

Electronic supplementary material

ESM Table 1 Phenotypic data from non-diabetic probands and accessible relatives of patients with the N1206S, N655Y and N785K *TBC1D4* variants

Characteristic	Genotype									
	N1206S				N655Y				N785K	
	Proband	Reference range/mean (95% CI ^a)	Mother	Reference range/mean (95% CI ^a)	Proband	Reference range/mean (95% CI ^a)	Mother	Reference range/mean (95% CI ^a)	Proband	Reference range/mean (95% CI ^a)
Age (years)	16		47		21		49		16	
Sex	F		F		F		F		F	
BMI (kg/m ²)	28.5		25		45		34		33.5	
BMI SD score	+2.1		+1		+3.9		+2.7		+2.9	
Fasting insulin (pmol/l)	58	55 (52–59)	48	55 (52–59)	231	101 (66–137)	55	72 (65–78)	212	72 (65–78)
Peak insulin (pmol/l)	5,220	415 (389–414)	506	415 (389–414)	1680	730 (392–1070)	717	532 (474–590)	2463	532 (474–590)
Ratio of peak to fasting insulin	90	8.5 (7.9–9)	10.5	8.5 (7.9–9)	7.3	7.3 (5–9.7)	13	8 (7.3–8.7)	11.6	8 (7.3–8.7)
Fasting glucose (mmol/l)	4.9	4.9 (4.9–5.0)	4.3	4.9 (4.9–5.0)	3.4	5.2 (4.8–5.6)	5	5 (4.9–5.1)	5.9	5 (4.9–5.1)
Peak glucose (mmol/l)	10.2	7.5 (7.3–7.8)	8	7.5 (7.3–7.8)	8	8.2 (6.5–10)	9.8	8.1 (7.7–8.5)	4.0	8.1 (7.7–8.5)
Cholesterol (mmol/l)	5.4	<5	4.3	<5	3.6	<5	6.7	<5	–	–
HDL (mmol/l)	1.65	<1	1.28	<1	0.96	<1	1.04	<1	–	–
Triacylglycerol (mmol/l)	2.9	<3	1.06	<3	1.2	<3	1.6	<3	–	–

^aData derived from OGTT data from BMI- and sex-matched controls from the Ely study [6]

BMI range 25–30 kg/m² (*n*=337), 30–35 kg/m² (*n*=126), ≥45 kg/m² (*n*=13)

F, female; M, male