Glucokinase (GCK) gene testing in pregnancy

NHS South East Genomic Medicine Service

You have been diagnosed with gestational diabetes (GDM) because you have been found to have a higher blood sugar (glucose) level in pregnancy.

Fewer than 2 in 100 women originally diagnosed with GDM will in fact have a change in the *Glucokinase* (*GCK*) gene which is causing their raised blood sugar.

Genes provide instructions to help our bodies develop, grow and work. Genes are inherited from our parents. Changes in the *GCK* gene can therefore be inherited, which means the change can be passed on from parent to child.

A change in the *GCK* gene causes mildly raised blood sugar, which is **not diabetes,** this is called glucokinase hyperglycaemia.

This raised blood sugar is often incorrectly diagnosed as diabetes, including GDM. Although it causes a slightly raised blood sugar level, it is not known to cause long term health problems. Outside of pregnancy people with a change in the *GCK* gene do not need specialist care or follow up. People with a change in the *GCK* gene have **no greater risk** of developing Type 2 diabetes than other members of the general population.

A Change in the *GCK* gene is more likely if you have:

- a high fasting Glucose Tolerance Test (GTT) result or several high fasting (pre breakfast) blood glucose results on home monitoring

and

- a Body Mass Index (BMI) of <30 kg/m^2 if white or <27 kg/m^2, if from an ethnic group with a high risk of type 2 diabetes

You can now be offered a blood test to look for a change in your *GCK* gene. This is available via the NHS and is free of charge.

What could the GCK gene test result be- three possible results?

1. **GCK gene variant not identified** (a negative result) - in most cases a change in the GCK gene will not be found and you will continue with recommended care for gestational diabetes.
2. **GCK gene variant identified** (glucokinase hyperglycaemia) - this is known to cause a raised blood glucose level.
3. **Variant of unknown significance** (VUS) - in extremely small numbers of cases, there is a change in the GCK gene which has been identified, but it is unclear if it is connected to the raised blood glucose level. If this happens, we will ask for more information about other family members to help understand if the gene change was the cause or not.

Why are you being offered a test for changes in the *GCK* gene in pregnancy?

If a change in the *GCK* gene is identified it can make a difference to you, your baby and possibly other family members. Treatment in pregnancy depends on whether your baby has inherited the same gene change. If you have a change in the *GCK* gene your baby has a 1 in 2 (50%) chance of also having the same gene change. In some cases, a specialised blood test can be offered during pregnancy to find out if your baby has inherited the gene change. This test works by analysing tiny fragments of the baby's DNA that naturally circulate in the pregnant person's bloodstream.

Do I have to have the test?

No, having any form of testing in pregnancy is a personal choice and one which only you can make. Your health care team will be happy to discuss any questions about the test that you have. You can then decide, based on your own circumstances, whether it is right for you.

Can my family be tested?

If you are diagnosed with a change in your *GCK* gene, your family **do not** need testing unless they have **already** been diagnosed with d

Further information can be found at:

DiabetesGenes

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It is not intended to replace clinical guidance or expertise.

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