Genotyping for disease association

The Wessex Genomics Laboratory Service (WGLS) is a combined service across two sites at Salisbury and Southampton (formerly Wessex Regional Genetics Laboratory and Molecular Pathology, respectively). The laboratories provide tests for haematological malignancies, solid organ cancers and pharmacogenomics, aligned with the National Genomics Test Directory. In addition, a select list of disease association tests are offered. These tests are not listed on the National Genomics Test Directory (NGTD).

Disease association testing provides genomic information to support a diagnosis or influence treatment. The table below aims to provide information about available genomic testing according to clinical indication.

The below assays test directly from peripheral blood (EDTA) and nucleic acids are not extracted or stored. For additional information on the WGLS policy on the retention and storage of samples, please contact the laboratory.

For any additional information or specific testing request, email wgls_cancergenomics@uhs.nhs.uk (Southampton).

Test indication	Sample type	WGLS Salisbury	WGLS Southampton
Thrombophilia			
	PB		Factor V Leiden and Prothrombin variant (G20210A) *If patient satisfies all criteria for R97.1 (Rare and Inherited disease NGTD) the sample should be sent to Oxford Genetics Laboratories for testing.*
Ankylosing spondylitis			
	PB		*HLA-B**27
Abacavir hypersensitivity			
Please clearly identify high risk specimens	PB		*HLA-B**57:01