**www.exeterlaboratory.com**

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**Genetic testing for neonatal diabetes**

**National Genomic Test Directory for Clinical Indication R143**

[www.england.nhs.uk](https://www.england.nhs.uk/wp-content/uploads/2018/08/Rare-and-inherited-disease-eligibility-criteria-2021-22-v2.pdf)



*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: Jayne Houghton ([rduh.betacellgenomics@nhs.net](mailto:rduh.betacellgenomics@nhs.net) )

**Please complete form electronically, e-mail to** [**rduh.exetergenomicslaboratory@nhs.net**](mailto:rduh.exetergenomicslaboratory@nhs.net) **and send a printed copy with the samples**

For clinical advice please contact Prof Andrew Hattersley by e-mail [a.t.hattersley@exeter.ac.uk](mailto:a.t.hattersley@exeter.ac.uk)

**Patient details**

|  |  |
| --- | --- |
| Surname: | Clinician(s) name(s): |
| Forename: | Clinician(s) e-mail address for report: |
| D.O.B. (dd/mm/yyyy): | Hospital/City/Country: |
| NHS number (hospital/patient id if non-uk) |
| Sex: | Invoice address: |
| Ethnic origin: | GP Practice Details: |

**Consent**

1. 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
2. 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. Please Tick: Yes No
3. We are also happy to be contacted about research into genetic diabetes and you may contact me directly at:   
   Name:       Address:       Telephone:       E-mail:

**Signed by patient/ guardian/advocate: ……………………………………. Date: ……………..………**

For more information (and patient information sheets) please see [www.diabetesgenes.org/content/genetic-beta-cell-research-bank](http://www.diabetesgenes.org/content/genetic-beta-cell-research-bank)

**Parent details**

|  |  |  |
| --- | --- | --- |
| Mother’s surname: | Mother’s forename: | Mother’s d.o.b.: |
| Father’s surname: | Father’s forename: | Father’s d.o.b.: |
| **Names of other family members sent and their relationship to proband:** | | |

**Clinical information**

|  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- |
| Date of diagnosis (dd/mm/yy): | Birth weight (g): | | | glucose at presentation (mmol/l):    DKA (Yes/No)? | |
| Gestation (weeks): | | |
| HbA1c at diagnosis: | Current HbA1c: | Current height (cm): | | Current weight (kg): | |
| GAD antibodies titre: | IA2 antibodies titre: | ICA antibodies titre: | | IAA antibodies titre: | |
| Insulin dose at presentation (U/KG/day): | Current treatment and dose: | | | C-peptide (pmol/l): | Fasting? |
| Post feed? |
| Diabetes remission? | Date of remission: | | Diabetes relapse? | | Date of relapse: |
| Muscle weakness? | Developmental delay? | | Epilepsy? | | Macroglossia? |
| Umbilical hernia? | Kidney disease? | | Skeletal abnormalities? | | Abnormal LFTs? |
| Exocrine pancreatic treatment?  (date started) | Biochemical evidence of malabsorption? | | Pancreatic hypoplasia? | | Anaemia? |
| Thyroid dysfunction? | Cardiac defects? | | Facial dysmorphism? | | |
| Further details/other features including **any imaging results** (a separate document with full details of any additional medical problems would be very helpful): | | | | | |

**Family history**

|  |  |
| --- | --- |
| Are parents related? if yes, how? | |
| Diabetic father? (age diagnosed, treatment): | Diabetic mother? (age diagnosed, treatment): |
| Diabetic sibling(s)? (age diagnosed, treatment): | Other Diabetes family members |

## Testing required

*For further information about testing options and current test costs for* ***non-NHS England*** *referrals please visit our website (*[*www.diabetesgenes.org*](http://www.diabetesgenes.org)*)*

|  |
| --- |
| * **Urgent Testing:*****KCNJ11* and *ABCC8***Report issued in 1-2 weeks followed by neonatal diabetes gene panel next generation sequencing test if no variant found. |
| * Next generation sequencing 35 gene test for monogenic neonatal diabetes (this method can also detect partial/whole gene deletions and duplications) |
| * Methylation analysis of the 6q24 transient neonatal diabetes locus by MS-MLPA: |
| * **Known variant test** (for families where a variant has already been identified)   Gene:       Variant:       Name and date of birth (DD/MM/YYYY) of relative with variant:       Relationship to this person: |