Genetic Testing In Lynch Syndrome

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There are three types of tests in Lynch syndrome; diagnostic, predictive, and pre-implantation genetic diagnosis.

1) Diagnostic. This test is offered to someone who might have Lynch syndrome (LS) because they have had cancer. Usually they are the first person in their family to be diagnosed with LS. Diagnostic testing is often done in two steps. Firstly, a test is done on a stored sample of the individual's tumour. Sometimes obtaining the sample takes several months. This sample is tested in the lab to check for signs of LS.

Nowadays, this test is usually requested by a colorectal team soon after someone is diagnosed with colon cancer. In addition a dermatologist may request tumour testing for sebaceous adenomas at the point of diagnosis.

The test can take a few months to be processed in the lab. If such changes are found in the tumour, the individual is asked to provide a blood sample for a genetic test; which is the second step to this process.

This test looks for a genetic change (technically known as a mutation) in a LS gene or genes. The genetic test can take a few months. A genetic test is most often offered by regional genetic centres.

A) When a mutation is found, a genetic counsellor or clinical geneticist will discuss the implications of this test and agree on a screening a management plan with you. They will refer you to have colonoscopies and discuss gynaecological surgery if you wish.

B) Sometimes, these tests are done and no sign of LS or no mutation is found. This means that person is unlikely to have LS. Your genetics team may suggest extra bowel screening for family members. Having a "negative" test does not exclude a genetic

diagnosis. You may be offered further genetic tests or you can request a review after a few years as advances in genetic testing could introduce more tests for you and the family.

2) Predictive. This test is offered to unaffected relatives of individuals with LS. This test can be offered by regional genetic services. If you have a relative with LS, you can ask your GP to refer you to your regional genetics service.

It would be helpful for your genetics specialist if you had information such as your relative's name, date of birth, address or clinical genetics reference number in hand.

A predictive genetic test appointment usually takes 45 minutes to 1 hour. Some centres in the UK prefer to offer two appointments for the purpose of predictive genetic testing. Patients may also ask for more appointments if they wish to take more time to consider their options.

Once a blood sample is drawn it may take up to 4-6 weeks to have a result. This type of test can only be offered to adults. This is because LS affects individuals in adulthood (except in very rare circumstances) and also has practical implications. If a relative has been affected with an LS related tumour at a very young age, testing may be considered for a young person under the age of 18 for the consideration of early screening.

Here is a link to The British Society Of Genomic Medicine statement on genetic testing for children:

http://www.bsgm.org.uk/media/678741/gtoc_booklet_final_new.pdf

3) Tests for pregnancies

A) Preimplantation Genetic diagnosis: This test can be offered to couples hoping to have (more) children, when one partner is an LS carrier. PGD can help stop the mutation from being passed on the next generation, by offering a predictive genetic test to very young embryos produced by IVF techniques. The embryos that do not

have the familial mutation, are implanted following IVF protocols or saved for future use. This process can be requested by your genetic counsellor.

You may find more information here: http://www.hfea.gov.uk/preimplantation-genetic-diagnosis.html

B) Prenatal diagnosis: This test can be offered during the pregnancy to check if the fetus is carrying the familial alteration. Such a test is usually offered to couples who wish to terminate an affected pregnancy. As LS is a condition that does not affect children and can be managed well during adulthood with screening and management options, not all obstetricians will agree to offer it.