NHS
Blood and Transplant

# Immunogenetic markers and diagnosing diseases

## Genetic differences and the immune system

While most genetic information (in other words, DNA) is identical between humans, there are huge differences in a small part of our DNA. Some of these differences are in the genes which control our immune systems. The human leucocyte antigen (HLA) genes are the most varied in humans, and these genes play a vital role in controlling how our immune systems respond to foreign agents such as bacteria and viruses.

HLA genes can show how some people's immune systems can respond more effectively to these foreign agents, as a direct result of their 'HLA type' (also known as 'tissue type'). Unfortunately, some people's immune systems will instead attack their own tissues, leading to diseases such as rheumatoid arthritis and ankylosing spondylitis. Also, people who have particular HLA genes or other genes related to HLA are at an increased risk of developing certain diseases, or developing more aggressive forms of a disease. Identifying these people's HLA types plays a useful role in diagnosing these diseases and can also guide the treatment a patient receives.

## What is HLA typing?

There are many thousands of different tissue types as a result of the differences in our HLA genes. Some of these tissue types are associated with disease including ankylosing spondylitis, Behcet's disease, birdshot chorioretinopathy, coeliac disease, narcolepsy, rheumatoid arthritis and selective IgA deficiency. Different variants of closely related genes are also associated with other diseases such as hereditary haemochromatosis and 21-hydroxylase deficiency. In fact, there are probably several hundred different diseases that are more common in people who have particular tissue types.

We find your HLA type by taking a blood sample and getting your DNA from this. We then analyse this DNA to identify your HLA type and the type of other similar genes. The laboratory will store a sample of your DNA in case we need it to carry out more tests in the future.

We will give the results of your HLA typing (and other gene typing) to your hospital consultant and, in some cases, to your GP. The laboratory will also store all your test results securely.

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# What happens to my sample?

When we no longer need your samples for testing, or we have more samples than we need, the law allows us to use these anonymously for quality control (making sure our tests are working correctly), research (depending on whether this is approved by an ethics committee) or introducing new procedures, or for educating and training doctors, nurses, scientists and other professionals working in healthcare. This helps us maintain accurate testing procedures and improve our knowledge, and so provide the best possible care for all patients.

However, if you do not want us to use your samples for any of the purposes above, you must tell your doctor or the person taking your blood (or both). We will respect your wishes and dispose of any samples we no longer need.

**This patient information leaflet does not replace the guidance provided by your treating clinical team. Your treating clinical team should advise you of the options for treatment, advise of any alternative treatment and associated risks. Your treating clinical team should ensure that you are aware of the material risks associated with the treatment advised.**

**It is the responsibility of the requester submitting your sample, to ensure informed consent has been obtained for all tests, including genetic tests in accordance with current guidance and legislation.**

**If you are unsure about any aspects of the treatment/care, ask your treating clinical team to explain.**

## NHS Blood and Transplant

NHS Blood and Transplant (NHSBT) saves and improves lives by providing a safe, reliable and efficient supply of blood and associated services to the NHS in England and North Wales. We are the organ donor organisation for the UK and are responsible for matching and allocating donated organs. We rely on thousands of members of the public who voluntarily donate their blood, organs, tissues and stem cells.

All information provided to NHS Blood and Transplant is used in accordance with the General Data Protection Regulation (GDPR) and all other applicable privacy legislation. For more information on how we look after your personal details or to find out more about your privacy rights visit www.nhsbt.nhs.uk/privacy or call 0300 123 23 23. NHSBT are committed to keeping your data safe and confidential.

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