# Additional findings conditions and genes

The following conditions will be looked for as part of the additional findings analysis.

# Adults only

## Lynch syndrome

Genes: *MLH1, MSH2, MSH6*

Changes in these genes increase the risk of developing various cancers, most commonly bowel, womb and ovarian. Approximately 1 in 200 people in the general population have one of these gene alterations.

There are other genes that are linked to a risk of bowel cancer that we won’t be looking for.

For more information on genetic causes of bowel cancer and Lynch syndrome, visit the [Bowel Cancer UK website](https://www.bowelcanceruk.org.uk/about-bowel-cancer/risk-factors/family-history/) or the [Genomics Education Programme, part of Health Education England, website](https://www.genomicseducation.hee.nhs.uk/documents/lynch-syndrome/).

## Bowel cancer predisposition

Genes: *MUTYH*, *APC*

Changes in these genes increase the risk of developing bowel polyps and cancer.

For more information on genetic causes of bowel cancer and Lynch syndrome, visit the [Bowel Cancer UK website](https://www.bowelcanceruk.org.uk/about-bowel-cancer/risk-factors/family-history/) or the [Genomics Education Programme website](https://www.genomicseducation.hee.nhs.uk/documents/lynch-syndrome/).

## Breast and ovarian cancer predisposition

Genes: *BRCA1*, *BRCA2*

Alterations in the two *BRCA* genes have been shown to increase the risk of breast cancer, as well as ovarian cancer and prostate cancer. These changes have a 50% chance of being inherited from parent to child. For more information about the *BRCA* genes, visit the [Macmillan website](https://www.macmillan.org.uk/cancer-information-and-support/worried-about-cancer/causes-and-risk-factors/brca-gene) or the Genomics Education Programme website ([*BRCA1*](https://www.genomicseducation.hee.nhs.uk/glossary/brca1/) and [*BRCA2*](https://www.genomicseducation.hee.nhs.uk/glossary/brca2/)).

## Other rare cancer predispositions

Genes: *VHL*, *MEN1*, *RET*

Von Hippel-Lindau disease is a rare condition caused by changes in the *VHL* gene that affects 1 in 36,000 births. Parents with Von Hippel-Lindau disease have a 50% chance of passing it down to their children. Von Hippel-Lindau disease can present with lots of different complications and at various ages, even within the same family. Find out more about Von Hippel-Lindau disease from [VHL UK/Ireland’s website](https://vhl-uk-ireland.org/about/what-is-vhl-disease/) or the [Genomics Education Programme website](https://www.genomicseducation.hee.nhs.uk/documents/von-hippel-lindau-disease/).

Multiple endocrine neoplasia type 1 (MEN1) is a condition, caused by changes in the *MEN1* gene, that can lead to tumours growing in the endocrine glands. Most people inherit the mutated *MEN1* gene from a parent, and there is a 50% chance of a parent passing it down to their children. More information about MEN1 can be found on the [Association for Multiple Endocrine Neoplasia Disorders (AMEND) website](https://www.amend.org.uk/patients/information/men-1/), [Macmillan’s website](https://www.macmillan.org.uk/cancer-information-and-support/worried-about-cancer/pre-cancerous-and-genetic-conditions/multiple-endocrine-neoplasia-1-men1#planning_a_family), or the [Genomics Education Programme website](https://www.genomicseducation.hee.nhs.uk/documents/multiple-endocrine-neoplasia-type-1/).

Changes in the *RET* gene significantly increases the risk in medullary thyroid cancer. Parents with an altered *RET* gene have a 50% chance of passing this down to their children. For more information, visit the [Genomics Education Programme website](https://www.genomicseducation.hee.nhs.uk/documents/familial-medullary-thyroid-cancer/).

## Familial hypercholesterolaemia

Genes: *LDLR*, *APOB*, *PCSK9*

Familial hypercholesterolaemia is a condition that causes high cholesterol, which can lead to early heart attacks and heart disease. Changes in any of the three genes we are looking for can cause familial hypercholesterolaemia because they remove cholesterol from the blood. People who have changes in one of these genes have a 50% chance of passing it down to their children. Find out more about familial hypercholesterolaemia and high cholesterol on the [British Heart Foundation website](https://www.bhf.org.uk/informationsupport/conditions/familial-hypercholesterolaemia), [Heart UK’s website](https://www.heartuk.org.uk/cholesterol/what-is-fh), or the [Genomics Education Programme website](https://www.genomicseducation.hee.nhs.uk/documents/familial-hypercholesterolaemia/).

## Carrier status

Some gene alterations have no effect on the individual who has them, but may affect future children. If someone has such an alteration, they are said to be a ‘carrier’. At the moment we are only looking for one of these conditions – cystic fibrosis. For a child to have this condition both parents will be carriers. We will therefore only look for alterations in the cystic fibrosis gene if both partners in a couple took part in the Project and both asked us to look for this.

## Cystic Fibrosis

Gene: *CFTR*

Cystic fibrosis is an inherited condition caused by changes in the *CFTR* gene, and affects 1 in 2,000 to 1 in 3,000 babies of Northern European ancestry. It is less common in other ethnic groups. Between 1 in 22 and 1 in 27 people in the UK are carriers. Read more about cystic fibrosis on the [Genomics Education Programme website](https://www.genomicseducation.hee.nhs.uk/documents/cystic-fibrosis/).

## Adults and Children

For children whose parents have asked for additional findings, we are looking at a very small number of genes where there is something that could be done to prevent illness in a child.

## Bowel cancer predisposition

Gene: *APC*

Changes in this gene increases the risk of developing bowel polyps and cancer.

For more information on genetic causes of bowel cancer and Lynch syndrome, visit the [Bowel Cancer UK website](https://www.bowelcanceruk.org.uk/about-bowel-cancer/risk-factors/family-history/) or the [Genomics Education Programme website](https://www.genomicseducation.hee.nhs.uk/documents/lynch-syndrome/).

## Other rare cancer predispositions

Genes: *VHL, MEN1, RET*

Von Hippel-Lindau disease is a rare condition caused by changes in the *VHL* gene that affects 1 in 36,000 births. Find out more about Von Hippel-Lindau disease from [VHL UK/Ireland’s website](https://vhl-uk-ireland.org/about/what-is-vhl-disease/) or the [Genomics Education Programme website](https://www.genomicseducation.hee.nhs.uk/documents/von-hippel-lindau-disease/).

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Changes in the *RET* gene significantly increases the risk in medullary thyroid cancer. For more information, visit the [Genomics Education Programme website](https://www.genomicseducation.hee.nhs.uk/documents/familial-medullary-thyroid-cancer/).

## Familial hypercholesterolaemia

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