NHS East Genomics

R142 genetic test form completion:

Testing for glucokinase hyperglycaemia during pregnancy

This guide aims to assist clinicians with completing the form and facilitate testing.

The test request form for monogenic diabetes is available at www.diabetesgenes.org and is used for both the **R141 and R142** tests.

Some aspects of the form are not required when testing for glucokinase hyperglycaemia within the gestational diabetes pathway. **Mandatory fields are highlighted in yellow.**

Patient Details

All aspects of this section should be completed, ***excluding*** invoice address and genetic diabetes nurse. Please fill in the 'currently pregnant and gestation and date sample taken'.

www.exeterlaboratory.com
www.diabetesgenes.org
+44(0)1392 408229
rduh.exetergenomicslaboratory@nhs.net

(FOR LAB USE ONLY)
EX Number:

Royal Devon and Exeter NHS
NHS Foundation Trust

Genetic Testing for Monogenic Diabetes
National Genomic Test Directory For Clinical Indication R141 & R142

*Please send **EDTA** whole blood (minimum 5ml adults; 3ml children; 1ml neonates) or DNA (minimum of 5µg) direct to:*
Exeter Genomics Laboratory, RILD Level 3, Royal Devon & Exeter Hospital, Barrack Road, Exeter EX2 5DW
Clinical Scientist Lead: Kevin Colclough (rduh.betacellgenomics@nhs.net)

Patient details **Please complete form electronically, e-mail to rduh.exetergenomicslaboratory@nhs.net and send a printed copy with the samples**

SURNAME:	CLINICIAN NAME:	
FORENAME:	CLINICIAN TELEPHONE:	
D.O.B.: (DD/MM/YYYY)	CLINICIAN E-MAIL ADDRESS (reports can be issued as PDFs to @nhs.net accounts and to non-UK clinicians):	
PATIENT POSTCODE:	CLINICIAN ADDRESS:	
NHS NUMBER (HOSPITAL/PATIENT ID IF NON-UK):	INVOICE ADDRESS: NA	
SEX:	ETHNIC ORIGIN:	GENETIC DIABETES NURSE: NA

Is this patient currently pregnant: **Yes** **Gestation (weeks)** **No**	**Date Sample Taken: (DD/MM/YYYY)**

Consent

The patient agreement to the 1st line on diagnosis is mandatory for testing and this box should be ticked. The rest of the consent is optional to the patient, and is based on samples and information being used for research purposes.

Consent

We understand that our samples and clinical information will be used only for diagnostic and research purposes relevant to ourselves and others in my family. Please tick: ☐

We also consent for our samples and clinical information to be saved in the Genetic Beta Cell Bank for use in future research into all forms of genetic diabetes and other beta cell conditions, whether or not it is of direct clinical benefit to us. Tick here for consent: ☐

We are also happy to be contacted about research into genetic diabetes and you may contact me directly using these details: Tick here for consent: ☐

Name: Address: Telephone: E-mail:

I confirm: I am the patient. I am signing this form on behalf of someone else (children, adults without capacity or deceased patients).
I am the healthcare professional recording the patient's choices and consent has been recorded remotely, no patient signature.

Name of patient/guardian/advocate	**Electronic Signature:**	**Date:**

For more information (and patient information sheets) please see https://www.diabetesgenes.org/current-research/genetic-beta-cell-research-bank

R142 genetic test form completion:
Testing for glucokinase hyperglycaemia during pregnancy

NHS East Genomics

Clinical Information

Essential fields are shown below (age at diagnosis, diagnosed during pregnancy, height, weight, current or booking BMI, laboratory FBG or OGTT result and date, current HbA1c).

Clinical information

MODY PROBABILTY CALCULATOR SCORE: % (https://www.diabetesgenes.org/mody-probability-calculator)
AGE AT DIAGNOSIS:
INSULIN TREATED WITHIN 6 MONTHS OF DIAGNOSIS? YES ☐ NO ☐
DIAGNOSED DURING PREGNANCY? ☐
HEIGHT (METERS):
WEIGHT (KILOGRAMS):
BMI AT DIAGNOSIS:
CURRENT BMI:
FATHER'S BMI:
MOTHER'S BMI:

INITIAL THERAPY:	INSULIN SUBTYPE :	UNITS PER DOSE	INSULIN FREQUENCY:	CURRENT THERAPY:	INSULIN SUBTYPE :	UNITS PER DOSE	INSULIN FREQUENCY:
	OHA SUBTYPE :	UNITS PER DOSE	OHA FREQUENCY:		OHA SUBTYPE :	UNITS PER DOSE	OHA FREQUENCY:

ACANTHOSIS NIGRICANS? ☐ SENSITIVE TO SULPHONYLUREA? ☐ RENAL DISEASE? ☐ RENAL CYSTS? ☐ RENAL DYSPLASIA OR AGENESIS? ☐ LOW RENAL THRESHOLD FOR GLUCOSE? ☐

PARTIAL LIPODYSTROPHY? ☐ DEAFNESS? ☐ LIVER ADENOMA? ☐ NEONATAL HYPOGLYCAEMIA? ☐ DETAILS AND DURATION OF NEONATAL HYPOGLYCAEMIA TREATMENT:

FBG OR OGTT 0 HOUR RESULT:	OGTT 2 HOUR RESULT:	OGTT DATE:	C-PEPTIDE (pmol/l):	CURRENT HBA1C (mmol/mol):
PREVIOUS FBG OR OGTT 0 HOUR RESULT:	PREVIOUS OGTT 2 HOUR RESULT:	PREVIOUS OGTT DATE:	DATE OF C-PEPTIDE:	HIGHEST RECORDED HBA1C (mmol/mol):

GAD POSITIVE? ☐ GAD NEGATIVE? ☐	GAD RESULT:	IA-2 POSITIVE? ☐ IA-2 NEGATIVE? ☐	IA-2 RESULT:	ZnT8 POSITIVE? ☐ ZnT8 NEGATIVE? ☐	ZnT8 RESULT:	UCPCR (nmol/mmol):
BIRTH WEIGHT (GRAMS):	GESTATION:	DIABETIC COMPLICATIONS, OR ANY OTHER CLINICAL FEATURES:				

Family History

Please indicate if any other family members are known to have diabetes (please tick any / all that apply and add age of diagnosis and treatment as indicated). Family history of renal disease and deafness not applicable.

Family history

DIABETIC FATHER'S FATHER? ☐ DIABETIC FATHER'S MOTHER? ☐ DIABETIC MOTHER'S FATHER? ☐ DIABETIC MOTHER'S MOTHER? ☐

TOTAL NUMBER OF SIBLINGS: NUMBER OF SIBLINGS WITH DIABETES:
TOTAL NUMBER OF CHILDREN: NUMBER OF CHILDREN WITH DIABETES:

DIABETIC FATHER? ☐ AGE AT DIAGNOSIS? TREATMENT:
DIABETIC MOTHER? ☐ AGE AT DIAGNOSIS?: TREATMENT

PLEASE ADD THE AGE OF DIAGNOSIS FOR SIBLINGS WITH DIABETES:
SIBLING 1: SIBLING 2: SIBLING 3: SIBLING 4:

PLEASE ADD THE AGE OF DIAGNOSIS FOR CHILDREN WITH DIABETES:
CHILD 1: CHILD 2: CHILD 3: CHILD 4:

FAMILY HISTORY OF RENAL DISEASE (CYSTS, PROTEINURIA, RENAL FAILURE, RENAL DYSPLASIA, RENAL AGENESIS)? IF YES PLEASE ADD TO FAMILY HISTORY DETAILS: ☐ NA

FAMILY HISTORY OF DEAFNESS? IF YES PLEASE PLEASE ADD TO FAMILY HISTORY DETAILS: ☐ NA

FAMILY HISTORY DETAILS/COMMENTS: SUCH AS OTHER DIABETIC RELATIVES? (AGE AT DIAGNOSIS AND CURRENT TREATMENT OF AFFECTED FAMILY MEMBERS WOULD BE VERY HELPFUL):

IF SAMPLES FROM OTHER FAMILY MEMBERS HAVE BEEN SENT PREVIOUSLY PLEASE GIVE DETAILS:

Testing Required

Select GCK Sanger sequencing as indicated below.

Testing required If no boxes are ticked, testing will be performed according to the clinical information provided

Please visit our website for current test costs (www.diabetesgenes.org)

GCK Sanger sequencing (this method will not detect partial/whole gene deletions and duplications) ☐

m.3243A>G test for maternally inherited diabetes and deafness (MIDD) ☐

Next generation sequencing 34 gene test for monogenic diabetes; includes all MODY genes, MIDD and partial lipodystrophy (this method can also detect partial/whole gene deletions and duplications) ☐

For further information about this test please see: https://www.diabetesgenes.org/tests-for-diabetes-subtypes/a-new-test-for-all-mody-genes/

KNOWN VARIANT TEST (FOR FAMILIES WHERE A VARIANT HAS ALREADY BEEN IDENTIFIED) ☐

Gene: Variant: Name and date of birth of relative with variant: Relationship to this person:

Version No.: MG/MON/FOR014.03

DiabetesGenes

Additional Information for Clinicians can be found at www.diabetesgenes.org