NHS
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The POLE and POLD1 Genes

Why have I been given this information sheet?

In recent years two genes have been shown to be linked to cancer and to bowel polyps in some families, called the POLE and POLD1 genes. If there is a pattern of bowel cancer, endometrial cancer, or polyps in your family it may be due to a problem with one of these genes. Usually you will be offered testing of the genes associated with Lynch Syndrome first. If this testing does not find a gene alteration then testing of the POLE and POLD1 genes may be considered.

What are the POLE and POLD1 genes?

POLE and POLD1 are two separate genes. Both are involved in the process of copying DNA in the body. As with all genes, we each inherit one copy of each of these genes from our mother, and one from our father. This means we all have two copies of POLE and two copies of POLD1. However, in some people, an alteration in one of the copies of the gene stops it from working properly and causes that person to have a higher than usual chance of developing certain cancers.

How are POLE and POLD1 gene alterations inherited?

Changes to these genes are passed on in the same way as gene changes associated with Lynch Syndrome. The diagram included in the 'How is Lynch Syndrome inherited?' section of the Lynch Syndrome leaflet shows the way that POLE and POLD1 genes changes can be inherited.

What happens if an alteration to POLE or POLD is found in my family?

This would give an explanation for the pattern of cancer and/or polyps. However, as these genes were only shown to be linked to cancer quite recently, we do not yet know everything about how they can affect the risks of cancer and polyps in a family.

Finding a change to these genes in a family allows us to offer 'predictive' testing to relatives to see if they have inherited this gene change or not. It also allows us to give advice for risk management and screening to relatives based on the current understanding of these genes.

What checks are advised for individuals with a POLE or POLD gene change?

There is not yet clear guidance for what will best manage the risks of cancer for patients with a POLE or POLD1 gene change. If you have a change to one of these genes then recommendations will be made based on the current scientific knowledge and any guidelines that have been published. Your genetic counsellor or doctor will discuss this with you, and is likely to include:

- Colonoscopy surveillance to monitor the bowel for cancer and for polyps. This procedure is described in the Lynch Syndrome leaflet. The age to begin this screening, and how often colonoscopies will be done, will be decided based on the pattern of cancer in the family.
- Womb screening for women. At present the exact risks of endometrial (uterine) cancer are not known, but there is some evidence that suggests carrying a POLE or POLD1 gene change will increase this risk.

No other cancer screening is recommended.

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