

## Genetic testing for neonatal diabetes

Genetic testing is provided free of charge for any patient diagnosed before 9 months through funding from the Wellcome Trust.

Samples must be labelled with name and date of birth, please send either

(1) Our preferred option is 3-5 mls blood taken in tubes containing EDTA and transported fresh (not frozen) at room temperature to arrive in the UK within 5 days. Blood samples should be sent in leak-proof packaging and include absorbent material to absorb any leakage OR

(2) Send 2-10 micrograms of DNA (to allow repeats) at room temperature. Again please make sure the tube is very securely sealed. Additional DNA will help us to test for further causes of neonatal diabetes.

Please include samples from both parents whenever possible – whether affected or unaffected.

Please fill in this form electronically, e-mail to [E.De-Franco@exeter.ac.uk](mailto:E.De-Franco@exeter.ac.uk) and send a printed copy with the blood/DNA samples to: Prof Sian Ellard, Department of Molecular Genetics, RILD Level 3, Royal Devon and Exeter NHS Foundation Trust, Barrack Road, Exeter, EX2 5DW, UK

For clinical advice please contact Prof Andrew Hattersley by e-mail [a.t.hattersley@exeter.ac.uk](mailto:a.t.hattersley@exeter.ac.uk) or telephone +44 1392 408260

### Patient details

SURNAME:	CLINICIAN NAME:
FORENAME:	CLINICIAN E-MAIL ADDRESS FOR REPORT:
D.O.B. (DD/MM/YYYY):	HOSPITAL:
NHS/CHI NUMBER (for UK patients):	
GENDER:	CITY:
ETHNIC ORIGIN:	COUNTRY:

### Parent details

MOTHER'S SURNAME:	MOTHER'S FORENAME:	MOTHER'S D.O.B.:
FATHER'S SURNAME:	FATHER'S FORENAME:	FATHER'S D.O.B.:

### Clinical information

DATE OF DIAGNOSIS (DD/MM/YY):	BIRTH WEIGHT (g):	GLUCOSE AT PRESENTATION (mmol/l):	
	GESTATION (WEEKS):		
GAD ANTIBODIES TESTED?:	IA2 ANTIBODIES TESTED?:	ICA ANTIBODIES TESTED?:	IAA ANTIBODIES TESTED?:
GAD TITRE:	IA2 TITRE:	ICA TITRE:	IAA TITRE:
PRESENT AGE:	PRESENT HEIGHT (cm):	PRESENT WEIGHT (kg):	
INSULIN DOSE AT PRESENTATION (U/kg/day):	CURRENT INSULIN (U/kg/day):	HBA1C:	DATE HBA1C MEASURED:
		HBA1C NORMAL RANGE:	
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 FAMILY MEMBERS (A PEDIGREE SHOWING 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:			
IF A MUTATION HAS ALREADY BEEN IDENTIFIED IN A FAMILY MEMBER PLEASE GIVE DETAILS:			
Gene	Mutation	Name and date of birth of relative with mutation:	Relationship to this person