

Clinical characteristics of five patients carrying novel-CEL variants

CEL Gene Transcript	Mutation site	Pathogenicity prediction	Clinical diagnosis	Gender	Age of disease onset	Age of diagnosis	Clinical manifestations	Family history	Diabetic ketoacidosis (DKA)	Metabolic Syndrome (MetS)	Insulin therapy	Height (cm)	Weight (kg)	HbA1c (g/mol)	Diabetes autoantibodies	Insulin (μ U/dL) (1.6-26.9)	C-peptide (ng/dL) (1.1-4.4)	HbA1c (%) (6-9)	F _{1c} (mmol/L) (0.60-1.00)	TC (mmol/L) (3.1-5.2)	LDL (mmol/L) (0-3.36)	HDL (mmol/L) (1-1.5)
NM_00807.4	Ex.10.1140	Likely pathogenic	NDM	Male	31	31	hypoglycaemia, hyperlipidaemia, polyphagia, ketosis	His mother has diabetes and his mother, sister and daughter have hypoglycaemia	No	Yes	No	170	70	21.61	No	0.24	1.09	6.7	0.84	4.83	2.10	0.86
NM_00807.4	c.8942C>T p.C298F	Uncertain significance	T1DM	Male	1	1	hypoglycaemia, polyphagia, polyuria, ketonuria, weight loss	No	Yes	No	Yes	170	8.0kg	17	ICL, GADA(+)	0.1	0.14	6.7	0.69	4.82	2.70	1.07
NM_00807.4	c.2398_GCCAGGGGGAGCTC>GAGGGCTC	Uncertain significance	NDM	Male	12	12	polyphagia, polyuria, fatty urine, weight loss	His mother														