Supplementary Table 2. Summary of all CEL gene variants reported in HGMD and our study

Reference	Mutation type	Mutation sites	Exon	Diabetes type	Exocrine dysfunction
[36]	Missense variant	c.1A>G (p.M1V)	E1	MODY	/
[37]	Missense variant	c.7C>T (p.R3C)	E1	MODY	Yes
[37]	Missense variant	c.61G>T (p.A21S)	E1	MODY	Yes
[38]	Missense variant	c.62C>T (p.A21V)	E1	MODY	Yes
[39]	Missense variant	c.166G>A (p.A56T)	E2	T1DM	/
[39]	Missense variant	c.209G>A (p.G70D)	E2	T1DM	/
[40]	Nonsense variant	c.285C>G (p.Y95*)	E3	MODY	/
[38]	Missense variant	c.296A>C (p.D99A)	E3	MODY	Yes
[41]	Missense variant	c.314T>C (p.I105T)	E3	T1DM	/
[37]	Nonsense variant	c.337C>T (p.Q113*)	E3	MODY	Yes
[37]	Missense variant	c.353T>G (p.L118R)	E4	MODY	Yes
[39]	Missense variant	c.404A>G (p.H135R)	E4	T1DM	/
[37]	Missense variant	c.466G>A (p.V156I)	E4	MODY	Yes
[38]	Missense variant	c.472G>A (p.V158M)	E4	MODY	Yes
[36]	Missense variant	c.655A>G (p.S219G)	E5	MODY	/
Shakhtshneider, 2017	Missense variant	c.731T>C (p.L244P)	E6	MODY	/
[38]	Missense variant	c.787A>G (p.K263E)	E7	MODY	Yes

[39]	Missense variant	c.830G>A (p.C277Y)	E7	T1DM	/
Shakhtshneider, 2017	Missense variant	c.886G>T (p.G296C)	E7	MODY	/
[37]	Missense variant	c.928G>A (p.V310I)	E8	MODY	Yes
[39]	Missense variant	c.985G>A (p.D329N)	E8	T1DM	/
[42]	Missense variant	c.1226C>T (p.T409I)	E9	MODY	Yes
[43]	Missense variant	c.1412C>T (p.T471M)	E10	MODY	Yes
[44]	Missense variant	c.1454T>C (p.I485T)	E10	MODY	Yes
[37]	Missense variant	c.1468A>C (p.N490H)	E10	MODY	Yes
[41]	Missense variant	c.1477A>C (p.K493Q)	E10	T1DM	/
[37]	Missense variant	c.1493A>C (p.N498T)	E11	MODY	Yes
[39]	Missense variant	c.1556T>C (p.L519P)	E11	T1DM	/
[43]	Missense variant	c.1618C>T (p.R540C)	E11	MODY	No
[45]	Missense variant	c.1810G>C (p.V604L)	E11	MODY	No
[46]	Missense variant	c.1966G>C (p.A656P)	E11	T1DM	/
[45]	Missense variant	c.1975G>C (p.V659L)	E11	MODY	/
[44]	Missense variant	c.2110A>C (p.T704P)	E11	MODY	Yes
[44]	Missense variant	c.2129A>G (p.E710G)	E11	MODY	Yes
[44]	Missense variant	c.2131A>G (p.T711A)	E11	MODY	Yes
[44]	Missense variant	c.2134G>C (p.A712P)	E11	MODY	Yes
[44]	Missense variant	c.2143C>A (p.P715T)	E11	MODY	Yes

[37]	Missense variant	c.2164G>C (p.A722P)	E11	MODY	Yes
[39]	Missense variant	c.2222A>G (p.D741G)	E11	T1DM	/
[41]	Splicing variant	c.670-1G>A	/	T1DM	/
[39]	Splicing variant	c.778-3C>A	/	MODY	Yes
[15]	Small deletions	c.137_138delCT (p.(Ser46Cysfs*52)	/	MODY	No
[41]	Small deletions	c.361_362delAT (p.(Met121Aspfs*2	/	T1DM	/
[8]	Small deletions	c.363delG (p.(Met121Ilefs*74	/	T2DM	/
[6]	Small deletions	c.1677delT (p.(Val560Cysfs*1	/	MODY	Yes
[6]	Small deletions	c.1776delC (p.(Val593Cysfs*1	/	MODY	Yes
[37]	Small deletions	c.1809delC (p.(Val604Cysfs*1	/	MODY	Yes
[37]	Small deletions	c.1941delC (p.(Val648Cysfs*5	/	MODY	Yes
[37]	Small deletions	c.1974delC (p.(Val659Cysfs*4	/	MODY	Yes
[37]	Small deletions	c.2098delG (p.(Ala700Profs*4)	/	MODY	Yes
[37]	Small deletions	c.2214delC (p.(Thr739Glnfs*19	/	MODY	Yes
[37]	Small insertions	c.1785dupC (p.(Thr596Hisfs*3)	/	MODY	Yes
[37]	Small insertions	c.1983dupC (p.(Thr662Hisfs*3)	/	MODY	Yes
[37]	Small insertions	c.2016dupC (p.(Thr673Hisfs*3)	/	MODY	Yes
[44]	Small insertions	c.2030_2031insC (p.(Pro678Alafs*9)	/	MODY	Yes
[37]	Small insertions	c.2040dupC (p.(Val681Argfs*6)	/	MODY	Yes
[37]	Small insertions	c.2049dupC (p.(Thr684Hisfs*3)	/	MODY	Yes

[37]	Small insertions	c.2082dupC (p.(Thr695Hisfs*3)	/	MODY	Yes
[47]	Gross deletions	c.1733_2161del429 (p.(Glu578_Ser720	/	MODY	/
[48]	Gross deletions	c.2049_2082del34 (p.(Thr684Argfs*9)	/	MODY	/
[48]	Gross deletions	c.2184_2216del33 (p.(Gly729_Thr739	/	MODY	/
[49]	Complex rearrangement	CEL-CELP duplication hybrid	/	Chronic pancreatit	Yes
[50]	VNTR repeat variants	Proline-rich repeat	/	LDL cholester	Yes
[22]	VNTR repeat variants	33bp VNTR	/	MODY	Yes