping with disease identification before symptoms arise and improving patient care, treatment and potential outcomes.

Leftover samples from individuals who were non-diabetic (n=100), with early onset Type 1 Diabetes (n=100), adult-onset Type 1 Diabetes (n=100) or Type 2 Diabetes (n=200) were assessed. A genetic profile with a risk score was generated for 498 samples out of the 500 tested. Two failures were due to sample issues. The test was shown to provide the same result as a reference method. All study outcomes were met with the test showing good test performance especially in differentiating Type 1 Diabetes from other forms of diabetes such as Type 2 Diabetes. The test therefore met its intended use and labelling claims.

One limitation of the study however was the low numbers of other ethnic groups assessed. 98% of the study equated to ‘White British, White Irish, Other white’ with only 2% other ethnicities. It is well documented within the test instructions for use that the genetic risk score has been developed and validated in European ancestry populations and caution must be applied to interpreting results in non-European populations and is an ongoing area of research. A further study would be beneficial within other ancestries besides European.