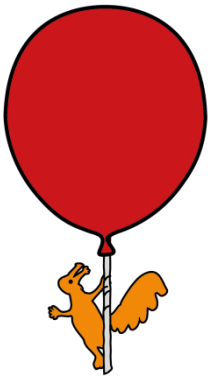


Alder Hey Children's NHS Foundation Trust

Ophthalmology Department

NGS Genetic Eye Disorders Testing

Information for parents

What is NGS Testing for Genetic Eye Disorders?

'NGS' stands for 'Next Generation Sequencing'. This is a new method of looking for changes in genes (genetic testing). Inherited eye disorders can be caused by changes in many different genes.

In the past genetic testing involved analysing one gene at a time. However new NGS technology allows us to sequence (read the genetic code) for many genes at the same time. This means that it is more likely that we can find the gene change(s) responsible for your child's eye condition.

How is the test done?

The test is done in the Genetic Medicine Laboratory in Manchester, using DNA (genetic information) taken from your / your child's blood sample. The sequence of the DNA (genetic code) is read on an automated machine to look for changes (like spelling mistakes) in the eye genes.

Will the test find the cause of my / my child's eye condidtion?

The test may or may not find the cause of your / your child's eye condition. As the tests are improving, we are finding the genetic cause in many more cases. However, in some cases the genetic cause is not found, and in other cases, a gene is found that is difficult to interpret.

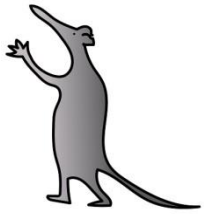
Will the test find changes in other genes?

A number of people in the population are carriers of changes in genes for genetic eye conditions. Since these changes are common, it is possible that the test may find a change in a gene that is not the cause of your / your child's eye condition. In a very small number of cases, it is also possible that the test may show a change in a gene that can have, in addition to the eye condition, other impacts on health. This information may also have implications for the wider family.

What are the benefits of having the test?

Finding the gene change(s) responsible for your / your child's eye condition can be helpful for several reasons:

- It can confirm your / your child's diagnosis.
- It tells us about the way in wich your / your child's eye condition is inherited, which helps us to work out the implications for family members.
- In the future, it may be helpful to know about the gene change, in terms of research ot if therapies beome available.



How long will it take to get results?

It may take 6-9 months to complete the test.

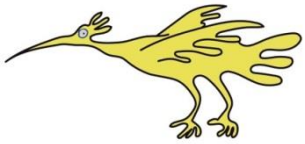
How will I find out the results?

We can explain your results at a clinic appointment, by telephone, or by letter. Your doctor or genetic counsellor will discuss this with you, to decide which option is best.

How can I find out more?

If you have questions about the chances of passing on the condition to future children or the changes of other people developing the eye condition in your family, you can take this information sheet to your GP and ask for a referral to your local genetic counsellor.

Referrals should be sent to:



Genetic Medicine
6th Floor, St Mary's Hospital
Oxford Road
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References

Content provided by the University of Manchester.
MCGM: Clinical Genetics Service ◆ Genomic Diagnostics Laboratory ◆ Willink Biochemical Genetics Unit ◆ Bioinformatics Group ◆ University of Manchester Genetic Medicine ◆ NIHR Genetics Clinical Trials Unit.
Partners: MAHSC Genomic Diagnostics and Innovation ◆ NGRL ◆ Nowgen ◆ EMQN ◆ ERNDIM

This leaflet only gives general information. You must always discuss the individual treatment of your child with the appropriate member of staff. Do not rely on this leaflet alone for information about your child's treatment.

This information can be made available in other languages and formats if requested.

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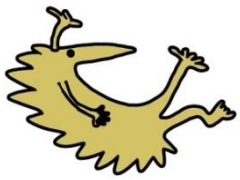

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