**Result letter examples**

**Positive: mutation identified**

The result of your genetic test for Lynch syndrome is now available. A genetic change was identified in a gene called ***MLH1***. This genetic change (or mutation) is considered pathogenic (disease-causing). This confirms your diagnosis of Lynch syndrome, and gives us an explanation of why you developed endometrial cancer. Your medical team will use this information in their medical management decisions, it give you access to personalised therapies, and a personalised surveillance programme.

This also help us to be able to offer your first degree relatives (parents, siblings, and children) a genetic blood test as they have a 50% chance of having the this genetic change, and therefore, a higher risk of developing bowel or other cancers.

For this reason, we have referred you to your local clinical genetics department who will give you an appointment to discuss your result in more detail with you, and facilitate genetic testing and surveillance for your first degree relatives. You should receive an appointment with them over the post in few weeks’ time.

Please continue your follow-up with Dr………... (Oncologist). Please do not hesitate to contact me should you have any clinical questions, or any queries about this result.

**Uncertain negative result: mutation not identified**

The results of your genetic testing to see if you have any genetic changes (mutations) in the genes associated with Lynch syndrome is now available. Analysis shows that you do not have any genetic changes in these genes. A negative result does not change your diagnosis nor does it rule out an inherited condition and we may need to look at other genes.

We have now referred you to your local clinical genetics department so they can ‘virtually’ assess if any further testing is available. They may give you an appointment if they can offer you further testing, or surveillance recommendations for your family.

Please continue your follow-up with Dr………... (Oncologist) . Please do not hesitate to contact me should you have any clinical questions.

**Uncertain result: variant of unknown significance**

The results of your genetic test to see if you have any genetic changes in the genes associated with Lynch syndrome are now available. Analysis has revealed a ***variant of unknown clinical significance*** in the ***MLH1*** gene. What this means is that we are not sure whether this genetic change is disease-causing (pathogenic), or part of your normal DNA variability that do not cause disease. For this reason, we are **unsure** that this could explain why you developed endometrial cancer, and we cannot use it to offer genetic testing to your family members.

We have now referred you to your local clinical genetics department so they can ‘virtually’ assess if any further testing is available. They may give you an appointment if they can offer you further testing, or surveillance recommendations for your family.