Appendix

Table A1 Demographic and clinical description of SIR patients

	Sex	Year of birth	Ethnic origin	Diagnosis	BMI	A/N	C/H	M/H	L/A
1	F	1967	Asian	HAIR-AN		Y	Y	N	N
2	F	1962	Asian	IR-DM	28.8	N	Y	N	N
3	F	1951	European	PA		Y	Y	Y	N
4	F	1964	European	Type A	24.1	Y	Y	N	N
5	F	1974	European	Type A	30.2	Y	Y	N	N
6	F	1956	European	IR-DM	34.9	N	N	N	N
7	F	1951	European	IR-DM	29.4	N	N	N	N
8	F	1950	European	HAIR-AN?	36.9	Y	Y	N	N
9	F	1924	European	Lipodystrophy	28.2	N	N	Y	Y
10	F	1939	European	HAIR-AN?		Y	Y	N	N
11	F	1972	Asian	Type A?		Y			
12	M	1975	European	MIR-AN	26.7	Y	N	N	N
13	M	1975	Asian	PA	46.3	Y	N		
14	F	1970	European	Type A					
15	F	1973	Afro-Carribean	HAIR-AN?	47.5	Y	Y	N	N
16	F	1976	European	Type A	31.7	Y	Y	N	
17	F	1960	European	HAIR-AN	31.6	Y	Y	N	N
18	F	1968		Lipodystrophy	24.8	Y	Y	Y	Y
19	F	1948	Afro-Carribean	Lipodystrophy	25	Y	N	Y	Y
20	F	1958	Iranian	HAIR-AN	34.5	Y	Y	N	N
21	F	1974	Asian	Type A	14.9	N	Y	N	Y
22	F	1975	Asian	HAIR-AN	33.5	Y	Y	N	N
23	F	1958	European	Type A		Y	Y	N	N
24	M			Type A					
25	F	1982	Somali	Type A	15.8	Y	N	N	Y?

Appendix

	Sex	Year of birth	Ethnic origin	Diagnosis	BMI	A/N	C/H	M/H	L/A
26	F	1963	European	HAIR-AN	41.2	Y	Y	N	N
27		1976	Cypriot	MIR-AN		Y			
28	F	1980	European	InsRes/short stature		Y			
29	F	1969	European	HAIR-AN	37	Y		N	N
30	F	1959		HAIR-AN		Y	Y		
31	F	1974	Asian	HAIR-AN?		Y	Y	N	N
32	F	1975	Asian	HAIR-AN	30.2	Y	Y	N	N
33	M			MIR-AN					
34	F	1947	European	Type A?		Y?	Y	N	N
35	F	1971	European	PA?	37.3	N	Y?	Y?	N
36	F	1991	European	Leprechaunism?					
37	F	1961	Mixed	HAIR-AN?	47.2	Y	Y	N	N
38	F	1980	Mixed	Type A?	27.5	Y	N	N	
39	F	1959	European	HAIR-AN?	30.7	Y	Y	N	N
40	F	1977	Asian	HAIR-AN	31.5	Y	Y	?	N
41	F	1943	European	Partial lipodystrophy	27.1	N	N	N	Y
42	F	1956		Lipodystrophy	30.6	N	Y	N	N
43	F	1944	European	IR-DM	32.6	Y	Y	N	N
44	F	1965	European	HAIR-AN	41.8	Y	N	N	N
45	F	1975	Asian	HAIR-AN	46.7	Y	N	N	N
46	M	1976		PA	35.8	Y			
47	F	1973	Asian	HAIR-AN	39.7	Y	Y	N	N
48	M	1974	Mixed	MIR-AN	48.9	Y		N	N
49		1977	Asian						
50	F	1964	European	HAIR-AN	34.8	N	Y	N	N
51	F	1972	Somali	InsRes/short stature	17.3	Y	Y	N	N
52	F	1972		Type A?	39.9	Y			

	Sex	Year of birth	Ethnic origin	Diagnosis	BMI	A/N	C/H	M/H	L/A
53	F	1976	European	HAIR-AN	37	Y	Y		N
54	F	1975	European	HAIR-AN	42.5	Y	N	N	N
55	M	1972	European	IR-DM	23	N			
56	F	1974		Lipodystrophy	21.3	N	N	N	Y
57	F	1958	European	HAIR-AN					
58	F	1977	European	HAIR-AN	31.5	Y	Y	Y	
59	F	1957	European	HAIR-AN	35	Y	Y	N	
60	F	1976	Asian	HAIR-AN	>47.4	N	Y		N
61	M	1990	European						
62	F	1954	Asian	HAIR-AN	36.3	Y		N	N
63	F	1979		Type II CMT	27	Y			
64	F	1959	Asian	Type A	22.5	Y	N		N
65	F	1953	European	HAIR-AN	38.5	N	Y	N	N
66	F	1982	European	PA?	28.2	Y	N	N	N
67	F	1975	Asian?	Type A		Y	Y	N	N
68	F	1978		Nerve deafness & diabetes					
69				Werner Syndrome					
70	M	1974	European		23.6	Y	N	Y	
71	F	1982	European	Type A?	33.4	Y	N	N	N
72	F	1974	European	HAIR-AN	49.6	Y	Y	N	N
73	M	1970	European		28.4	N		N	N
74	F	1954	European	HAIR-AN		Y	Y	N	N
75									
76	F	1980	Mixed	PA	30.7	N	N	N	N
77	F	1976	Asian	InsRes/short stature	28	Y	N	N	N
78	M	1982	Mauritius	MIR-AN	29	Y	N	N	N
79	F		Arabic	PA	17.3	Y	Y	Y	
80	F		Arabic	PA		Y	N	N	N

	Sex	Year of birth	Ethnic origin	Diagnosis	BMI	A/N	C/H	M/H	L/A
81	F	1973		Type A?	37.2	Y			
82	M	1985	European	MIR-AN	29.4	Y		N	N
83	F	1939	European	HAIR-AN	32.1	Y	N	Y	N
84	F	1968	European	IR-DM	33.1	Y	Y		Y?
85				HAIR-AN					
86	F			HAIR-AN		Y			
87	M	1982		InsRes/short stature		Y			
88	F	1976			37	Y			
89	F	1986	European		35.7	Y		N	N
90	F	1985	European	HAIR-AN	41.7	Y	N	N	N
91	M	1977	European	MOPDII (Osteodyplastic primordial dwarfism of Majewski type 2)	29	Y		N	N
92	F			OB-IR		Y			
93	F	1936	European	HAIR-AN	45.6	Y	Y		N
94	F	1980			30.1	N	Y	N	Y
95	F	1967	European	PA	38.9	N			
96	F	1964		HAIR-AN	35.8	Y	Y	N	N
97		1984	Asian	Type A	19.7	Y	N	Y	Y
98	M	1943							
99	F		European	OB-IR	34.6	Y	N	N	N
100	F	1988	European	InsRes/short stature		Y	N	N	Y?
101	F	1905	Jewish	PA	25.5	Y	N		N
102				Leprechaunism					
103	F	1988	European		21.1	Y	N	N	N
104				IR-DM (Type 1)					
105	F	1968	European	IR-DM	22.7	Y	Y	N	N
106	M	1960		MIR-AN					
107	M		European						
108	F	1959	European	HAIR-AN	42.3	Y	Y	Y	N

	Sex	Date of birth	Ethnic origin	Diagnosis	BMI	A/N	C/H	M/H	L/A
109	F	1974	European	OB-IR	39.6	N		N	N
110	F	1986	European		40.1	Y	?	N	?
111	F	1983	Irish	Type A	17.5	Y		N	N
112	M	1951		IR-DM	49.3				
113	F	1969		OB-IR	51.9	Y	N	N	N
114	F	1987	European	HAIR-AN		Y	Y	N	N
115	F	1989	European	Hemihypertrophy					
116	F	1983	European	HAIR-AN	31	Y	Y	N	N
117	F	1983	European		23.6	N	Y	N	N
118	M	1981		OB-IR	47.6	Y			
119	F	1959	European	PA? (originally Type A)	47.3	Y			
120	F	1987	European	HAIR-AN	32	Y	N	N	N
121	F	1956		Lipodystrophy					
122	F	1994	European	RM		Y	N	N	N
123	F	1973	Asian (Pakistani)		25.2	N	N	N	N
124	F	1990	European	Type A?	16	Y	Y	N	N
125	F	1980		Lipodystrophy (partial)	28				
126	F	1983		Lipodystrophy (partial)		Y			
127	M	1963	European	Lipodystrophy (partial)					
128	F	1940		Lipodystrophy (partial)					
129	F	1976		Lipodystrophy	29.2	Y?			Y
130	F	1991		Type A					
131	F	1989	Asian	HAIR-AN	30.3	Y	Y	N	N
132	F	1970	Brazilian	IR-DM	29				
133	M	1990		Leprechaunism					
134	F	1997	Asian (Indonesian)						
135	F	1956	From Azores	Lipodystrophy (partial)					
136	F	1983	European	Type A	24.5	Y	Y		N

	Sex	Year of birth	Ethnic origin	Diagnosis	BMI	A/N	C/H	M/H	L/A
137	F	1987	European	Type A	29	Y	N	?	N
138	F	1951	European	HAIR-AN	38.7	N	N	Y	Y
139	F	1989	Asian	Complex syndrome including severe insulin resistance		Y	Y	N	N
140	F	1990	European	Lipodystrophy (partial)?	23.9	Y mild		Y	Y partial
141	F	1977	Asian	HAIR-AN	36	Y	Y	N	N
142	F	1947	Cauc/Afro Caribbean	Lipodystrophy partial?	29.5	Y			Y
143	F	1983	European	HAIR-AN		Y			
144	F	1981	European	Lipodystrophy		Y	Y		Y partial
145	F	1975	European	HAIR-AN		Y	Y	N	N
146	F	1983		Lipodystrophy partial	22.9	Y			
147	M	1985		Lipodystrophy complex	18.1	Y			
148	M	1986	Asian	IR-DM	24.1	Y		Y	N
149	F	1937	European	Lipodystrophy partial	25.4	N	N	N	Y
150	F	1951	European	Lipodystrophy partial (peripheral)	45.1	?	N	?	Y
151	F	1976	European	Lipodystrophy partial	24.1	Y	Y	Y	Y
152	F	1975	European	Lipodystrophy partial	28.4	Y	Y	Y	
153	F	1981	Asian	Partial lipodystrophy	29.6	Y	Y	Y	Y
154	F	1979	Asian	Type A	20.4	Y	N	N	N
155	F	1984	European	Type A		Y	N	N	N
156	M	1988	European	MIR-AN	21.9	Y		N	N
157	F	1989	Asian Bengali	Type A?	21.8	Y			
158	M	1975	European	MIR-AN	25.2	Y		N	N

A/N = acanthosis nigricans, C/H = clinical hyperandrogenism, M/H = muscular hypertrophy , L/A = lipoatrophy. HAIR-AN = hyperandrogenism, insulin resistance, and acanthosis nigricans. IR-DM = insulin resistance and diabetes mellitus. PA = pseudoacromegaly. MIR-AN = male insulin resistance and acanthosis nigricans. OB-IR = obesity and insulin resistance. RM = Rabson-Mendenhall.

Table A2 CEPH samples

SampleID	Sex	Family	Relationship	HapMap CEU?
NA06985	Female	1341-14	maternal grandmother	Yes
NA06993	Male	1341-13	maternal grandfather	Yes
NA06994	Male	1340-9	paternal grandfather	Yes
NA07000	Female	1340-10	paternal grandmother	Yes
NA07022	Male	1340-11	maternal grandfather	Yes
NA07034	Male	1341-11	paternal grandfather	Yes
NA07055	Female	1341-12	paternal grandmother	Yes
NA07056	Female	1340-12	maternal grandmother	Yes
NA07345	Female	1345-13	maternal grandmother	Yes
NA11881	Male	1347-14	maternal grandfather	Yes
NA11882	Female	1347-15	maternal grandmother	Yes
NA11992	Male	1362-13	paternal grandfather	Yes
NA11993	Female	1362-14	paternal grandmother	Yes
NA11994	Male	1362-15	maternal grandfather	Yes
NA11995	Female	1362-16	maternal grandmother	Yes
NA12003	Male	1420-9	paternal grandfather	Yes
NA12004	Female	1420-10	paternal grandmother	Yes
NA12005	Male	1420-11	maternal grandfather	Yes
NA12006	Female	1420-12	maternal grandmother	Yes
NA12043	Male	1346-11	paternal grandfather	Yes
NA12044	Female	1346-12	paternal grandmother	Yes
NA12144	Male	1334-10	paternal grandfather	Yes
NA12145	Female	1334-11	paternal grandmother	Yes
NA12146	Male	1334-12	maternal grandfather	Yes
NA12154	Male	1408-10	paternal grandfather	Yes
NA12155	Male	1408-12	maternal grandfather	Yes
NA12156	Female	1408-13	maternal grandmother	Yes
NA12236	Female	1408-11	paternal grandmother	Yes
NA12239	Female	1334-13	maternal grandmother	Yes
NA12248	Male	1416-11	paternal grandfather	Yes
NA12249	Female	1416-12	paternal grandmother	Yes
NA07007	Male	1331-12	paternal grandfather	No
NA07340	Female	1331-13	paternal grandmother	No
NA07016	Male	1331-14	maternal grandfather	No
NA07050	Female	1331-15	maternal grandmother	No
NA07049	Male	1333-11	paternal grandfather	No
NA07002	Female	1333-12	paternal grandmother	No
NA07017	Male	1333-13	maternal grandfather	No
NA07341	Female	1333-14	maternal grandmother	No
NA12045	Male	1346-13	maternal grandfather	No
NA11879	Male	1347-12	paternal grandfather	No
NA11880	Female	1347-13	paternal grandmother	No
NA12250	Male	1416-13	maternal grandfather	No
NA12251	Female	1416-14	maternal grandmother	No
NA11917	Male	1423-11	paternal grandfather	No
NA11918	Female	1423-12	paternal grandmother	No
NA11919	Male	1423-13	maternal grandfather	No
NA11920	Female	1423-14	maternal grandmother	No

Table A3 HGDP-CEPH Human Genome Diversity panel

Sample ID	Sex	Population	Geographic origin
HGDP01253	M	Mozabite	Algeria (Mzab)
HGDP01254	F	Mozabite	Algeria (Mzab)
HGDP01255	M	Mozabite	Algeria (Mzab)
HGDP01256	M	Mozabite	Algeria (Mzab)
HGDP01257	M	Mozabite	Algeria (Mzab)
HGDP01258	M	Mozabite	Algeria (Mzab)
HGDP01259	M	Mozabite	Algeria (Mzab)
HGDP01260	M	Mozabite	Algeria (Mzab)
HGDP01261	M	Mozabite	Algeria (Mzab)
HGDP01262	M	Mozabite	Algeria (Mzab)
HGDP01263	M	Mozabite	Algeria (Mzab)
HGDP01264	M	Mozabite	Algeria (Mzab)
HGDP01265	M	Mozabite	Algeria (Mzab)
HGDP01266	M	Mozabite	Algeria (Mzab)
HGDP01267	F	Mozabite	Algeria (Mzab)
HGDP01268	M	Mozabite	Algeria (Mzab)
HGDP01269	M	Mozabite	Algeria (Mzab)
HGDP01270	F	Mozabite	Algeria (Mzab)
HGDP01271	M	Mozabite	Algeria (Mzab)
HGDP01272	M	Mozabite	Algeria (Mzab)
HGDP01273	F	Mozabite	Algeria (Mzab)
HGDP01274	F	Mozabite	Algeria (Mzab)
HGDP01275	F	Mozabite	Algeria (Mzab)
HGDP01276	F	Mozabite	Algeria (Mzab)
HGDP01277	F	Mozabite	Algeria (Mzab)
HGDP01278	M	Mozabite	Algeria (Mzab)
HGDP01279	M	Mozabite	Algeria (Mzab)
HGDP01280	F	Mozabite	Algeria (Mzab)
HGDP01281	F	Mozabite	Algeria (Mzab)
HGDP01282	M	Mozabite	Algeria (Mzab)
HGDP00490	M	NAN Melanesian	Bougainville
HGDP00491	M	NAN Melanesian	Bougainville
HGDP00655	M	NAN Melanesian	Bougainville
HGDP00656	F	NAN Melanesian	Bougainville
HGDP00657	F	NAN Melanesian	Bougainville
HGDP00658	F	NAN Melanesian	Bougainville
HGDP00659	F	NAN Melanesian	Bougainville
HGDP00660	F	NAN Melanesian	Bougainville
HGDP00661	F	NAN Melanesian	Bougainville
HGDP00662	M	NAN Melanesian	Bougainville
HGDP00663	F	NAN Melanesian	Bougainville
HGDP00664	F	NAN Melanesian	Bougainville
HGDP00787	F	NAN Melanesian	Bougainville
HGDP00788	M	NAN Melanesian	Bougainville
HGDP00789	M	NAN Melanesian	Bougainville
HGDP00823	M	NAN Melanesian	Bougainville
HGDP00824	M	NAN Melanesian	Bougainville
HGDP00825	F	NAN Melanesian	Bougainville
HGDP00826	F	NAN Melanesian	Bougainville

Sample ID	Sex	Population	Geographic origin
HGDP00978	F	NAN Melanesian	Bougainville
HGDP00979	F	NAN Melanesian	Bougainville
HGDP01027	F	NAN Melanesian	Bougainville
HGDP00995	F	Karitiana	Brazil
HGDP00996	F	Karitiana	Brazil
HGDP00997	M	Karitiana	Brazil
HGDP00998	M	Karitiana	Brazil
HGDP00999	F	Karitiana	Brazil
HGDP01000	M	Karitiana	Brazil
HGDP01001	F	Karitiana	Brazil
HGDP01003	F	Karitiana	Brazil
HGDP01004	M	Karitiana	Brazil
HGDP01005	M	Karitiana	Brazil
HGDP01006	F	Karitiana	Brazil
HGDP01007	F	Karitiana	Brazil
HGDP01008	F	Karitiana	Brazil
HGDP01009	M	Karitiana	Brazil
HGDP01010	F	Karitiana	Brazil
HGDP01011	F	Karitiana	Brazil
HGDP01012	M	Karitiana	Brazil
HGDP01013	M	Karitiana	Brazil
HGDP01014	F	Karitiana	Brazil
HGDP01015	M	Karitiana	Brazil
HGDP01016	F	Karitiana	Brazil
HGDP01017	F	Karitiana	Brazil
HGDP01018	F	Karitiana	Brazil
HGDP01019	M	Karitiana	Brazil
HGDP00830	F	Surui	Brazil
HGDP00832	F	Surui	Brazil
HGDP00833	F	Surui	Brazil
HGDP00834	M	Surui	Brazil
HGDP00835	M	Surui	Brazil
HGDP00837	M	Surui	Brazil
HGDP00838	F	Surui	Brazil
HGDP00839	M	Surui	Brazil
HGDP00840	F	Surui	Brazil
HGDP00841	F	Surui	Brazil
HGDP00842	M	Surui	Brazil
HGDP00843	M	Surui	Brazil
HGDP00844	M	Surui	Brazil
HGDP00845	M	Surui	Brazil
HGDP00846	F	Surui	Brazil
HGDP00847	M	Surui	Brazil
HGDP00848	F	Surui	Brazil
HGDP00849	M	Surui	Brazil
HGDP00850	F	Surui	Brazil
HGDP00851	M	Surui	Brazil
HGDP00852	F	Surui	Brazil
HGDP00711	M	Cambodian	Cambodia
HGDP00712	F	Cambodian	Cambodia
HGDP00713	F	Cambodian	Cambodia

Sample ID	Sex	Population	Geographic origin
HGDP00714	M	Cambodian	Cambodia
HGDP00715	M	Cambodian	Cambodia
HGDP00716	M	Cambodian	Cambodia
HGDP00717	M	Cambodian	Cambodia
HGDP00718	F	Cambodian	Cambodia
HGDP00719	F	Cambodian	Cambodia
HGDP00720	M	Cambodian	Cambodia
HGDP00721	F	Cambodian	Cambodia
HGDP00448	M	Biaka Pygmies	Central African Republic
HGDP00461	M	Biaka Pygmies	Central African Republic
HGDP00464	M	Biaka Pygmies	Central African Republic
HGDP00465	M	Biaka Pygmies	Central African Republic
HGDP00466	M	Biaka Pygmies	Central African Republic
HGDP00469	M	Biaka Pygmies	Central African Republic
HGDP00470	M	Biaka Pygmies	Central African Republic
HGDP00472	M	Biaka Pygmies	Central African Republic
HGDP00473	M	Biaka Pygmies	Central African Republic
HGDP00475	M	Biaka Pygmies	Central African Republic
HGDP00477	M	Biaka Pygmies	Central African Republic
HGDP00451	M	Biaka Pygmies	Central African Republic
HGDP00452	M	Biaka Pygmies	Central African Republic
HGDP00453	M	Biaka Pygmies	Central African Republic
HGDP00454	M	Biaka Pygmies	Central African Republic
HGDP00455	M	Biaka Pygmies	Central African Republic
HGDP00457	M	Biaka Pygmies	Central African Republic
HGDP00458	M	Biaka Pygmies	Central African Republic
HGDP00459	M	Biaka Pygmies	Central African Republic
HGDP00460	M	Biaka Pygmies	Central African Republic
HGDP00479	M	Biaka Pygmies	Central African Republic
HGDP00980	F	Biaka Pygmies	Central African Republic
HGDP00981	M	Biaka Pygmies	Central African Republic
HGDP00985	M	Biaka Pygmies	Central African Republic
HGDP00986	M	Biaka Pygmies	Central African Republic
HGDP01084	F	Biaka Pygmies	Central African Republic
HGDP01085	F	Biaka Pygmies	Central African Republic
HGDP01086	M	Biaka Pygmies	Central African Republic
HGDP01087	M	Biaka Pygmies	Central African Republic
HGDP01088	M	Biaka Pygmies	Central African Republic
HGDP01089	M	Biaka Pygmies	Central African Republic
HGDP01090	M	Biaka Pygmies	Central African Republic
HGDP01091	M	Biaka Pygmies	Central African Republic
HGDP01092	M	Biaka Pygmies	Central African Republic
HGDP01093	M	Biaka Pygmies	Central African Republic
HGDP01094	M	Biaka Pygmies	Central African Republic
HGDP01307	M	Dai	China
HGDP01308	M	Dai	China
HGDP01309	M	Dai	China
HGDP01310	M	Dai	China
HGDP01311	M	Dai	China
HGDP01312	M	Dai	China
HGDP01313	M	Dai	China

Sample ID	Sex	Population	Geographic origin
HGDP01314	F	Dai	China
HGDP01315	F	Dai	China
HGDP01316	F	Dai	China
HGDP01213	M	Daur	China
HGDP01214	M	Daur	China
HGDP01215	F	Daur	China
HGDP01216	M	Daur	China
HGDP01217	M	Daur	China
HGDP01218	M	Daur	China
HGDP01219	F	Daur	China
HGDP01220	M	Daur	China
HGDP01221	M	Daur	China
HGDP01222	F	Daur	China
HGDP00774	M	Han	China
HGDP00775	M	Han	China
HGDP00776	F	Han	China
HGDP00777	M	Han	China
HGDP00778	M	Han	China
HGDP00779	M	Han	China
HGDP00780	M	Han	China
HGDP00781	F	Han	China
HGDP00782	M	Han	China
HGDP00783	F	Han	China
HGDP00784	F	Han	China
HGDP00785	M	Han	China
HGDP00786	M	Han	China
HGDP00811	F	Han	China
HGDP00812	F	Han	China
HGDP00813	F	Han	China
HGDP00814	F	Han	China
HGDP00815	M	Han	China
HGDP00817	F	Han	China
HGDP00818	F	Han	China
HGDP00819	M	Han	China
HGDP00820	F	Han	China
HGDP00821	M	Han	China
HGDP00822	M	Han	China
HGDP00971	M	Han	China
HGDP00972	F	Han	China
HGDP00973	M	Han	China
HGDP00974	F	Han	China
HGDP00975	F	Han	China
HGDP00976	F	Han	China
HGDP00977	M	Han	China
HGDP01021	F	Han	China
HGDP01022	F	Han	China
HGDP01023	F	Han	China
HGDP01024	F	Han	China
HGDP01287	F	Han	China
HGDP01288	M	Han	China
HGDP01289	M	Han	China

Sample ID	Sex	Population	Geographic origin
HGDP01290	M	Han	China
HGDP01291	F	Han	China
HGDP01292	M	Han	China
HGDP01293	M	Han	China
HGDP01294	M	Han	China
HGDP01295	M	Han	China
HGDP01296	M	Han	China
HGDP01233	M	Hezhen	China
HGDP01234	F	Hezhen	China
HGDP01235	M	Hezhen	China
HGDP01236	M	Hezhen	China
HGDP01237	M	Hezhen	China
HGDP01238	F	Hezhen	China
HGDP01239	F	Hezhen	China
HGDP01240	M	Hezhen	China
HGDP01241	M	Hezhen	China
HGDP01242	F	Hezhen	China
HGDP01317	M	Lahu	China
HGDP01318	M	Lahu	China
HGDP01319	M	Lahu	China
HGDP01320	M	Lahu	China
HGDP01321	M	Lahu	China
HGDP01322	M	Lahu	China
HGDP01323	F	Lahu	China
HGDP01324	F	Lahu	China
HGDP01325	F	Lahu	China
HGDP01326	M	Lahu	China
HGDP01189	M	Miaozu	China
HGDP01190	M	Miaozu	China
HGDP01191	M	Miaozu	China
HGDP01192	M	Miaozu	China
HGDP01193	M	Miaozu	China
HGDP01194	M	Miaozu	China
HGDP01195	M	Miaozu	China
HGDP01196	F	Miaozu	China
HGDP01197	F	Miaozu	China
HGDP01198	F	Miaozu	China
HGDP01223	F	Mongola	China
HGDP01224	M	Mongola	China
HGDP01225	M	Mongola	China
HGDP01226	M	Mongola	China
HGDP01227	M	Mongola	China
HGDP01228	M	Mongola	China
HGDP01229	M	Mongola	China
HGDP01230	M	Mongola	China
HGDP01231	F	Mongola	China
HGDP01232	F	Mongola	China
HGDP01337	M	Naxi	China
HGDP01338	M	Naxi	China
HGDP01339	M	Naxi	China
HGDP01340	M	Naxi	China

Sample ID	Sex	Population	Geographic origin
HGDP01341	M	Naxi	China
HGDP01342	M	Naxi	China
HGDP01343	M	Naxi	China
HGDP01344	M	Naxi	China
HGDP01345	F	Naxi	China
HGDP01346	F	Naxi	China
HGDP01203	M	Oroqen	China
HGDP01204	M	Oroqen	China
HGDP01205	M	Oroqen	China
HGDP01206	M	Oroqen	China
HGDP01207	M	Oroqen	China
HGDP01208	M	Oroqen	China
HGDP01209	F	Oroqen	China
HGDP01210	M	Oroqen	China
HGDP01211	F	Oroqen	China
HGDP01212	F	Oroqen	China
HGDP01327	M	She	China
HGDP01328	M	She	China
HGDP01329	M	She	China
HGDP01330	M	She	China
HGDP01331	M	She	China
HGDP01332	M	She	China
HGDP01333	M	She	China
HGDP01334	F	She	China
HGDP01335	F	She	China
HGDP01336	F	She	China
HGDP01347	M	Tu	China
HGDP01348	M	Tu	China
HGDP01349	M	Tu	China
HGDP01350	M	Tu	China
HGDP01351	M	Tu	China
HGDP01352	M	Tu	China
HGDP01353	M	Tu	China
HGDP01354	F	Tu	China
HGDP01355	F	Tu	China
HGDP01356	F	Tu	China
HGDP01095	M	Tujia	China
HGDP01096	M	Tujia	China
HGDP01097	M	Tujia	China
HGDP01098	F	Tujia	China
HGDP01099	M	Tujia	China
HGDP01100	M	Tujia	China
HGDP01101	M	Tujia	China
HGDP01102	M	Tujia	China
HGDP01103	M	Tujia	China
HGDP01104	M	Tujia	China
HGDP01297	M	Uygur	China
HGDP01298	M	Uygur	China
HGDP01299	M	Uygur	China
HGDP01300	M	Uygur	China
HGDP01301	M	Uygur	China

Sample ID	Sex	Population	Geographic origin
HGDP01302	M	Uygur	China
HGDP01303	M	Uygur	China
HGDP01304	M	Uygur	China
HGDP01305	F	Uygur	China
HGDP01306	F	Uygur	China
HGDP01243	M	Xibo	China
HGDP01244	M	Xibo	China
HGDP01245	M	Xibo	China
HGDP01246	M	Xibo	China
HGDP01247	M	Xibo	China
HGDP01248	M	Xibo	China
HGDP01249	M	Xibo	China
HGDP01250	M	Xibo	China
HGDP01251	F	Xibo	China
HGDP01179	M	Yizu	China
HGDP01180	M	Yizu	China
HGDP01181	M	Yizu	China
HGDP01182	M	Yizu	China
HGDP01183	M	Yizu	China
HGDP01184	M	Yizu	China
HGDP01185	M	Yizu	China
HGDP01186	M	Yizu	China
HGDP01187	M	Yizu	China
HGDP01188	F	Yizu	China
HGDP00702	F	Colombian	Colombia
HGDP00703	M	Colombian	Colombia
HGDP00704	F	Colombian	Colombia
HGDP00705	M	Colombian	Colombia
HGDP00706	F	Colombian	Colombia
HGDP00707	F	Colombian	Colombia
HGDP00708	F	Colombian	Colombia
HGDP00709	M	Colombian	Colombia
HGDP00710	M	Colombian	Colombia
HGDP00792	M	Colombian	Colombia
HGDP00793	F	Colombian	Colombia
HGDP00827	F	Colombian	Colombia
HGDP00970	F	Colombian	Colombia
HGDP00449	M	Mbuti Pygmies	Democratic Republic of Congo
HGDP00450	M	Mbuti Pygmies	Democratic Republic of Congo
HGDP00456	M	Mbuti Pygmies	Democratic Republic of Congo
HGDP00462	M	Mbuti Pygmies	Democratic Republic of Congo
HGDP00463	M	Mbuti Pygmies	Democratic Republic of Congo
HGDP00467	M	Mbuti Pygmies	Democratic Republic of Congo
HGDP00468	M	Mbuti Pygmies	Democratic Republic of Congo
HGDP00471	F	Mbuti Pygmies	Democratic Republic of Congo
HGDP00474	M	Mbuti Pygmies	Democratic Republic of Congo
HGDP00476	F	Mbuti Pygmies	Democratic Republic of Congo
HGDP00478	M	Mbuti Pygmies	Democratic Republic of Congo
HGDP00982	M	Mbuti Pygmies	Democratic Republic of Congo
HGDP00983	M	Mbuti Pygmies	Democratic Republic of Congo
HGDP00984	M	Mbuti Pygmies	Democratic Republic of Congo

Sample ID	Sex	Population	Geographic origin
HGDP01081	M	Mbuti Pygmies	Democratic Republic of Congo
HGDP00511	M	French	France
HGDP00512	M	French	France
HGDP00513	F	French	France
HGDP00514	F	French	France
HGDP00515	M	French	France
HGDP00516	F	French	France
HGDP00517	F	French	France
HGDP00518	M	French	France
HGDP00519	M	French	France
HGDP00520	F	French	France
HGDP00521	M	French	France
HGDP00522	M	French	France
HGDP00523	F	French	France
HGDP00524	F	French	France
HGDP00525	M	French	France
HGDP00526	F	French	France
HGDP00527	F	French	France
HGDP00528	M	French	France
HGDP00529	F	French	France
HGDP00530	M	French	France
HGDP00531	F	French	France
HGDP00532	F	French	France
HGDP00533	M	French	France
HGDP00534	F	French	France
HGDP00535	F	French	France
HGDP00536	F	French	France
HGDP00537	F	French	France
HGDP00538	M	French	France
HGDP00539	F	French	France
HGDP01357	M	French Basque	France
HGDP01358	M	French Basque	France
HGDP01359	M	French Basque	France
HGDP01360	M	French Basque	France
HGDP01361	M	French Basque	France
HGDP01362	M	French Basque	France
HGDP01363	F	French Basque	France
HGDP01364	M	French Basque	France
HGDP01365	F	French Basque	France
HGDP01366	F	French Basque	France
HGDP01367	F	French Basque	France
HGDP01368	F	French Basque	France
HGDP01369	F	French Basque	France
HGDP01370	M	French Basque	France
HGDP01371	M	French Basque	France
HGDP01372	M	French Basque	France
HGDP01373	F	French Basque	France
HGDP01374	M	French Basque	France
HGDP01375	M	French Basque	France
HGDP01376	M	French Basque	France
HGDP01377	M	French Basque	France

Sample ID	Sex	Population	Geographic origin
HGDP01378	M	French Basque	France
HGDP01379	M	French Basque	France
HGDP01380	F	French Basque	France
HGDP00557	F	Druze	Israel (Carmel)
HGDP00558	F	Druze	Israel (Carmel)
HGDP00559	F	Druze	Israel (Carmel)
HGDP00560	F	Druze	Israel (Carmel)
HGDP00561	F	Druze	Israel (Carmel)
HGDP00562	M	Druze	Israel (Carmel)
HGDP00563	F	Druze	Israel (Carmel)
HGDP00564	F	Druze	Israel (Carmel)
HGDP00565	F	Druze	Israel (Carmel)
HGDP00566	F	Druze	Israel (Carmel)
HGDP00567	F	Druze	Israel (Carmel)
HGDP00568	F	Druze	Israel (Carmel)
HGDP00569	F	Druze	Israel (Carmel)
HGDP00570	F	Druze	Israel (Carmel)
HGDP00571	F	Druze	Israel (Carmel)
HGDP00572	F	Druze	Israel (Carmel)
HGDP00573	F	Druze	Israel (Carmel)
HGDP00574	F	Druze	Israel (Carmel)
HGDP00575	F	Druze	Israel (Carmel)
HGDP00576	M	Druze	Israel (Carmel)
HGDP00577	F	Druze	Israel (Carmel)
HGDP00578	F	Druze	Israel (Carmel)
HGDP00579	F	Druze	Israel (Carmel)
HGDP00580	M	Druze	Israel (Carmel)
HGDP00581	F	Druze	Israel (Carmel)
HGDP00582	F	Druze	Israel (Carmel)
HGDP00583	F	Druze	Israel (Carmel)
HGDP00584	F	Druze	Israel (Carmel)
HGDP00585	F	Druze	Israel (Carmel)
HGDP00586	F	Druze	Israel (Carmel)
HGDP00587	F	Druze	Israel (Carmel)
HGDP00588	M	Druze	Israel (Carmel)
HGDP00589	F	Druze	Israel (Carmel)
HGDP00590	F	Druze	Israel (Carmel)
HGDP00591	F	Druze	Israel (Carmel)
HGDP00592	F	Druze	Israel (Carmel)
HGDP00594	M	Druze	Israel (Carmel)
HGDP00595	M	Druze	Israel (Carmel)
HGDP00597	M	Druze	Israel (Carmel)
HGDP00598	M	Druze	Israel (Carmel)
HGDP00599	M	Druze	Israel (Carmel)
HGDP00600	M	Druze	Israel (Carmel)
HGDP00601	F	Druze	Israel (Carmel)
HGDP00602	M	Druze	Israel (Carmel)
HGDP00603	M	Druze	Israel (Carmel)
HGDP00604	M	Druze	Israel (Carmel)
HGDP00605	M	Druze	Israel (Carmel)
HGDP00606	F	Druze	Israel (Carmel)

Sample ID	Sex	Population	Geographic origin
HGDP00675	M	Palestinian	Israel (Central)
HGDP00676	M	Palestinian	Israel (Central)
HGDP00677	M	Palestinian	Israel (Central)
HGDP00678	M	Palestinian	Israel (Central)
HGDP00679	F	Palestinian	Israel (Central)
HGDP00680	F	Palestinian	Israel (Central)
HGDP00681	F	Palestinian	Israel (Central)
HGDP00682	F	Palestinian	Israel (Central)
HGDP00683	F	Palestinian	Israel (Central)
HGDP00684	F	Palestinian	Israel (Central)
HGDP00685	F	Palestinian	Israel (Central)
HGDP00686	F	Palestinian	Israel (Central)
HGDP00687	F	Palestinian	Israel (Central)
HGDP00688	F	Palestinian	Israel (Central)
HGDP00689	F	Palestinian	Israel (Central)
HGDP00690	F	Palestinian	Israel (Central)
HGDP00691	F	Palestinian	Israel (Central)
HGDP00692	F	Palestinian	Israel (Central)
HGDP00693	F	Palestinian	Israel (Central)
HGDP00694	F	Palestinian	Israel (Central)
HGDP00695	F	Palestinian	Israel (Central)
HGDP00696	F	Palestinian	Israel (Central)
HGDP00697	F	Palestinian	Israel (Central)
HGDP00698	F	Palestinian	Israel (Central)
HGDP00699	F	Palestinian	Israel (Central)
HGDP00700	F	Palestinian	Israel (Central)
HGDP00722	M	Palestinian	Israel (Central)
HGDP00723	M	Palestinian	Israel (Central)
HGDP00724	M	Palestinian	Israel (Central)
HGDP00725	M	Palestinian	Israel (Central)
HGDP00726	M	Palestinian	Israel (Central)
HGDP00727	M	Palestinian	Israel (Central)
HGDP00728	M	Palestinian	Israel (Central)
HGDP00729	M	Palestinian	Israel (Central)
HGDP00730	M	Palestinian	Israel (Central)
HGDP00731	M	Palestinian	Israel (Central)
HGDP00732	M	Palestinian	Israel (Central)
HGDP00733	M	Palestinian	Israel (Central)
HGDP00734	M	Palestinian	Israel (Central)
HGDP00735	F	Palestinian	Israel (Central)
HGDP00736	F	Palestinian	Israel (Central)
HGDP00737	F	Palestinian	Israel (Central)
HGDP00738	F	Palestinian	Israel (Central)
HGDP00739	F	Palestinian	Israel (Central)
HGDP00740	F	Palestinian	Israel (Central)
HGDP00741	F	Palestinian	Israel (Central)
HGDP00742	F	Palestinian	Israel (Central)
HGDP00743	F	Palestinian	Israel (Central)
HGDP00744	F	Palestinian	Israel (Central)
HGDP00745	F	Palestinian	Israel (Central)
HGDP00746	F	Palestinian	Israel (Central)

Sample ID	Sex	Population	Geographic origin
HGDP00607	F	Bedouin	Israel (Negev)
HGDP00608	M	Bedouin	Israel (Negev)
HGDP00609	M	Bedouin	Israel (Negev)
HGDP00610	M	Bedouin	Israel (Negev)
HGDP00611	M	Bedouin	Israel (Negev)
HGDP00612	F	Bedouin	Israel (Negev)
HGDP00613	F	Bedouin	Israel (Negev)
HGDP00614	F	Bedouin	Israel (Negev)
HGDP00615	F	Bedouin	Israel (Negev)
HGDP00616	M	Bedouin	Israel (Negev)
HGDP00617	M	Bedouin	Israel (Negev)
HGDP00618	M	Bedouin	Israel (Negev)
HGDP00619	M	Bedouin	Israel (Negev)
HGDP00620	M	Bedouin	Israel (Negev)
HGDP00621	M	Bedouin	Israel (Negev)
HGDP00622	M	Bedouin	Israel (Negev)
HGDP00623	M	Bedouin	Israel (Negev)
HGDP00624	M	Bedouin	Israel (Negev)
HGDP00625	M	Bedouin	Israel (Negev)
HGDP00626	M	Bedouin	Israel (Negev)
HGDP00627	M	Bedouin	Israel (Negev)
HGDP00628	M	Bedouin	Israel (Negev)
HGDP00629	M	Bedouin	Israel (Negev)
HGDP00630	M	Bedouin	Israel (Negev)
HGDP00631	M	Bedouin	Israel (Negev)
HGDP00632	F	Bedouin	Israel (Negev)
HGDP00633	F	Bedouin	Israel (Negev)
HGDP00634	F	Bedouin	Israel (Negev)
HGDP00635	F	Bedouin	Israel (Negev)
HGDP00636	F	Bedouin	Israel (Negev)
HGDP00637	F	Bedouin	Israel (Negev)
HGDP00638	F	Bedouin	Israel (Negev)
HGDP00639	M	Bedouin	Israel (Negev)
HGDP00640	M	Bedouin	Israel (Negev)
HGDP00641	M	Bedouin	Israel (Negev)
HGDP00642	M	Bedouin	Israel (Negev)
HGDP00643	F	Bedouin	Israel (Negev)
HGDP00644	M	Bedouin	Israel (Negev)
HGDP00645	M	Bedouin	Israel (Negev)
HGDP00646	F	Bedouin	Israel (Negev)
HGDP00647	F	Bedouin	Israel (Negev)
HGDP00648	M	Bedouin	Israel (Negev)
HGDP00649	F	Bedouin	Israel (Negev)
HGDP00650	F	Bedouin	Israel (Negev)
HGDP00651	F	Bedouin	Israel (Negev)
HGDP00652	F	Bedouin	Israel (Negev)
HGDP00653	F	Bedouin	Israel (Negev)
HGDP00654	M	Bedouin	Israel (Negev)
HGDP00701	F	Bedouin	Israel (Negev)
HGDP00665	M	Sardinian	Italy
HGDP00666	M	Sardinian	Italy

Sample ID	Sex	Population	Geographic origin
HGDP00667	F	Sardinian	Italy
HGDP00668	M	Sardinian	Italy
HGDP00669	F	Sardinian	Italy
HGDP00670	M	Sardinian	Italy
HGDP00671	M	Sardinian	Italy
HGDP00672	F	Sardinian	Italy
HGDP00673	F	Sardinian	Italy
HGDP00674	M	Sardinian	Italy
HGDP01062	F	Sardinian	Italy
HGDP01063	M	Sardinian	Italy
HGDP01064	F	Sardinian	Italy
HGDP01065	F	Sardinian	Italy
HGDP01066	M	Sardinian	Italy
HGDP01067	M	Sardinian	Italy
HGDP01068	F	Sardinian	Italy
HGDP01069	M	Sardinian	Italy
HGDP01070	F	Sardinian	Italy
HGDP01071	M	Sardinian	Italy
HGDP01072	F	Sardinian	Italy
HGDP01073	M	Sardinian	Italy
HGDP01074	F	Sardinian	Italy
HGDP01075	M	Sardinian	Italy
HGDP01076	M	Sardinian	Italy
HGDP01077	M	Sardinian	Italy
HGDP01078	F	Sardinian	Italy
HGDP01079	M	Sardinian	Italy
HGDP01161	M	Tuscan	Italy
HGDP01162	M	Tuscan	Italy
HGDP01163	M	Tuscan	Italy
HGDP01164	M	Tuscan	Italy
HGDP01166	M	Tuscan	Italy
HGDP01167	M	Tuscan	Italy
HGDP01168	F	Tuscan	Italy
HGDP01169	F	Tuscan	Italy
HGDP01147	M	North Italian	Italy (Bergamo)
HGDP01149	M	North Italian	Italy (Bergamo)
HGDP01151	M	North Italian	Italy (Bergamo)
HGDP01152	M	North Italian	Italy (Bergamo)
HGDP01153	M	North Italian	Italy (Bergamo)
HGDP01154	M	North Italian	Italy (Bergamo)
HGDP01155	M	North Italian	Italy (Bergamo)
HGDP01156	F	North Italian	Italy (Bergamo)
HGDP01157	F	North Italian	Italy (Bergamo)
HGDP01171	F	North Italian	Italy (Bergamo)
HGDP01172	F	North Italian	Italy (Bergamo)
HGDP01173	M	North Italian	Italy (Bergamo)
HGDP01174	M	North Italian	Italy (Bergamo)
HGDP01177	F	North Italian	Italy (Bergamo)
HGDP00747	M	Japanese	Japan
HGDP00748	M	Japanese	Japan
HGDP00749	M	Japanese	Japan

Sample ID	Sex	Population	Geographic origin
HGDP00750	M	Japanese	Japan
HGDP00751	M	Japanese	Japan
HGDP00752	M	Japanese	Japan
HGDP00753	M	Japanese	Japan
HGDP00754	F	Japanese	Japan
HGDP00755	M	Japanese	Japan
HGDP00756	F	Japanese	Japan
HGDP00757	M	Japanese	Japan
HGDP00758	M	Japanese	Japan
HGDP00759	M	Japanese	Japan
HGDP00760	F	Japanese	Japan
HGDP00761	F	Japanese	Japan
HGDP00762	M	Japanese	Japan
HGDP00763	M	Japanese	Japan
HGDP00764	M	Japanese	Japan
HGDP00765	F	Japanese	Japan
HGDP00766	M	Japanese	Japan
HGDP00767	M	Japanese	Japan
HGDP00768	M	Japanese	Japan
HGDP00769	M	Japanese	Japan
HGDP00770	M	Japanese	Japan
HGDP00771	F	Japanese	Japan
HGDP00772	F	Japanese	Japan
HGDP00773	F	Japanese	Japan
HGDP00790	M	Japanese	Japan
HGDP00791	M	Japanese	Japan
HGDP00828	M	Japanese	Japan
HGDP01025	M	Japanese	Japan
HGDP01405	M	Bantu N.E.	Kenya
HGDP01406	M	Bantu N.E.	Kenya
HGDP01408	M	Bantu N.E.	Kenya
HGDP01411	M	Bantu N.E.	Kenya
HGDP01412	M	Bantu N.E.	Kenya
HGDP01413	M	Bantu N.E.	Kenya
HGDP01414	F	Bantu N.E.	Kenya
HGDP01415	M	Bantu N.E.	Kenya
HGDP01416	M	Bantu N.E.	Kenya
HGDP01417	M	Bantu N.E.	Kenya
HGDP01418	M	Bantu N.E.	Kenya
HGDP01419	M	Bantu N.E.	Kenya
HGDP00854	F	Maya	Mexico
HGDP00855	F	Maya	Mexico
HGDP00856	M	Maya	Mexico
HGDP00857	F	Maya	Mexico
HGDP00858	F	Maya	Mexico
HGDP00859	F	Maya	Mexico
HGDP00860	F	Maya	Mexico
HGDP00861	F	Maya	Mexico
HGDP00862	F	Maya	Mexico
HGDP00863	F	Maya	Mexico
HGDP00864	F	Maya	Mexico

Sample ID	Sex	Population	Geographic origin
HGDP00865	F	Maya	Mexico
HGDP00866	F	Maya	Mexico
HGDP00867	F	Maya	Mexico
HGDP00868	F	Maya	Mexico
HGDP00869	F	Maya	Mexico
HGDP00870	F	Maya	Mexico
HGDP00871	F	Maya	Mexico
HGDP00872	F	Maya	Mexico
HGDP00873	F	Maya	Mexico
HGDP00874	F	Maya	Mexico
HGDP00875	F	Maya	Mexico
HGDP00876	F	Maya	Mexico
HGDP00877	M	Maya	Mexico
HGDP00878	M	Maya	Mexico
HGDP01037	M	Pima	Mexico
HGDP01038	F	Pima	Mexico
HGDP01039	M	Pima	Mexico
HGDP01040	M	Pima	Mexico
HGDP01041	F	Pima	Mexico
HGDP01042	M	Pima	Mexico
HGDP01043	M	Pima	Mexico
HGDP01044	F	Pima	Mexico
HGDP01045	M	Pima	Mexico
HGDP01046	F	Pima	Mexico
HGDP01047	M	Pima	Mexico
HGDP01048	F	Pima	Mexico
HGDP01049	F	Pima	Mexico
HGDP01050	M	Pima	Mexico
HGDP01051	F	Pima	Mexico
HGDP01052	M	Pima	Mexico
HGDP01053	F	Pima	Mexico
HGDP01054	F	Pima	Mexico
HGDP01055	M	Pima	Mexico
HGDP01056	F	Pima	Mexico
HGDP01057	M	Pima	Mexico
HGDP01058	F	Pima	Mexico
HGDP01059	M	Pima	Mexico
HGDP01060	M	Pima	Mexico
HGDP01061	M	Pima	Mexico
HGDP00987	M	San	Namidia
HGDP00988	M	San	Namidia
HGDP00991	M	San	Namidia
HGDP00992	M	San	Namidia
HGDP01029	M	San	Namidia
HGDP01032	M	San	Namidia
HGDP01036	M	San	Namidia
HGDP00540	M	Papuan	New Guinea
HGDP00541	M	Papuan	New Guinea
HGDP00542	M	Papuan	New Guinea
HGDP00543	M	Papuan	New Guinea
HGDP00544	F	Papuan	New Guinea

Sample ID	Sex	Population	Geographic origin
HGDP00545	M	Papuan	New Guinea
HGDP00546	M	Papuan	New Guinea
HGDP00547	M	Papuan	New Guinea
HGDP00548	M	Papuan	New Guinea
HGDP00549	M	Papuan	New Guinea
HGDP00550	F	Papuan	New Guinea
HGDP00551	M	Papuan	New Guinea
HGDP00552	F	Papuan	New Guinea
HGDP00553	M	Papuan	New Guinea
HGDP00554	F	Papuan	New Guinea
HGDP00555	M	Papuan	New Guinea
HGDP00556	M	Papuan	New Guinea
HGDP00920	F	Yoruba	Nigeria
HGDP00921	F	Yoruba	Nigeria
HGDP00922	F	Yoruba	Nigeria
HGDP00923	M	Yoruba	Nigeria
HGDP00924	F	Yoruba	Nigeria
HGDP00925	F	Yoruba	Nigeria
HGDP00926	F	Yoruba	Nigeria
HGDP00927	M	Yoruba	Nigeria
HGDP00928	F	Yoruba	Nigeria
HGDP00929	M	Yoruba	Nigeria
HGDP00930	M	Yoruba	Nigeria
HGDP00931	M	Yoruba	Nigeria
HGDP00932	M	Yoruba	Nigeria
HGDP00933	F	Yoruba	Nigeria
HGDP00934	F	Yoruba	Nigeria
HGDP00935	F	Yoruba	Nigeria
HGDP00936	M	Yoruba	Nigeria
HGDP00937	M	Yoruba	Nigeria
HGDP00938	F	Yoruba	Nigeria
HGDP00939	F	Yoruba	Nigeria
HGDP00940	M	Yoruba	Nigeria
HGDP00941	M	Yoruba	Nigeria
HGDP00942	M	Yoruba	Nigeria
HGDP00943	M	Yoruba	Nigeria
HGDP00944	M	Yoruba	Nigeria
HGDP00794	F	Orcadian	Orkney Islands
HGDP00795	M	Orcadian	Orkney Islands
HGDP00796	F	Orcadian	Orkney Islands
HGDP00797	F	Orcadian	Orkney Islands
HGDP00798	M	Orcadian	Orkney Islands
HGDP00799	F	Orcadian	Orkney Islands
HGDP00800	F	Orcadian	Orkney Islands
HGDP00801	F	Orcadian	Orkney Islands
HGDP00802	F	Orcadian	Orkney Islands
HGDP00803	M	Orcadian	Orkney Islands
HGDP00804	M	Orcadian	Orkney Islands
HGDP00805	F	Orcadian	Orkney Islands
HGDP00806	F	Orcadian	Orkney Islands
HGDP00807	M	Orcadian	Orkney Islands

Sample ID	Sex	Population	Geographic origin
HGDP00808	M	Orcadian	Orkney Islands
HGDP00810	M	Orcadian	Orkney Islands
HGDP00052	M	Balochi	Pakistan
HGDP00054	M	Balochi	Pakistan
HGDP00056	M	Balochi	Pakistan
HGDP00057	M	Balochi	Pakistan
HGDP00058	M	Balochi	Pakistan
HGDP00060	M	Balochi	Pakistan
HGDP00062	M	Balochi	Pakistan
HGDP00064	M	Balochi	Pakistan
HGDP00066	M	Balochi	Pakistan
HGDP00068	M	Balochi	Pakistan
HGDP00070	M	Balochi	Pakistan
HGDP00072	M	Balochi	Pakistan
HGDP00074	M	Balochi	Pakistan
HGDP00076	M	Balochi	Pakistan
HGDP00078	M	Balochi	Pakistan
HGDP00080	M	Balochi	Pakistan
HGDP00082	M	Balochi	Pakistan
HGDP00084	M	Balochi	Pakistan
HGDP00086	M	Balochi	Pakistan
HGDP00088	M	Balochi	Pakistan
HGDP00090	M	Balochi	Pakistan
HGDP00092	M	Balochi	Pakistan
HGDP00094	M	Balochi	Pakistan
HGDP00096	M	Balochi	Pakistan
HGDP00098	M	Balochi	Pakistan
HGDP00001	M	Brahui	Pakistan
HGDP00003	M	Brahui	Pakistan
HGDP00005	M	Brahui	Pakistan
HGDP00007	M	Brahui	Pakistan
HGDP00009	M	Brahui	Pakistan
HGDP00011	M	Brahui	Pakistan
HGDP00013	M	Brahui	Pakistan
HGDP00015	M	Brahui	Pakistan
HGDP00017	M	Brahui	Pakistan
HGDP00019	M	Brahui	Pakistan
HGDP00021	M	Brahui	Pakistan
HGDP00023	M	Brahui	Pakistan
HGDP00025	M	Brahui	Pakistan
HGDP00027	M	Brahui	Pakistan
HGDP00029	M	Brahui	Pakistan
HGDP00031	M	Brahui	Pakistan
HGDP00033	M	Brahui	Pakistan
HGDP00035	M	Brahui	Pakistan
HGDP00037	M	Brahui	Pakistan
HGDP00039	M	Brahui	Pakistan
HGDP00041	M	Brahui	Pakistan
HGDP00043	M	Brahui	Pakistan
HGDP00045	M	Brahui	Pakistan
HGDP00047	M	Brahui	Pakistan

Sample ID	Sex	Population	Geographic origin
HGDP00049	M	Brahui	Pakistan
HGDP00336	F	Burusho	Pakistan
HGDP00338	F	Burusho	Pakistan
HGDP00341	M	Burusho	Pakistan
HGDP00346	M	Burusho	Pakistan
HGDP00351	M	Burusho	Pakistan
HGDP00356	F	Burusho	Pakistan
HGDP00359	M	Burusho	Pakistan
HGDP00364	M	Burusho	Pakistan
HGDP00371	F	Burusho	Pakistan
HGDP00372	M	Burusho	Pakistan
HGDP00376	M	Burusho	Pakistan
HGDP00382	M	Burusho	Pakistan
HGDP00388	M	Burusho	Pakistan
HGDP00392	M	Burusho	Pakistan
HGDP00397	M	Burusho	Pakistan
HGDP00402	M	Burusho	Pakistan
HGDP00407	M	Burusho	Pakistan
HGDP00412	M	Burusho	Pakistan
HGDP00417	M	Burusho	Pakistan
HGDP00423	M	Burusho	Pakistan
HGDP00428	M	Burusho	Pakistan
HGDP00433	M	Burusho	Pakistan
HGDP00438	M	Burusho	Pakistan
HGDP00444	F	Burusho	Pakistan
HGDP00445	M	Burusho	Pakistan
HGDP00099	M	Hazara	Pakistan
HGDP00100	M	Hazara	Pakistan
HGDP00102	M	Hazara	Pakistan
HGDP00103	M	Hazara	Pakistan
HGDP00104	M	Hazara	Pakistan
HGDP00105	M	Hazara	Pakistan
HGDP00106	M	Hazara	Pakistan
HGDP00108	M	Hazara	Pakistan
HGDP00109	M	Hazara	Pakistan
HGDP00110	M	Hazara	Pakistan
HGDP00111	M	Hazara	Pakistan
HGDP00112	M	Hazara	Pakistan
HGDP00113	M	Hazara	Pakistan
HGDP00115	M	Hazara	Pakistan
HGDP00116	M	Hazara	Pakistan
HGDP00118	M	Hazara	Pakistan
HGDP00119	M	Hazara	Pakistan
HGDP00120	M	Hazara	Pakistan
HGDP00121	M	Hazara	Pakistan
HGDP00122	M	Hazara	Pakistan
HGDP00124	M	Hazara	Pakistan
HGDP00125	M	Hazara	Pakistan
HGDP00127	M	Hazara	Pakistan
HGDP00128	M	Hazara	Pakistan
HGDP00129	M	Hazara	Pakistan

Sample ID	Sex	Population	Geographic origin
HGDP00267	M	Kalash	Pakistan
HGDP00274	F	Kalash	Pakistan
HGDP00277	M	Kalash	Pakistan
HGDP00279	M	Kalash	Pakistan
HGDP00281	M	Kalash	Pakistan
HGDP00285	M	Kalash	Pakistan
HGDP00286	F	Kalash	Pakistan
HGDP00288	M	Kalash	Pakistan
HGDP00290	M	Kalash	Pakistan
HGDP00292	M	Kalash	Pakistan
HGDP00298	F	Kalash	Pakistan
HGDP00302	M	Kalash	Pakistan
HGDP00304	F	Kalash	Pakistan
HGDP00307	M	Kalash	Pakistan
HGDP00309	M	Kalash	Pakistan
HGDP00311	M	Kalash	Pakistan
HGDP00313	M	Kalash	Pakistan
HGDP00315	M	Kalash	Pakistan
HGDP00319	M	Kalash	Pakistan
HGDP00321	M	Kalash	Pakistan
HGDP00323	F	Kalash	Pakistan
HGDP00326	M	Kalash	Pakistan
HGDP00328	M	Kalash	Pakistan
HGDP00330	M	Kalash	Pakistan
HGDP00333	M	Kalash	Pakistan
HGDP00130	M	Makrani	Pakistan
HGDP00131	M	Makrani	Pakistan
HGDP00133	M	Makrani	Pakistan
HGDP00134	M	Makrani	Pakistan
HGDP00135	M	Makrani	Pakistan
HGDP00136	M	Makrani	Pakistan
HGDP00137	M	Makrani	Pakistan
HGDP00139	M	Makrani	Pakistan
HGDP00140	M	Makrani	Pakistan
HGDP00141	M	Makrani	Pakistan
HGDP00143	M	Makrani	Pakistan
HGDP00144	M	Makrani	Pakistan
HGDP00145	M	Makrani	Pakistan
HGDP00146	M	Makrani	Pakistan
HGDP00148	M	Makrani	Pakistan
HGDP00149	M	Makrani	Pakistan
HGDP00150	M	Makrani	Pakistan
HGDP00151	F	Makrani	Pakistan
HGDP00153	F	Makrani	Pakistan
HGDP00154	F	Makrani	Pakistan
HGDP00155	F	Makrani	Pakistan
HGDP00157	F	Makrani	Pakistan
HGDP00158	M	Makrani	Pakistan
HGDP00160	M	Makrani	Pakistan
HGDP00161	M	Makrani	Pakistan
HGDP00213	M	Pathan	Pakistan

Sample ID	Sex	Population	Geographic origin
HGDP00214	M	Pathan	Pakistan
HGDP00216	M	Pathan	Pakistan
HGDP00218	M	Pathan	Pakistan
HGDP00220	M	Pathan	Pakistan
HGDP00222	M	Pathan	Pakistan
HGDP00224	M	Pathan	Pakistan
HGDP00226	M	Pathan	Pakistan
HGDP00228	M	Pathan	Pakistan
HGDP00230	M	Pathan	Pakistan
HGDP00232	F	Pathan	Pakistan
HGDP00234	M	Pathan	Pakistan
HGDP00237	F	Pathan	Pakistan
HGDP00239	F	Pathan	Pakistan
HGDP00241	M	Pathan	Pakistan
HGDP00243	M	Pathan	Pakistan
HGDP00244	F	Pathan	Pakistan
HGDP00247	F	Pathan	Pakistan
HGDP00248	M	Pathan	Pakistan
HGDP00251	M	Pathan	Pakistan
HGDP00254	M	Pathan	Pakistan
HGDP00258	M	Pathan	Pakistan
HGDP00259	M	Pathan	Pakistan
HGDP00262	M	Pathan	Pakistan
HGDP00264	M	Pathan	Pakistan
HGDP00163	M	Sindhi	Pakistan
HGDP00165	M	Sindhi	Pakistan
HGDP00167	M	Sindhi	Pakistan
HGDP00169	M	Sindhi	Pakistan
HGDP00171	M	Sindhi	Pakistan
HGDP00173	M	Sindhi	Pakistan
HGDP00175	M	Sindhi	Pakistan
HGDP00177	M	Sindhi	Pakistan
HGDP00179	M	Sindhi	Pakistan
HGDP00181	M	Sindhi	Pakistan
HGDP00183	M	Sindhi	Pakistan
HGDP00185	M	Sindhi	Pakistan
HGDP00187	M	Sindhi	Pakistan
HGDP00189	M	Sindhi	Pakistan
HGDP00191	M	Sindhi	Pakistan
HGDP00192	F	Sindhi	Pakistan
HGDP00195	F	Sindhi	Pakistan
HGDP00197	M	Sindhi	Pakistan
HGDP00199	M	Sindhi	Pakistan
HGDP00201	M	Sindhi	Pakistan
HGDP00203	M	Sindhi	Pakistan
HGDP00205	M	Sindhi	Pakistan
HGDP00206	F	Sindhi	Pakistan
HGDP00208	M	Sindhi	Pakistan
HGDP00210	F	Sindhi	Pakistan
HGDP00879	M	Russian	Russia
HGDP00880	M	Russian	Russia

Sample ID	Sex	Population	Geographic origin
HGDP00881	F	Russian	Russia
HGDP00882	M	Russian	Russia
HGDP00883	M	Russian	Russia
HGDP00884	F	Russian	Russia
HGDP00885	F	Russian	Russia
HGDP00886	M	Russian	Russia
HGDP00887	M	Russian	Russia
HGDP00888	M	Russian	Russia
HGDP00889	F	Russian	Russia
HGDP00890	M	Russian	Russia
HGDP00891	M	Russian	Russia
HGDP00892	M	Russian	Russia
HGDP00893	M	Russian	Russia
HGDP00894	M	Russian	Russia
HGDP00895	M	Russian	Russia
HGDP00896	M	Russian	Russia
HGDP00897	M	Russian	Russia
HGDP00898	F	Russian	Russia
HGDP00899	F	Russian	Russia
HGDP00900	M	Russian	Russia
HGDP00901	F	Russian	Russia
HGDP00902	F	Russian	Russia
HGDP00903	F	Russian	Russia
HGDP01381	F	Adygei	Russia Caucasus
HGDP01382	F	Adygei	Russia Caucasus
HGDP01383	M	Adygei	Russia Caucasus
HGDP01384	F	Adygei	Russia Caucasus
HGDP01385	M	Adygei	Russia Caucasus
HGDP01386	F	Adygei	Russia Caucasus
HGDP01387	F	Adygei	Russia Caucasus
HGDP01388	F	Adygei	Russia Caucasus
HGDP01396	M	Adygei	Russia Caucasus
HGDP01397	M	Adygei	Russia Caucasus
HGDP01398	F	Adygei	Russia Caucasus
HGDP01399	F	Adygei	Russia Caucasus
HGDP01400	F	Adygei	Russia Caucasus
HGDP01401	F	Adygei	Russia Caucasus
HGDP01402	M	Adygei	Russia Caucasus
HGDP01403	M	Adygei	Russia Caucasus
HGDP01404	M	Adygei	Russia Caucasus
HGDP00904	M	Mandenka	Senegal
HGDP00905	M	Mandenka	Senegal
HGDP00906	M	Mandenka	Senegal
HGDP00907	M	Mandenka	Senegal
HGDP00908	M	Mandenka	Senegal
HGDP00909	F	Mandenka	Senegal
HGDP00910	F	Mandenka	Senegal
HGDP00911	M	Mandenka	Senegal
HGDP00912	M	Mandenka	Senegal
HGDP00913	M	Mandenka	Senegal
HGDP00914	F	Mandenka	Senegal

Sample ID	Sex	Population	Geographic origin
HGDP00915	F	Mandenka	Senegal
HGDP00916	F	Mandenka	Senegal
HGDP00917	F	Mandenka	Senegal
HGDP00918	F	Mandenka	Senegal
HGDP00919	M	Mandenka	Senegal
HGDP01199	M	Mandenka	Senegal
HGDP01200	M	Mandenka	Senegal
HGDP01201	F	Mandenka	Senegal
HGDP01202	M	Mandenka	Senegal
HGDP01283	M	Mandenka	Senegal
HGDP01284	M	Mandenka	Senegal
HGDP01285	M	Mandenka	Senegal
HGDP01286	M	Mandenka	Senegal
HGDP00945	M	Yakut	Siberia
HGDP00946	M	Yakut	Siberia
HGDP00947	M	Yakut	Siberia
HGDP00948	M	Yakut	Siberia
HGDP00949	M	Yakut	Siberia
HGDP00950	M	Yakut	Siberia
HGDP00951	M	Yakut	Siberia
HGDP00952	M	Yakut	Siberia
HGDP00953	M	Yakut	Siberia
HGDP00954	M	Yakut	Siberia
HGDP00955	F	Yakut	Siberia
HGDP00956	F	Yakut	Siberia
HGDP00957	F	Yakut	Siberia
HGDP00958	M	Yakut	Siberia
HGDP00959	F	Yakut	Siberia
HGDP00960	M	Yakut	Siberia
HGDP00961	M	Yakut	Siberia
HGDP00962	M	Yakut	Siberia
HGDP00963	F	Yakut	Siberia
HGDP00964	M	Yakut	Siberia
HGDP00965	M	Yakut	Siberia
HGDP00966	F	Yakut	Siberia
HGDP00967	F	Yakut	Siberia
HGDP00968	M	Yakut	Siberia
HGDP00969	M	Yakut	Siberia
HGDP00993	M	Bantu S.E. Pedi	South Africa
HGDP00994	M	Bantu S.E. S.Sotho	South Africa
HGDP01030	M	Bantu S.E. Tswana	South Africa
HGDP01034	M	Bantu S.E. Tswana	South Africa
HGDP01033	M	Bantu S.E. Zulu	South Africa
HGDP01028	M	Bantu S.W. Herero	South Africa
HGDP01035	M	Bantu S.W. Herero	South Africa
HGDP01031	M	Bantu S.W. Ovambo	South Africa

Table A4 European-Indian (CIN) control panel

Sample ID	Ethnic Origin	Sample ID	Ethnic Origin
POD-AB41	European	POD-BD462	Indian
POD-AB44	European	POD-BD56	Indian
POD-AB54	European	POD-BD57	Indian
POD-AB56	European	POD-BD59	Indian
POD-AB79	European	POD-BD61	Indian
POD-AB80	European	POD-BD78	Indian
POD-AC315	Indian	POD-BD781	Indian
POD-AC332	European	POD-BD891	Indian
POD-AF124	Indian	POD-BO101	European
POD-AF15	European	POD-BO19	European
POD-AF236	Indian	POD-BO590	European
POD-AF24	European	POD-BO65	European
POD-AF3	European	POD-BO71	European
POD-AF50	European	POD-BO96	European
POD-AF6	European	POD-BQ63	Indian
POD-AG0033	European	POD-BQ68	European
POD-AG11	European	POD-BV278	Indian
POD-AG17	European	POD-BW152	Indian
POD-AG547	European	POD-BW156	Indian
POD-AG646	Indian	POD-BW158	Indian
POD-AG66	European	POD-BW160	Indian
POD-AJ10	European	POD-CB276	Indian
POD-AJ2	Indian	POD-CB464	Indian
POD-AK353	European	POD-CB874	Indian
POD-AK400	Indian	POD-CD59	Indian
POD-AK503	European	POD-CD68	Indian
POD-AM107	Indian	POD-CD79	Indian
POD-AM125	Indian	POD-DB104	Indian
POD-AM148	Indian	POD-GG1	European
POD-AM16	Indian	POD-HH15	European
POD-AM17	Indian	POD-HH170	European
POD-AM21	Indian	POD-HH176	European
POD-AM31	Indian	POD-HH195	European
POD-AM37	Indian	POD-HH198	European
POD-AM38	Indian	POD-HH30	European
POD-AM40	Indian	POD-HH74	European
POD-AM42	Indian	POD-HH8	European
POD-AM54	Indian	POD-MM16	European
POD-AM69	European	POD-QQ63	Indian
POD-AR55	European	POD-QQ65	Indian
POD-AR70	Indian	POD-QQ67	Indian
POD-AW165	Indian	POD-TT100	European
POD-AW53	Indian	POD-TT37	European
POD-AX17	European	POD-TT71	European
POD-BC45	Indian	POD-WW001	European
POD-BD238	Indian	POD-WW5	European
POD-BD245	European	POD-YY4	European

Table A5 *LPIN2* sequence variants detected in a cohort of severe insulin resistant patients and 11 Indian and 23 CEPH controls

Genic position	Genomic position	Minor|Major allele	Protein consequence	MAF in SIR	Detected in controls?	rs ID
Upstream	3002144	C|G		0.0055		rs3810061
Upstream	3002078	G|C		0.0989		rs3810062
Upstream	3001969	T|C		0.0026		
5prime_UTR	3001787	A|T		0.0026		
5prime_UTR	3001772	A|G		0.0026		
5prime_UTR	3001756	C|T		0.0026		
5prime_UTR	3001743	C|G		0.0078		
5prime_UTR	3001742	C|G		0.0078		
Intron 1	3001646	T|C		0.0026		
Intron 1	3001587	A|G		0.1		
Intron 1	3001563	C|T		0.1099		
Intron 1	3001480	T|C		0.0026		
Intron 1	3001440	G|T		0.006		
Intron 1	3001415	A|G		0.0186		rs12455800
Intron 1	2972876	C|T		0.0027		
Intron 1	2972733	T|A		0.0026		
Intron 3	2944448	A|G		0.0026		
Intron 3	2944439	T|C		0.3803		rs7226624
Intron 4	2940926	G|A		0.0026		
Exon 5	2930695	G|A	A202A	0.0027		
Exon 5	2930693	A|G	S203F	0.0081	Yes	
Intron 5	2929627	G|T		0.0026		
Intron 6	2929424	C|T		0.0026		
Intron 6	2929307	A|T		0.0026		
Intron 7	2924536	C|G		0.1219		rs3765622
Intron 9	2921225	C|T		0.0133		rs16944068
Intron 9	2921074	C|G		0.0081		
Intron 9	2919186	G|T		0.0026		
Exon 10	2919124	T|C	**E497K**	0.0026	No	
Intron 12	2917669	C|A		0.0026		
Intron 12	2917023	G|T		0.4274		rs602233
Exon 13	2916779	G|A	S579P	0.0026	Yes	
Exon 14	2915359	T|C	E601K	0.0111	Yes	
Exon 14	2915284	A|G	**P626S**	0.0027	No	
Intron 14	2915104	T|C		0.0028		
Intron 15	2914278	G|A		0.0026		
Intron 15	2913884	C|A		0.0052		
Exon 16	2913790	A|G	L719L	0.0052		
Intron 16	2913699	A|T		0.0026		
Intron 16	2912450	C|T		0.378		rs12458532
Intron 16	2912360	A|C		0.0026		
Intron 17	2911979	T|C		0.032		
Intron 17	2911777	C|T		0.2771		rs2282636
Intron 17	2911752	C|T		0.0027		
Intron 18	2911444	C|T		0.2771		rs2282635
Intron 18	2911433	C|A		0.0027		
Intron 19	2910725	T|A		0.1648		rs3737514

Genic position	Genomic position	Minor|Major allele	Protein consequence	MAF in SIR	Detected in controls?	rs ID
Intron 19	2910631	G|A		0.014		
Intron 19	2910482	A|G		0.0083		rs16944043
Exon 20	2910387	G|A	S865S	0.0029		
Exon 20	2910372	A|G	S870S	0.0026		
3prime_UTR	2910288	A|G		0.293		rs3745012
3prime_UTR	2910201	A|G		0.0108		
3prime_UTR	2910186	A|G		0.0026		
3prime_UTR	2910124	A|G		0.0026		
3prime_UTR	2909933	T|C		0.0052		
3prime_UTR	2909853	A|G		0.0052		
3prime_UTR	2909843	A|G		0.0026		
3prime_UTR	2909790	G|A		0.0629		rs35176958
3prime_UTR	2909734	T|C		0.05		
3prime_UTR	2909639	A|G		0.0026		
3prime_UTR	2909634	G|T		0.086		rs16944040
3prime_UTR	2909606	A|T		0.2783		rs607549
3prime_UTR	2909538	G|A		0.0026		
3prime_UTR	2909494	C|A		0.0026		rs3810064
3prime_UTR	2909478	G|T		0.0026		
3prime_UTR	2909377	G|A		0.0028		
3prime_UTR	2909341	G|T		0.0058		rs3810065
3prime_UTR	2909239	G|A		0.0364		
3prime_UTR	2909214	C|G		0.0027		
3prime_UTR	2909209	T|C		0.0026		
3prime_UTR	2909170	T|C		0.0058		
3prime_UTR	2908933	T|C		0.0026		
3prime_UTR	2908888	T|C		0.0026		
3prime_UTR	2908789	T|C		0.009		
3prime_UTR	2908699	C|T		0.0026		
3prime_UTR	2908669	G|A		0.0026		
3prime_UTR	2908426	A|C		0.0027		
3prime_UTR	2908215	T|G		0.0028		
3prime_UTR	2908188	T|C		0.0081		
3prime_UTR	2907884	T|C		0.0029		
3prime_UTR	2907874	A|G		0.0031		
3prime_UTR	2907798	C|G		0.037		rs8091401
3prime_UTR	2907756	G|A		0.0026		
3prime_UTR	2907738	T|C		0.0052		
3prime_UTR	2907357	G|A		0.3882		rs7980
3prime_UTR	2907345	T|C		0.0106		rs4781
3prime_UTR	2907276	G|A		0.0026		rs14916
3prime_UTR	2907223	A|T		0.2777		rs1985
3prime_UTR	2907185	A|C		0.0026		
3prime_UTR	2907152	C|T		0.2063		rs1164
Downstream	2906904	G|C		0.0846		

Genomic coordinates are NCBI build 36 (B36).

Table A6 *LPIN3* sequence variants detected in a cohort of severe insulin resistant patients and 11 Indian and 23 CEPH controls

Genic position	Genomic position	Minor|Major allele	Protein consequence	MAF in SIR	Detected in controls?	rs ID
Upstream	39402927	A|C		0.2807		rs6029636
Intron 1	39403067	C|A		0.0027		
Intron 1	39403147	T|C		0.0026		
Intron 1	39407730	G|A		0.2884		rs6129820
Exon 2	39407866	T|C	Y3Y	0.0027		
Exon 2	39407896	T|G	G13G	0.0026		rs16985673
Exon 2	39407916	A|G	R20Q	0.0132	Yes	
Exon 2	39407947	T|C	G30G	0.0026		
Exon 2	39407978	A|G	**G41S**	0.0026	No	
Exon 2	39407998	G|C	P47P	0.0165		
Exon 2	39408038	T|C	R61W	0.0239	Yes	
Intron 2	39408175	G|C		0.0029		
Intron 2	39408256	A|G		0.0058		
Intron 3	39409732	T|C		0.0026		
Exon 4	39410714	C|G	**W110C**	0.0027	No	
Intron 4	39410998	T|C		0.0225		rs6072349
Intron 4	39411052	C|G		0.0304		
Intron 5	39411282	G|A		0.0179		
Intron 6	39411978	T|G		0.2549		rs6029646
Intron 6	39412063	G|T		0.0235		
Exon 7	39412412	C|G	V355L	0.0161	Yes	
Intron 7	39412493	T|C		0.0026		
Intron 7	39412628	A|G		0.0026		
Exon 8	39413957	A|G	A395A	0.0026		
Exon 9	39414271	T|C	L454L	0.0052		
Intron 9	39414286	T|C		0.0263		rs6102366
Intron 9	39414375	C|G		0.0265		
Intron 9	39414593	T|A		0.0217		
Intron 11	39414984	T|C		0.0026		rs8119824
Intron 11	39416616	C|T		0.0026		
Exon 12	39416786	A|G	**E539K**	0.0026	No	
Exon 12	39416791	T|G	E540D	0.0026	Yes	
Intron 14	39419011	G|A		0.0263		rs6102369
Intron 14	39419066	T|C		0.0026		
Intron 15	39419261	C|T		0.1421		rs6072350
Exon 17	39419954	A|G	L686L	0.0315		rs2072969
Exon 17	39419963	A|G	S689S	0.0026		rs41277020
Intron 17	39420265	T|C		0.0494		rs41277022
Intron 17	39420272	A|G		0.0027		
Intron 19	39420739	T|C		0.0186		
3prime_UTR	39421026	A|G		0.1548		
3prime_UTR	39421362	A|G		0.1538		
3prime_UTR	39421519	C|T		0.0027		
3prime_UTR	39421553	C|G		0.2158		rs6072351
3prime_UTR	39421666	A|C		0.1587		rs6065338
3prime_UTR	39421761	T|C		0.0238		
3prime_UTR	39422072	C|T		0.0026		rs2235595

Genic position	Genomic position	Minor|Major allele	Protein consequence	MAF in SIR	Detected in controls?	rs ID
3prime_UTR	39422160	A|G		0.0216		rs12625592
3prime_UTR	39422218	C|T		0.0026		
3prime_UTR	39422426	A|G		0.0027		
3prime_UTR	39422490	A|C		0.0027		rs6102378
3prime_UTR	39422595	T|C		0.0376		rs2235594
Downstream	39422749	A|G		0.0246		
Downstream	39422867	C|A		0.0093		rs41278102

Genomic coordinates are NCBI build 36 (B36).

Table A7 *LPIN* family PCR primer sequences

Gene	Genic position	Forward	Reverse	Amplicon length	Annealing temperature
LPIN1	5'UTR/Exon 1	CAGGAACGCTAAGATAGGCG	GAACCACTTCCCGTAGAGGG	404	FAIL
	Exon 2	ATGGGGGTGGATAGAACACA	GTTCAAGACAGGTGGCATCA	435	60
	Exon 3	TTTGGGTTCCATCTTTCCTG	AAATCCCCCACACAGATCAA	481	53
	Exon 4	CTCTAGCTGGGCCATCTGC	CTGGTCCCTAGTGCCAAAAA	593	60
	Exon 5	TTAGAATTCGGCCAAAGCAT	CCGTGTTTTTCCTGGTCCTA	460	60
	Exon 6	GGGCCACTTCTTTTTCTCCT	CTGGCTCCTGGTAGCTGTGT	491	60
	Exon 7	CCAGGTGATTGTTGAGGCTT	TGGAAAGGACCTGTCATGTG	514	55
	Exon 8	ACTGTGCCCAAACCTCAAAG	CCCAGCTGTCTTGCACACT	418	60
	Exon 9	ATACAGGGAGATGCAGCCAG	GCACTCCTTGGAGAAGCCTA	492	60
	Exon 10	TAAAATGGTGCGGCCTTTAC	CATGGAACCCTGACCACAGT	414	60
	Exon 11	TGCCTAGGGGTGATATGGAG	GCCTTGATAAGGAAGAGGGG	435	60
	Exon 12	TCATGTAATTTTGGACGCCAT	GAGCTGGCTCATGGAAACTC	408	60
	Exon 13	CGTCATTGGGGAATGGTCTA	AAGGACTATTGCATGCCAGG	478	60
	Exon 14	GCAAGTGAATGAGGAGAGGC	GGGTACCCAGTTGAGGGATT	496	60
	Exon 15	AAGGGTGGGGAAAGAAAGAA	AGAATTCGAACCGTGCAAGT	413	60
	Exon 16	TCGACTTGGTTGCTTCTGTG	TGCTGGGACAGGAAAGATTC	463	60
	Exon 17	GGACTTTTGTATGTGGGGGA	AGGAGCCAATGCAGGTGTAG	456	60
	Exon 18	CTCGTGCTTTTCTGGGATGT	TGGCTCTTCACCAGGCTACT	407	60
	Exon 19	TTCACTGCACCACTTTGAGG	AGACCCAGAAGGGATCTGATT	446	60
	Exon 20	CGTGTCGATAAGTAGGCGGT	TGCTTAGAAATGTCAGCCCC	459	60
	3'UTR	TCCTCTGCGCTTGTTCACTA	CCAGGGCTCACATTGAACTT	371	60
	3'UTR	AGGCGGAGTTTTCAGAGGAT	TGTGCTTACAGTGGTGATCCA	489	60
	3'UTR	TTTCATTGGCATATCTCCCC	AGGCCCTTTCGATAGTCTAACAC	453	60
	3'UTR	TGGGAGCTCTCCAAAGAAAC	GGAAAATGTTTCAATCGTTGG	463	60
	3'UTR	TAACACCTGCCTTCTGGCTT	TTGCATCCAGTACAACACAATG	420	60
	3'UTR	ACTCAAATTAAGGGCAAGCG	GATGGCAGATCTGTTTGCCT	389	60
	3'UTR	AAATGGAACGGTTGAATGAAA	AACCGTCTGAACCTTTGCAG	473	60

Gene	Genic position	Forward	Reverse	Amplicon length	Annealing temperature
*LPIN2**	Exon 1	ATCAAAAGTTGGGCATGGAG	CACACAGTGCAGGGGATG	526	60
	Exon 1	TACAATGTAATTGGAGGGAAGAGC	ACGAGGCTCTAGACTTGGTGTC	450	60
	Exon 2	CATTGTCTCAACATCATTCAGG	CGTGTGTGGAAATGCGTAGA	540	60
	Exon 3	TGTCTTGCCAACAACAGTCC	TGGTTGTCCATCTCTCCCTC	419	60
	Exon 4	GAGCTCACGCCTACATCACA	TGGCATGTCAGTTCCTTCAG	524	60
	Exon 4	AACTGCCTTTGCTGCTTGAT	CTCAAGGAAAAGCTTCCTGC	460	60
	Exon 5	GATGGCTAAATTCCATGCTGA	ATAGTGGGTGGCATTGGTGT	525	60
	Exon 6	GAAATCCTTCGGTTGCAAAT	AATAATTGATGCTCCCTTCCA	467	60
	Exon 7	CAGCCATCACGTTATGTGGA	AAGGCAATTATTAGTTTTTCTCACTT	600	FAIL
	Exon 8	TTCCAAAACCTGAAGCCATC	GGCTGGGAACCAACCTAAG	401	60
	Exon 9	CACTACCTCAAAGCCACTCCA	CCCTATCTAAGAAAAGTTATGCCTGA	545	60
	Exon 10	ACCTTCGCTTCAAGCACAAC	TCTTTCTTGTCCTGGGGAAA	520	60
	Exon 11	CCAGAGTTTGGGCATGAGAT	AATTAGCCAGTGGGCAATGA	541	60
	Exon 12	ACCTGCACGTCCTAAGTGCT	ATGAGATTTGCCCACCTGTC	508	60
	Exon 13	GTCATCTGCTGAGTGGCACA	AGAAGGAAGCTGGGGACCTA	535	60
	Exon 14	AGGTGGGCCTGATAGCTCTT	GGATATGGGAGAATGGCAGA	468	60
	Exon 15	ACCATCTCCTCCATGCAAAA	CTGTGTCTAAGACCCCGGAA	467	60
	Exon 16	GACACAGGTTCGATGCCAAT	GACTCCGCTTTCAGAAGCAC	473	60
	Exon 17	ATTGACCCAGTGCAGGTAGC	TGGTTTTCACAGTAAATGAGAGGA	515	60
	Exon 17	CCCCAGTGAGTGGTCCTAAA	GCCAGAACATGTGGGCTAAC	543	60
	Exon 18	AGGACCTCCCCAGTTGCTAT	TGGTGAAGAAAGGCAGGTTC	516	60
	Exon 19	ACAGGGCTCAAACTCCCTCT	GCTCAGTTTTGGTGGCAACT	461	60
	Exon 19 and 20	GACAGGTCATCCAGGTCCAC	TGAACCCCAAGGGTGAATTA	533	60
	Exon 20	AGGAAACATGTGTGCGACC	TGAGTGACAGCTTCACAGCC	498	60
	3'UTR	CCAAAGAGGGCCAGGTTTA	GCTGACCTCAGATACCAGCC	549	60
	3'UTR	TTCACTGTGCTTCCCTGACA	GTCAGTGATGCTGGCTGAAG	512	60
	3'UTR	AGCAGCTTATGGCACACCTT	TTAAAACAGACCAAACCCCG	491	60
	3'UTR	ATGGCTGAAATCAGTCAGGG	GTGGAAGCAGCGTCAAAGA	543	60
	3'UTR	CAAGAGGACACACAGATGGC	GTGGTTCCTGAGCAGAGAGG	505	60

Gene	Genic position	Forward	Reverse	Amplicon length	Annealing temperature
	3'UTR	TCAGCAAGAGGGACTCCTTT	AAGCTTCCCCCAGCTTAAAT	536	60
	3'UTR	AGCACACAGACAGCAAAGCA	TTCTCTTTGGACACATGGGC	509	60
	3'UTR	TTCCCAGTCATCCAATCTCC	GAGATGCCCTCGAGTGTGTT	453	60
	3'UTR	GGCTGAACATAATTATTAAAAGAGCA	GTGCTCGTGTAGACCTGGAG	511	60**
	3'UTR	AGGTTAGTTCCTGGGGGTGT	TCCCGTGTTGTAGAGCTTCC	549	60
*LPIN3**	Exon 1	TCTTTCCAGGGAGTGGAGC	CGACGTCTCCTCTCCAGACT	437	FAIL
	Exon 2	TTTAGAGGCACGAGGCAACT	GAGCACTCACCACCTTCTCC	510	60
	Exon 2	AGCTTCTTGTGGAGGAGCTG	GGATGAGCCCTACTGGTCTG	490	60
	Exon 3	GATGGGTAGGGAGCTCAGTG	CATATCACGTGTGCCAGAGG	485	60
	Exon 4	CCACACCTGTTTTCCTGACC	GCCACAAGGAAATGGTCACT	595	FAIL
	Exon 5	CCAGGGTGTGTAAGGACCAA	GAAGATGGCAAAGTTGTAGCTG	637	60
	Exon 6	TTCTTACTGGATTGTCTTCTTCTTG	CCTTCAAGGACCACTGAGGA	528	60
	Exon 7	ACACCCCAGAGACAGAGGAA	ACCTGAGCTTGTGAGCTGGA	618	60
	Exon 8	TTCAGAGTCAGAGGGCACTT	GTCCACCACAGAGGGACAGT	451	60
	Exon 9	GCTTTACTTCCCCCAAAGGT	GGAGTGTGGACTGCACAGG	462	FAIL
	Exon 10	AGTCCCCATGAAGCAAAATG	TCTCCGTGCCCTGAGTCTAT	541	60
	Exon 11	CAGCACAGCGTCTCTTACCA	CTTCCCCTCTGGTTTGTCCT	509	60
	Exons 12 and 13	AGGTGAGCACACTTCCTTGG	ACAGGGTGCTGGCTCCTC	468	FAIL
	Exon 14	TCTCCAAGGCAGACACTGAG	GCTCCCAAGGGGATAGATTC	530	60
	Exon 15	ATGGATGTGTCTGTGCATGG	TTACTCCAAGGCAGCTCCAC	471	60
	Exon 16	GTCTTCAGCGTGACCACTCA	GGATGAGCCTTTCCCAGAAC	543	60
	Exon 17	GTCCAGCTGAGTCCACTGC	GGGCCAAAGTAGGGAAGAAG	487	60
	Exon 18	CTCTGCCCTCCACAGGTAAC	TAGCCTCACGTGGATTTGTG	532	60
	Exon 19	CCTGCCTGAGTGACATCCA	GAGGAGCTCGACCACTTCAC	481	60
	Exon 20	GCTAAACCCTGCCATGTTCC	GCTAATTGCCCCAGGACAG	550	60**
	3'UTR	AAACCCACTGAAGGGGAAG	AGGGCATAGAAAACCCTGC	500	FAIL
	3'UTR	GGGGCAATTAGCTTGTCATC	TGTACCACACTCCCACTGCT	497	FAIL
	3'UTR	CCTTGCAGGGTTTTCTATGC	CTGCTTCCCACAGCTCCTTA	476	FAIL
	3'UTR	CATGACACACACCCACACCT	AAAATGCATGTTCCTCAGGC	487	FAIL

Genic position	Genomic position	Minor|Major allele	Protein consequence	MAF in SIR	Detected in controls?
	3'UTR	GTGGAGGGCAGAATACCAAA	CCCCCACTCCAAATTTATCC	460	FAIL
	3'UTR	CCTCATTCAGGCCACTTTGT	TCAGCAACTGTGCAGAGACC	539	60**

* *LPIN2* and *LPIN3* primers are M13-tailed. Forward primer sequences are preceded by TGTAAAACGACGGCCAGT and reverse sequences are preceded by AGGAAACAGCTATGACCAT. ** 7-deaza-dGTP was added to dNTP mix.

Table A8 *LPIN1* sequenom primers

Plex	SNP_ID	2nd PCR primer	1st PCR primer	MassEXTEND primer
hME1	G582R	ACGTTGGATGGTAAGCCAGAGCAGTGCTTG	ACGTTGGATGATCCCAAGAAGCCTCCTACC	GCAAGGCCCATAGCACC
hME1	R552K	ACGTTGGATGAGGCCACTGTGGAATCTATC	ACGTTGGATGTTGTGGTGTTTCTTCCCCTC	GCCCAAAAAGGGAGGAA
hME1	A353T	ACGTTGGATGAGCCTCAGACAGAAATGCAG	ACGTTGGATGGTTTGAGCTCCTCGATCATG	AGACCTGGAGACCTTAGGA
iPlex1	Novel2	ACGTTGGATGAGGTATCCGAACAGGGAAAG	ACGTTGGATGGAGACTCTGTGAAGTAGTCG	TGGCTTCTTTTCAGCAA
iPlex1	rs33997857	ACGTTGGATGCTCCCAATCTTTACCACGAG	ACGTTGGATGGGAGCAAGCTGTGTCATATC	AATAGCGGGGTTGTCCA
iPlex1	rs2577264	ACGTTGGATGCAGCCAAACTCAGAATTCCC	ACGTTGGATGTGTGCTTCACAGGCACAAG	CCATACCCTGGTGCCTTCC
iPlex1	Novel1	ACGTTGGATGAGCGTATCTGTGGAGACTTG	ACGTTGGATGAACCTGAAGGGAAAACATGG	GTCCCCATGAGGTCTCCTGT
iPlex1	rs4669778	ACGTTGGATGTGCTCATTCTTCCCGCCTAC	ACGTTGGATGCGAGAACTCTGAAGTTGTCC	GAAGGTATTCTGGGCCTGTG
iPlex1	rs2577262	ACGTTGGATGTGGTGACAAGCTGTTTGCTC	ACGTTGGATGAGGTGTCAGAATGTTTTCC	CGGGGACTATTCTGTCAATCA
iPlex1	rs13412852	ACGTTGGATGTATGTCCCATGCCTGGTTTC	ACGTTGGATGATGCAGGGACTTTGTCACAG	CCTTCCAGCCAATACTGATGTCG
iPlex1	rs6744695	ACGTTGGATGAGAGAGCAGAGCAACGATTC	ACGTTGGATGTTCCAAGACAAAGGCTCCAC	CAACGATTCAGTTATTAAAGATGC
iPlex1	rs2278513	ACGTTGGATGGAGCAAGCTTTGTCTAAAGG	ACGTTGGATGTGGCTATAAGTTCTTTGCAG	GGATTTCATTTAGTGACAGTGAGA
iPlex1	rs6729430	ACGTTGGATGAAAGGACCGTAGATTTCCTC	ACGTTGGATGAGAGAGAAATGCCAAGGGAC	CCAGGACCGTAGATTTCCTCTCCTGA
iPlex2	rs17603420	ACGTTGGATGGCATGTGTTGTGTTTTGAGG	ACGTTGGATGAACCACTCTGGAGAACATGC	GTGTTTTGAGGACCGCA
iPlex2	rs2577256	ACGTTGGATGATGCTAATATCCCTTCCCCC	ACGTTGGATGGCTAAGCAGTCTCAAAAGCC	CTTCTCCAGGCACTTCAC
iPlex2	rs3795974	ACGTTGGATGACTGTCCCAGTTCAGGGTC	ACGTTGGATGATCTGTGCTGAGATGCAGCC	CCAGTTCAGGGTCCTACATC
iPlex2	rs893346	ACGTTGGATGAGCATAGCAGCCACTGTTTC	ACGTTGGATGACAGGGAAAGAGACCTCAAC	CCCATGGAGTTCTCTCCTCCC
iPlex2	rs16857866	ACGTTGGATGACGTGTGCTGCTGATCATTC	ACGTTGGATGCTGTGTGTCTCAGGTGTAG	GCTGATCATTCAAATAGGTTT
iPlex2	rs7595221	ACGTTGGATGGGTGTATTATCTGTTTGGTTC	ACGTTGGATGCATCCCACTGGAATACCATC	CTCCAACAGTTTCTAACCCAGAG
iPlex2	rs17603350	ACGTTGGATGCAAAAAACCGAGGCTCAGAC	ACGTTGGATGTGAGCTCTTTAAGGGCCTCG	GGTAAAGTAGTCAAGGAAAGCTC
iPlex2	rs1050800	ACGTTGGATGAGTTTCTTTGGAGAGCTCCC	ACGTTGGATGTGCTGACAGTGTGGACTTGA	CAGAAGTCAGTATCATTATCATTT
iPlex3	rs893345	ACGTTGGATGTAGATAGGAGCTGCCCCAAG	ACGTTGGATGGCTTGGAGAGCTTTGTCCAG	CACTTGACCAGATGGAC
iPlex3	rs2716609	ACGTTGGATGATCTCAAGCTTTGACCCACC	ACGTTGGATGTGCCCACATCTAAAGGCAAG	AACCTTCAAAGAACCCTGGA
iPlex3	rs4669781	ACGTTGGATGACATTAGGCAGAAGAGGGAG	ACGTTGGATGTGTGTGTTCCAGGGTAAAGC	GAGGTGGCCTGCGTTTGATG
iPlex3	rs17603755	ACGTTGGATGCCGAAGTCCCATAATTGAGC	ACGTTGGATGTTAGCACTTGTCAGTGTACG	TGTCACCATACAAATAATTAAAATA
iPlex4	rs2577261	ACGTTGGATGAACTTCCTCTCGCCTTACTG	ACGTTGGATGAAATGAACACTTATGGGGTC	ATCTCCTATGCAACCTAC
iPlex5	rs4315495	ACGTTGGATGACTGTAGCATTAATTGTAAT	ACGTTGGATGCCAGAGTCATACAGATATTA	GCATTAATTGTAATAGGGAAAAT
iPlex6*	rs4669781	ACGTTGGATGTGTGTGTTCCAGGGTAAAGC	ACGTTGGATGACATTAGGCAGAAGAGGGAG	AGCGCGCAGCTGCCAAG

Plex	SNP_ID	2nd PCR primer	1st PCR primer	MassEXTEND primer
iPlex6*	rs2577261	ACGTTGGATGAAATGAACACTTATGGGGTC	ACGTTGGATGAACTTCCTCTCGCCTTACTG	CTTATGGGGTCAGAAAGC
iPlex6*	rs17603755	ACGTTGGATGCCGAAGTCCCATAATTGAGC	ACGTTGGATGTTAGCACTTGTCAGTGTACG	TGTCACCATACAAATAATTAAAATA

* = Redesigned for Ely cohort only.

Table A9 *mTOR* sequence variants detected in a cohort of severe insulin resistant patients and 11 Indian and 23 CEPH controls

Genic position	Genomic position	Minor|Major allele	Protein consequence	MAF in SIR	Detected in controls?	rs ID
Upstream	11245215	C|A		0.118		rs2295080
Upstream	11245152	G|C		0.116		rs2295079
5prime_UTR	11245131	A|C		0.013		
Intron 1	11242153	A|G		0.0395		rs17229137
Intron 2	11241394	G|C		0.004		rs28730695
Intron 2	11241375	T|A		0.328		
Intron 2	11241350	G|C		0.003		rs2076657
Intron 3	11241106	A|C		0.008		
Intron 3	11239989	T|C		0.01		
Intron 3	11239920	A|G		0.43		
Intron 3	11239907	G|A		0.011		rs2092642
Intron 3	11239897	A|G		0.01		rs12121319
Intron 4	11239523	A|G		0.011		
Intron 4	11238929	A|G		0.011		
Intron 4	11238912	C|G		0.005		
Intron 4	11238849	T|-		0.005		
Intron 5	11238610	G|A		0.005		
Intron 5	11236649	T|C		0.0026		
Exon 7	11230691	T|C	H296H	0.006		
Intron 7	11230378	A|G		0.024		
Exon 9	11225807	C|G	V455L	0.004	No	
Intron 9	11225740	T|G		0.0195		rs12141961
Intron 9	11225674	A|G		0.0375		rs28730692
Intron 9	11224410	A|G		0.005		
Exon 10	11224301	C|T	D479D	0.301		rs1135172
Intron 11	11222925	C|T		0.017		rs17848533
Intron 11	11221377	A|G		0.2735		rs2024627
Intron 11	11221326	G|A		0.016		rs12132215
Intron 11	11221279	G|C		0.007		
Intron 12	11220965	A|C		0.007		
Intron 12	11220814	T|G		0.004		
Intron 13	11217061	A|T		0.247		rs718206
Intron 14	11216229	C|T		0.005		rs17229172
Intron 14	11216164	T|C		0.004		rs28730690
Exon 15	11216130	G|C	A778G	0.003	Yes	
Exon 15	11216051	G|A	E804E	0.003		
Intron 15	11215215	G|A		0.041		
Intron 16	11214996	T|C		0.006		
Intron 16	11214865	A|G		0.017		
Intron 17	11213751	-|TGT		0.016		
Intron 18	11213456	T|C		0.011		
Exon 19	11211537	A|G	L935L	0.004		
Exon 19	11211345	C|T	N999N	0.253		rs1064261
Intron 19	11211248	C|G		0.005		
Intron 19	11198940	T|G		0.0275		rs28730687

Genic position	Genomic position	Minor|Major allele	Protein consequence	MAF in SIR	Detected in controls?	rs ID
Intron 19	11198928	A|G		0.0275		rs28730686
Intron 20	11198706	T|-		0.0405		
Intron 21	11196007	C|A		0.008		rs1211695
Intron 21	11196005	C|A		0.026		rs12116957
Intron 21	11195578	T|C		0.004		
Intron 22	11195204	A|G		0.021		
Intron 22	11195180	G|A		0.007		
Exon 23	11195055	C|G	R1154R	0.05		rs17036536
Exon 24	11193482	C|T	D1210D	0.005		
Intron 25	11191890	G|T		0.011		
Intron 25	11187415	G|T		0.024		rs2273127
Intron 26	11182424	A|G		0.004		
Intron 26	11182417	G|T		0.005		rs11121697
Intron 30	11133037	A|C		0.0235		
Intron 30	11132980	G|A		0.0105		
Intron 30	11132924	C|T		0.01		
Intron 30	11132918	A|G		0.005		rs28730683
Intron 32	11129279	A|T		0.005		
Intron 32	11129277	A|T		0.034		rs2275942
Intron 32	11127797	T|C		0.002		
Exon 33	11127645	G|A	A1577A	0.004		rs1057079
Intron 35	11122165	A|C		0.2845		
Intron 35	11122145	A|G		0.011		
Intron 35	11122128	T|C		0.0263		rs17848548
Intron 35	11122105	G|A		0.05		rs28730682
Intron 36	11121736	G|A		0.0265		rs2275528
Intron 36	11121717	-|AT		0.021		
Intron 36	11121582	A|G		0.0455		rs17417751
Intron 36	11121571	T|C		0.006		
Intron 36	11117178	T|C		0.021		rs3730381
Intron 36	11117169	G|C		0.003		
Intron 37	11116856	A|C		0.022		
Intron 37	11115995	A|G		0.0175		rs12122483
Intron 37	11115993	A|G		0.0175		rs17235612
Intron 37	11115988	G|T		0.0215		rs12141233
Intron 38	11115543	C|T		0.019		rs2275526
Intron 38	11115495	C|T		0.0045		
Intron 38	11115376	C|G		0.007		
Intron 38	11115280	C|T		0.005		
Intron 38	11114926	A|G		0.016		
Intron 38	11114721	A|C		0.003		
Intron 38	11114509	G|A		0.235		rs1417131
Intron 38	11114072	G|C		0.005		
Intron 38	11113819	T|C		0.038		rs12732063
Exon 39	11113317	T|C	A1832A	0.044		rs17848553
Exon 39	11113233	T|C	S1851S	0.281		rs2275527
Intron 39	11113114	A|G		0.007		
Intron 39	11112638	A|T		0.046		
Intron 39	11112511	A|G		0.005		
Intron 40	11112314	G|A		0.005		

Genic position	Genomic position	Minor|Major allele	Protein consequence	MAF in SIR	Detected in controls?	rs ID
Intron 40	11111794	T|C		0.005		rs3730378
Intron 40	11111778	T|C		0.029		rs3730379
Intron 40	11111757	A|G		0.004		
Intron 40	11111664	G|A		0.016		rs3730380
Intron 41	11111486	A|G		0.005		
Intron 41	11111469	T|G		0.016		rs17848558
Intron 41	11111292	T|C		0.047		
Intron 41	11111215	A|G		0.004		
Intron 41	11111213	A|G		0.004		
Intron 42	11111081	A|G		0.005		
Exon 43	11110729	A|G	T1984T	0.006		
Intron 43	11110599	A|G		0.005		
Intron 45	11109484	C|T		0.032		rs3737611
Exon 47	11107180	C|T	L2208L	0.007		
Intron 47	11104816	G|A		0.005		
Exon 48	11104650	T|C	L2261L	0.003		
Intron 48	11104572	T|C		0.032		
Intron 48	11104044	A|C		0.0215		rs17235633
Exon 49	11103914	G|A	L2303L	0.1735		rs1112169
Intron 50	11098268	T|C		0.007		
Intron 50	11098129	G|T		0.005		
Intron 52	11097438	T|C		0.01		rs2275523
Intron 53	11096918	G|A		0.0227		rs17235654
Intron 53	11095790	G|A		0.005		rs17229256
Intron 54	11092376	C|T		0.005		
Intron 55	11092263	G|A		0.0705		rs2275525
Exon 56	11092007	T|C	D2485D	0.01		
Intron 56	11090972	G|T		0.005		
Intron 57	11090233	G|A		0.005		
3prime_UTR	11090104	T|C		0.005		
3prime_UTR	11089889	-|A		0.01		
3prime_UTR	11089831	A|G		0.005		
3prime_UTR	11089733	T|C		0.021		rs12139042
3prime_UTR	11089300	G|A		0.019		rs2536

Genomic coordinates are NCBI build 36 (B36).

Table A10 *Rictor* sequence variants detected in a cohort of severe insulin resistant patients and 11 Indian and 23 CEPH controls

Genic position	Genomic position	Minor|Major allele	Protein consequence	MAF in SIR	Detected in controls?	rs ID
Upstream	39110453	T|C		0.3966		rs6869095
Upstream	39110425	T|C		0.003		
Upstream	39110388	G|A		0.0027		
Upstream	39110376	A|G		0.0027		
Upstream	39110365	A|G		0.0027		
Upstream	39110328	C|A		0.0027		
Exon 1	39110229	A|G	A3V	0.0054	No	
Intron 1	39110173	C|T		0.0027		
Intron 1	39110053	C|G		0.3655		rs2303808
Intron 2	39057126	C|T		0.302		rs13177690
Intron 4	39039278	A/-		0.017		
Intron 4	39039245	G|C		0.003		
Intron 4	39038633	G|A		0.003		
Exon 5	39038462	G|A	R108R	0.003		
Intron 5	39032802	A|G		0.021		rs2548796
Intron 8	39017548	G|C		0.004		
Intron 8	39017167	A|G		0.003		
Intron 8	39017144	-|T		0.037		
Intron 8	39017022	G|A		0.006		
Intron 8	39016743	G|A		0.003		
Intron 8	39016629	A|G		0.003		
Intron 8	39016620	T|C		0.007		
Intron 8	39016444	T|G		0.004		
Intron 8	39014597	C|G		0.011		
Intron 9	39014386	T|G		0.001		
Intron 9	39014355	T|C		0.003		
Intron 9	39014354	G|A		0.003		
Intron 9	39011565	C|T		0.007		
Intron 9	39011487	T|C		0.003		
Intron 10	39008092	A|G		0.027		rs10440634
Intron 11	39007669	A|T		0.011		rs7732615
Intron 11	39007666	G|A		0.008		
Intron 11	39006764	A|G		0.003		
Intron 11	39006641	C|G		0.003		
Intron 11	39006506	A|G		0.002		
Intron 11	39006439	G|T		0.182		
Intron 11	39006390	G|A		0.139		
Intron 12	39003781	A|G		0.0026		
Intron 13	39003134	A|G		0.024		rs41271107
Intron 15	39000949	G|A		0.419		
Exon 17	38998810	C|T	I497I	0.003		
Intron 17	38998551	G|A		0.003		
Intron 18	38998300	T|C		0.017		rs7729745
Intron 21	38995551	A|G		0.021		rs2115946
Intron 21	38995205	G|A		0.007		

Genic position	Genomic position	Minor|Major allele	Protein consequence	MAF in SIR	Detected in controls?	rs ID
Exon 23	38994634	G|A	E745E	0.007		
Exon 23	38994622	C|T	N749N	0.007		
Intron 23	38994507	T|C		0.491		rs6868087
Intron 23	38994500	G|A		0.003		
Intron 23	38994406	G|A		0.003		
Intron 24	38993701	G|A		0.003		
Exon 25	38993577	A|T	I811I	0.003		
Intron 25	38991719	C|T		0.008		
Intron 25	38991591	T|G		0.003		
Exon 26	38991553	T|C	S837F	0.299	Yes	rs2043112
Intron 26	38991435	C|G		0.003		
Intron 26	38991401	T|C		0.001		
Intron 26	38991377	A|G		0.004		
Intron 26	38990751	A|G		0.018		rs9292726
Intron 27	38990582	A/-		0.011		
Intron 29	38988818	C|T		0.014		
Exon 31	38986533	C|T	S1058S	0.033		rs2115949
Exon 31	38986023	T|C	G1228G	0.003		
Intron 31	38985522	C|A		0.006		
Exon 33	38982400	C|T	I1442I	0.002		
Intron 34	38981000	C|T		0.004		
Intron 36	38978913	G|A		0.0107		
Intron 36	38978644	T|G		0.0292		
Intron 36	38978564	T|C		0.0026		
Exon 38	38978206	T|C	T1695I	0.017	Yes	
3prime_UTR	38978160	G|T		0.003		
3prime_UTR	38977943	A|C		0.017		
3prime_UTR	38977847	A|G		0.02		rs1975089
3prime_UTR	38977184	-|C		0.011		rs3214763
3prime_UTR	38977096	A|G		0.004		
3prime_UTR	38977035	T|C		0.015		
3prime_UTR	38976615	C|A		0.029		rs443039
3prime_UTR	38976567	A|C		0.014		
3prime_UTR	38975560	C|T		0.003		
3prime_UTR	38975492	T|C		0.019		rs16867885
3prime_UTR	38975361	T|C		0.028		rs637747
3prime_UTR	38975147	G|C		0.004		
3prime_UTR	38974902	T|-		0.006		
3prime_UTR	38974878	T|A		0.003		
3prime_UTR	38974664	T|C		0.003		
3prime_UTR	38974518	G|A		0.003		
3prime_UTR	38974421	C|T		0.053		rs10941413
Downstream	38973706	T|C		0.003		

Genomic coordinates are NCBI build 36 (B36).

Table A11 *GβL* sequence variants detected in a cohort of severe insulin resistant patients and 11 Indian and 23 CEPH controls

Genic position	Genomic position	Minor|Major allele	Protein consequence	MAF in SIR	Detected in controls?	rs ID
5prime_UTR	2196052	-|C		0.386		rs34390502
Intron 2	2196327	T|C		0.421		rs27699
Intron 2	2196331	A|C		0.029		
Intron 3	2196462	G|C		0.366		rs26863
Exon 4	2196583	T|C	A88V	0.006	No	
Intron 4	2196690	A|G		0.006		
Exon 5	2197106	C|G	P137P	0.372		rs26862
Intron 6	2198188	T|C		0.0277		
Exon 9	2198768	A|G	S289S	0.007		rs11863256
Exon 9	2198798	C|G	E299D	0.006	No	

Genomic coordinates are NCBI build 36 (B36).

Table A12 *MAPKAP1* sequence variants detected in a cohort of severe insulin resistant patients and 11 Indian and 23 CEPH controls

Genic position	Genomic position	Minor|Major allele	Protein consequence	MAF in SIR	Detected in controls?	rs ID
Intron 2	127472130	C|A		0.0058		rs7039012
Intron 2	127472128	C|T		0.0067		
Intron 2	127472119	G|A		0.0119		
Intron 2	127472034	A|G		0.0268		
Intron 3	127471810	A|G		0.0114		
Intron 4	127452497	G|A		0.4042		rs631287
Exon 5	127387762	T|C	D188D	0.005		
Exon 5	127387705	T|C	I207I	0.005		
Intron 5	127367562	G|T		0.0053		
Exon 6	127361802	T|C	S260N	0.0161	No	
Intron 6	127361648	T|A		0.0283		
Intron 6	127361592	T|C		0.3947		rs562004
Exon 7	127345211	T|C	K302K	0.0069		rs11542134
Intron 7	127323575	A|G		0.0058		
Intron 7	127308724	G|T		0.4843		rs7875713
Exon 8	127286607	A|G	H345H	0.2173		rs2070113
Intron 8	127286442	C|T		0.0054		
Intron 8	127273911	A|G		0.0111		rs10986771
Intron 8	127273723	T|C		0.0063		
Intron 8	127273602	A|G		0.0062		
Intron 8	127273601	C|A		0.0079		
Intron 8	127273327	A|G		0.271		rs7018948
Intron 8	127273302	T|C		0.0112		
Intron 8	127273091	G|A		0.008		
Intron 8	127270219	C|A		0.027		
Intron 9	127270063	C|A		0.009		
Intron 9	127269936	T|G		0.0119		
Intron 10	127246538	C|G		0.0633		rs907501
Intron 10	127246521	A|G		0.0063		
Intron 10	127246488	T|C		0.0053		
Intron 10	127246422	T|C		0.0163		
3prime_UTR	127240959	T|C		0.0053		
3prime_UTR	127240919	A|G		0.0054		rs7864593
3prime_UTR	127240660	C|T		0.0054		
3prime_UTR	127240169	A|G		0.009		
3prime_UTR	127240120	G|C		0.3908		rs12202
3prime_UTR	127239694	T|C		0.3674		rs1129

Genomic coordinates are NCBI build 36 (B36).

Table A13 *AS160* sequence variants detected in a cohort of severe insulin resistant patients and 11 Indian and 23 CEPH controls

Genic position	Genomic position	Minor|Major allele	Protein consequence	MAF in SIR	Detected in controls?	rs ID
Exon 1	74953821	G|C	P28P	0.255		rs7327548
Exon 1	74953603	T|C	A101V	0.075	Yes	
Intron 1	74953398	A|G		0.249		rs9573565
Intron 1	74953322	G|A		0.003		
Exon 2	74834347	A|G	R299Q	0.0104	No	
Intron 2	74832061	C|T		0.005		
Exon 3	74831989	T|C	R363X	0.0053	No	
Intron 4	74821464	C|A		0.0058		
Exon 10	74798404	T|A	N655Y	0.0052	No	
Intron 10	74797597	G|T		0.358		
Intron 10	74796597	G|T		0.3066		rs2297210
Intron 10	74796580	G|A		0.005		
Intron 12	74785128	A|G		0.4821		rs2274757
Exon 13	74785004	G|A	T752A	0.0053	Yes	
Exon 13	74784903	G|C	N785K	0.0106	No	
Intron 13	74782291	T|C		0.3947		rs2297203
Exon 14	74782217	G|A	V819I	0.14	Yes	rs1062087
Intron 14	74778682	T|C		0.022		rs17064121
Intron 14	74778669	G|A		0.005		
Intron 14	74778668	T|C		0.323		rs2297209
Exon 16	74778517	A|G	K895K	0.005		
Exon 16	74774390	C|T	L967L	0.3333		rs2297208
Intron 16	74771788	T|C		0.4107		rs2297207
Intron 16	74767163	G|A		0.003		
Intron 18	74766937	T|C		0.021		
Exon 19	74764285	T|C	T1147M	0.1	Yes	rs9600455
Intron 19	74764193	A|G		0.3		rs471822
Intron 19	74761316	G|C		0.1636		rs7332292

Genomic coordinates are NCBI build 36 (B36).

Table A14 *Raptor* sequence variants detected in a cohort of severe insulin resistant patients and 11 Indian and 23 CEPH controls

Genic position	Genomic position	Minor|Major allele	Protein consequence	MAF in SIR	Detected in controls?	rs ID
5prime_UTR	76133375	T|C		0.2838		rs11547301
5prime_UTR	76133439	A|G		0.0026		
5prime_UTR	76133741	T|C		0.0026		
5prime_UTR	76133764	A|G		0.1825		rs12602885
5prime_UTR	76133969	A|G		0.0135		
Exon 1	76134114	C|T	F30F	0.0967		
Intron 1	76213908	A|G		0.0687		rs17848677
Exon 2	76214157	G|C	T78T	0.2005		rs17848685
Intron 2	76214241	A|C		0.0027		
Intron 2	76231933	T|C		0.0355		
Intron 2	76231989	G|A		0.0053		
Intron 2	76231998	A|G		0.1078		
Intron 2	76232034	T|C		0.2921		rs6565469
Intron 2	76232105	A|T		0.0026		
Intron 3	76232235	G|C		0.013		
Intron 3	76232272	A|C		0.0026		
Intron 3	76296115	G|A		0.1326		rs9900445
Intron 3	76296185	A|G		0.3844		rs2306690
Intron 3	76296221	T|C		0.0026		
Intron 4	76296527	T|C		0.0026		
Intron 4	76297225	G|A		0.2052		rs12450876
Intron 4	76297351	A|G		0.2026		rs12453034
Intron 4	76297497	C|G		0.0894		rs34461761
Intron 4	76318494	A|G		0.307		rs9896771
Intron 4	76318562	T|C		0.0053		
Intron 4	76318707	A|G		0.0026		
Intron 4	76318751	C|T		0.2827		
Intron 4	76318835	C|A		0.2026		rs12946115
Exon 5	76319006	A|G	G187S	0.0026	No	
Exon 5	76319008	G|C	G187G	0.0053		
Intron 5	76319108	A|G		0.0026		
Intron 5	76319162	A|G		0.0027		
Intron 5	76319173	A|G		0.0027		
Intron 5	76342329	C|T		0.2159		rs12944923
Intron 5	76342351	T|G		0.0026		
Intron 5	76342387	A|G		0.0026		
Exon 6	76342542	G|C	T264T	0.0026		
Intron 6	76342685	A|G		0.0026		
Intron 6	76342690	T|A		0.0081		
Intron 6	76379675	C|A		0.0629		rs17848624
Intron 6	76379814	A|G		0.2708		rs4969429
Intron 7	76410463	G|A		0.3931		rs9911574
Intron 8	76410728	A|G		0.0106		
Intron 9	76411661	A|T		0.1382		rs17848690
Intron 9	76412007	T|C		0.1961		rs9895174

Genic position	Genomic position	Minor|Major allele	Protein consequence	MAF in SIR	Detected in controls?	rs ID
Intron 9	76426092	A|G		0.1082		
Intron 10	76434762	T|C		0.0502		
Intron 10	76434807	G|A		0.3783		rs3829572
Exon 11	76434924	C|T	G423G	0.3617		rs3751945
Exon 11	76434969	A|G	Q438Q	0.1058		rs2589156
Intron 11	76434991	T|C		0.0026		
Intron 11	76435091	T|C		0.0053		
Intron 12	76443970	A|G		0.2154		rs12943041
Intron 12	76444079	G|T		0.2608		rs9895380
Intron 12	76444126	T|C		0.206		rs17848633
Intron 12	76446117	C|T		0.0345		
Intron 13	76468651	G|A		0.4563		rs6420478
Intron 13	76468684	A|G		0.0027		
Intron 13	76468733	A|G		0.0026		
Intron 13	76468759	T|C		0.0026		
Exon 14	76468818	G|A	Q506Q	0.2526		rs2289759
Intron 14	76468905	C|T		0.0052		
Intron 14	76468911	T|C		0.2552		rs2289760
Intron 14	76468946	A|G		0.0026		
Intron 14	76468971	T|C		0.0052		
Intron 14	76468972	A|G		0.0026		rs2289761
Intron 14	76469072	T|C		0.0028		
Intron 14	76469082	A|G		0.4692		rs2248843
Intron 14	76469091	T|C		0.1075		rs12937510
Intron 14	76471640	G|A		0.0055		
Intron 14	76471697	C|T		0.0053		
Exon 15	76471873	A|G	T548T	0.0053		rs34848699
Intron 15	76471913	T|C		0.0027		
Exon 16	76472295	A|G	S590S	0.0058		
Intron 16	76472480	A|G		0.238		rs11656246
Intron 16	76473364	T|C		0.0085		
Exon 17	76473438	T|C	F626F	0.0028		
Exon 17	76473495	T|C	A645A	0.0028		
Intron 17	76473599	T|C		0.2926		rs2016817
Intron 17	76473704	T|C		0.0028		
Intron 17	76473705	A|G		0.2926		rs2289762
Intron 17	76479975	C|G		0.0078		
Intron 17	76480086	G|C		0.1349		rs2289763
Exon 18	76480141	C|T	L670L	0.2526		rs2289764
Exon 18	76480225	G|A	A698A	0.1349		rs2289765
Intron 18	76480328	T|C		0.0052		
Intron 19	76481354	G|C		0.0027		
Intron 20	76482307	T|C		0.0029		
Intron 20	76482328	G|A		0.0143		rs3751943
Intron 21	76497401	A|G		0.335		rs1468029
Intron 21	76497420	A|G		0.1492		rs12936933
Intron 21	76497435	G|A		0.3246		rs1468030
Intron 21	76511083	T|C		0.3345		rs2271602
Exon 22	76511124	T|C	T842T	0.2919		rs2271603
Exon 22	76511182	A|G	A862T	0.0036	No	

Genic position	Genomic position	Minor|Major allele	Protein consequence	MAF in SIR	Detected in controls?	rs ID
Intron 22	76511246	T|C		0.0037		
Intron 22	76511357	T|C		0.3219		rs2271605
Intron 22	76511363	T|C		0.1718		rs2271606
Intron 23	76512142	T|C		0.304		rs7217786
Intron 23	76512156	C|T		0.1715		rs6565491
Intron 23	76512182	A|G		0.0423		rs6565492
Intron 23	76513718	A|G		0.0084		
Exon 24	76513818	A|G	T954T	0.0028		
Intron 24	76513913	T|C		0.0112		
Intron 24	76528840	T|G		0.3453		rs17848659
Exon 25	76528905	T|C	H978H	0.0054		
Exon 25	76528961	A|G	R997H	0.0031	No	
Intron 25	76529088	T|C		0.003		rs8082065
Intron 25	76529118	T|C		0.0059		
Intron 25	76529148	C|T		0.0089		rs7211235
Intron 25	76529243	G|A		0.0031		
Intron 25	76529343	G|A		0.174		rs6420480
Intron 25	76529346	A|G		0.1836		rs908237
Intron 25	76529356	C|A		0.2659		rs17848664
Exon 26	76534153	T|C	A1039A	0.3521		rs1567962
Intron 26	76534210	T|C		0.0026		
Intron 26	76534246	A|G		0.0053		
Intron 26	76534432	C|G		0.3567		rs2138119
Intron 26	76534454	G|C		0.1729		rs2271610
Intron 26	76534487	A|G		0.4486		rs2271611
Intron 26	76534515	T|C		0.4805		rs2271612
Intron 27	76536814	C|T		0.25		rs2138123
Intron 27	76536867	T|C		0.0133		
Intron 27	76537027	A|G		0.0053		
Intron 27	76537072	T|C		0.2005		rs2138124
Intron 27	76537073	A|G		0.0054		
Intron 27	76537150	C|T		0.3005		rs8082382
Intron 27	76537179	G|T		0.2121		rs8082395
Intron 27	76537721	G|T		0.0267		rs35638552
Intron 27	76537773	T|C		0.3972		rs3751940
Exon 28	76537938	A|G	T1122T	0.0026		
Intron 28	76538065	G|T		0.0026		
Intron 28	76538155	A|G		0.0028		
Intron 28	76546009	G|A		0.0135		
Intron 29	76548346	T|C		0.0086		
Intron 29	76548432	A|G		0.0029		rs4436840
Exon 30	76548509	T|C	L1172L	0.0029		
Intron 30	76548629	C|G		0.0174		
Intron 30	76548683	A|G		0.0294		
Intron 30	76548735	A|G		0.0174		rs9889472
Intron 30	76548765	T|C		0.0029		
Intron 30	76549666	A|G		0.311		rs9897319
Intron 30	76549670	A|G		0.0058		
Intron 30	76549691	A|C		0.0087		
Intron 30	76549710	A|G		0.0087		

Genic position	Genomic position	Minor|Major allele	Protein consequence	MAF in SIR	Detected in controls?	rs ID
Intron 30	76549715	C|T		0.0029		
Exon 31	76549792	T|C	R1203R	0.3411		rs9899178
Exon 31	76549870	C|T	S1229S	0.0088		
Intron 31	76549896	T|G		0.0029		
Intron 32	76551241	C|T		0.0068		
Intron 32	76551247	T|C		0.0103		
Intron 32	76551300	T|C		0.0068		
Intron 33	76551484	T|C		0.0035		
Intron 33	76552592	A|C		0.0069		
Intron 33	76552606	A|C		0.1758		rs3751937
3prime_UTR	76552729	T|C		0.0172		
3prime_UTR	76552799	C|G		0.1938		rs3751936
3prime_UTR	76552818	T|C		0.0034		
3prime_UTR	76552901	A|G		0.0034		
3prime_UTR	76552977	T|C		0.0055		
3prime_UTR	76552980	A|G		0.1868		rs11547302
3prime_UTR	76553093	A|C		0.4093		rs3751934
3prime_UTR	76553106	A|G		0.0054		
3prime_UTR	76553134	A|G		0.0027		
3prime_UTR	76553167	A|G		0.0333		
3prime_UTR	76553168	T|A		0.0274		rs3751933
3prime_UTR	76553250	A|G		0.0054		
3prime_UTR	76553338	T|C		0.0053		
3prime_UTR	76553354	T|C		0.0026		
3prime_UTR	76553370	A|G		0.0054		
3prime_UTR	76553468	T|C		0.0053		
3prime_UTR	76553485	G|A		0.0026		
3prime_UTR	76553497	G|A		0.0053		
3prime_UTR	76553509	T|C		0.0053		
3prime_UTR	76553545	A|G		0.0026		
3prime_UTR	76553635	A|G		0.0113		
3prime_UTR	76553680	C|T		0.0027		
3prime_UTR	76553730	T|C		0.0026		
3prime_UTR	76553996	A|G		0.0081		
3prime_UTR	76554009	C|T		0.0882		rs3751932
3prime_UTR	76554056	A|G		0.0027		
3prime_UTR	76554120	G|A		0.0027		
3prime_UTR	76554236	T|C		0.0086		
3prime_UTR	76554312	G|A		0.0028		
3prime_UTR	76554452	C|T		0.4099		rs1062935

Genomic coordinates are NCBI build 36 (B36).

Table A15 mTORC PCR primer sequences

Gene	Genic position	Forward	Reverse	Amplicon length	Annealing temperature
mTOR	Exon 1	GCAATACCAGAGCTGATGGG	CGGGTCCTGAAGCTTCTCTT	476	60
	Exon 2	ATGGATGTGACAGGTTGGGT	CCTTTTTAGAATCCCCTGCC	442	60
	Exon 3	AAGCCACGGGCTTCTTAAA	TCAGATGATTGCAAGTTCAGG	415	60
	Exon 4	TTTCCATGACAACTGGGTCA	GTAGAATGGAGGGTTCACTCG	554	60
	Exon 4	CCACCACACCATGCTAGATTCC	TCATCACCTTTTGCCTTCCTC	440	60
	Exon 5	GAGAGCAAACCAAAACGTGA	TTGCTTTCCTTCCTCTGTCC	478	60
	Exon 6	CCTGGGCAATAGAGCAAGAC	CCCCTCCCCATCACTCTCTA	545	60
	Exon 7	CACTGGCACACCTGAGAGAG	CCACCCTTCACTTTGGAAGTA	499	60
	Exon 8	GCGTTGCTTCAAAGGAAAAG	GGTACAGCTCTCACCAAGGC	404	60
	Exon 9	GCAAAACTAGCAGATTCCCAGTA	TGAATTGCTTTATAGGACCTTTG	550	60
	Exon 10	TCCAGTTGAATGTGCAAACA	GTGGTAGCCCTGCCTGAGTA	343	60
	Exon 11	CTTTCCCTCAAACCATTTGC	CTTGCTGGGAGAGAAAGCTG	554	60
	Exon 12	TATCAGCTTTTTGAGCCGAA	GAGTTCCGGGTCAGTATCGT	475	60
	Exon 13	CGCTTCCTCAAGCAGTTCTC	CGTGTTAGGCATGGCTACTGT	497	60
	Exon 14	CTGAAACACTCCCATCTCCC	CTCTGCTTTAGAGAGGGCCA	472	60
	Exon 15	GCTGGGTTCCGCAATAAATA	TCACATTATGGTGCTTCCCA	433	60
	Exon 16	CTTCATAGGCAGTGGCCAAG	CGGCCTCCAAAATACTTTCT	481	60
	Exon 17	CAAAATTGGAGAGGGACAGG	CAGTGATTGTTGCAGGGTTG	461	60
	Exon 18	TTCCGGGATTCAAGAAGAAA	GGACGATGGCAAGAGAGAGA	499	60
	Exon 19	GCACAATTAAGAAGCTGAAGCA	GACCTTGAACTCCCAACAGTG	487	60*
	Exon 20	TCTCTTCAGCCTTCATGCCT	CCTCCAAACCTAGCCAAGC	470	60
	Exon 21	GGCATCAACCTGTCACTCAG	GAGGAGTTGATCTCGGCG	398	60
	Exon 22	AGAAAGATGGCCTGGGAACT	CCAGCAACGGATTCCTTATC	434	60
	Exon 23	TTCCACGTTCTCTGATGGTG	TGGATCCATGTCATCCAAAA	414	60
	Exon 24	GCCAAAAAGCTACACAAACTCC	ACAGAGCTAGCCACAAACCC	309	60
	Exon 25	GTTTTCGGTTGCCCAGTTTA	TTTGCTAGTTGCATGGTTGC	492	60
	Exon 26	CCAGCCCCTTGATTATTACTTC	CCAGCACAAAGGAGTTGTGA	460	60

Gene	Genic position	Forward	Reverse	Amplicon length	Annealing temperature
	Exon 27	TCTCACCCAGCAGAACAATG	GCACTGGTCTGTATAGCACTGG	480	60
	Exon 28	GTGACTCCAGTGAAGTGGCA	CAAGGTGAGACTTTTCCCCA	373	60
	Exon 29	TGAAGACTGGTAAGTCAGCATCA	CCACAGAGCAGCAAGATGAG	462	60
	Exon 30	AAGTGAGAACTCCGTGTGGG	AGCAGCACTCCTCCAGATTG	412	60
	Exon 31	TACTCTTCTGATGCGAGCCC	GCCCAGAATTCTTTGGCATA	486	60
	Exon 32	GCTTCTTTACCAAAGCACCG	ATAGGGGCTCTGCAGGATTT	360	60
	Exon 33	AAGCAAATCGCATTCCAAAC	TTCAGTTCTGGGAAAATGCC	438	60
	Exon 34	GGCAGGAAAAGCAAGTTGAG	GTTACAGTCGGGCATATGGG	447	60
	Exon 35	GCCTGTAATCCCCGCTACTC	GGGCACTCTTCCACATGT	528	60
	Exon 36	ACAGGAAGAGACTCCCTGACA	CCTGGCTCAAGTATGCAAGC	410	60
	Exon 36	CCCTCCTGGTTTCAGCATTA	TGTTGCTCCTGGGAGTTGAT	562	60
	Exon 37	CCTAGGAAAGCAAGTGGCAG	GGTCCTCTGTCCTATGGGGT	455	60
	Exon 38	ACTTGTAGGGGGAAATGCAG	TGAGAGACAAAGGTCAAGATGG	519	60
	Exon 39	AGCTCCATCTGGCACTTCAG	TCCTTGAAGTGTTTGCTTTATGG	499	60
	Exon 40	GGCCCAGTCAGCTGTTACTC	CCTGTTCAGGAAGCCACTGT	496	60
	Exon 41	GGACACTTGACACTGGGACC	GTGACCTGGGATGAAATGCT	469	60
	Exon 42	AGACTACCAGGAAGGGGCTC	GGGAAGGGAATGAACCATTT	435	60
	Exon 43	GTCAGAGGAAGTGCACAGCA	AGCCCCTTCCTGGTAGTCTC	363	60
	Exon 44	CTTGGGTCACGTCCTTTCAT	TTTTTCTTCCTCGCATTGGT	419	60
	Exon 45	CTGCCTCCAGGGAAGAATTT	ATGCTCTTGCTTTCTCCAGC	462	60
	Exon 46	TTTCAGAAGAGGGAAGGGGT	TGTGTTCCGACGAATCTCAA	496	60
	Exon 47	CATGACTACACGAGACAAATGTAGG	TCATTTCCATGCCTAACCGT	470	60
	Exon 48	TCTCCATATGGCCAGTGCTT	TCTGCCTGTGTTCTGAGCTG	506	60
	Exon 49	TGTCTTGCTCACCCATTTCA	TCTCCTTTGGCTATGATGGG	411	60
	Exon 51	ACAGTGTCAGAGGAGGGGG	TGATTCACACATAAGCCCACA	479	60
	Exon 52	TTTGGAGAACTGATGGGCTC	GTTCCCATGCCCTCTGAGTA	478	60
	Exon 53	CCAAATGGGATCAGGACAGT	CAAGCAACTCCTCTGCCTTG	557	60
	Exon 54	GAGCCTCTGTAACGTCCTGC	CCTTAGGGGATGTTCACCAA	472	60
	Exon 55	CCTTTTCTGCTCAAAGGCAG	TCCCCTTTAGGGTAGGTAGGG	322	60

Gene	Genic position	Forward	Reverse	Amplicon length	Annealing temperature
	Exon 56	TGCCAAAGCTCGTCACTAAC	TGCCTTTGAGCAGAAAAGGT	366	60
	Exon 57	AACAATGGGCACATGCAGTA	TCAAATCCAACTATGATGGTATCC	407	60*
	Exon 58	CCCATGTTGAGAGGAGCAAC	GTGGGCATTAGCCTTGACTG	570	60
	3'UTR	GTGATCCCCTCTGTGCATCT	GAAACACAAGGCTTGATTTGGT	488	60
	3'UTR	CTTCCCTGTGTTCAGCACCT	TGGGTACAGCAAACTCAGCA	437	60
	3'UTR	CACCCTTACGTCCTCTGTCA	GATGCACAGAGGGGATCACT	409	60
Rictor	Exon 1	CACACAGCCCCAATTAGGAT	GCATCAATGGGACTTTCCTC	476	60*
	Exon 3	TCCATGAGAAGCAGGAAAGAA	CCATGGGTACAGGGGAGTAG	439	60
	Exon 4	TTTGAACATTCACTTTGGTTCTTC	CTGGCAGGTCAAATGAGAAA	469	60
	Exon 5	ATGGCAGGCATGCTAAAAAC	CAGAAGGTAAGAAAGAACTCAATAACA	454	60
	Exon 6	TTGCCCTACCTATACCCAATCA	TGACAGATATTTATCCCTCGAGATG	466	60
	Exon 7	TTGACTACTATTGTCCCCTCTGG	TTCATTACTTGAAGAAAATTCTGACC	504	60
	Exon 8	TGTCGGCTAATTTAGGCTTGA	TTAAATATCAGGTTGTGGGGAAGT	486	60
	Exon 8	GCCCTGTTACTTTTGACTAACTGT	CGACAGTATGTGCGAGCTGA	548	60
	Exon 9	CAATGACCTTGGTTTCTGTCA	TCACCAAACATATTCCCTGATG	474	60
	Exon 10	TTTTGTACGAGTTCCTTTGGTTC	AATAGTTTTCAGGGAGCAGGAA	478	60
	Exon 11	AACAACTAATGAGGAAAACACATCA	TCTCCCGTGTTCTTACTCCC	494	60
	Exon 12	TGACAGACAATATGACTGCTGCT	AGAGCCAAGGTGATAAAGAAGAA	529	60
	Exon 13	TGCAGAGAGTATCAGTGCCAA	CCGAAGTCCCAAAATGAAGA	439	60
	Exon 14	GAGGCTTTCAGATGGCTTTG	GGGTTCAAGCAATTCTCGTG	509	60
	Exon 16	TTTCCCACCATAACCTATCCA	AACTGGGTACCACTTAAAACCATAA	488	60
	Exon 17	TGCATGAAGAGAAGTGTAAACTCA	CAATTGCATCTAAATAATCAGTGTGT	476	60
	Exon 18	ACATTTGGCCACTAGAAAAACA	ACATCTTAATTCCTTAACCACTGGA	488	60
	Exon 19	TCCAAAACTAAAAGCAGAATGTAAA	TTTTAGGATACAGAGGAAGCTCTTTT	500	60
	Exon 20	TCATTTCTTATGTTTCCAAATACGG	TTGCCATTTATCCCAATGGT	400	60
	Exon 21	GCTTTTTGGAAAGAAAGCATTTA	TGGATTAGAAAACAACAGGTACTCAC	531	60
	Exon 22	GCAGTTCCTTGGAGAAACTAAAA	TCTTCCCACTCCAGCATTC	499	60
	Exon 23	CCTCAGCAGGAGAAGCAAAC	CAGGCATTCTTCCCTTTTCA	497	60
	Exon 24	TTCAGCGTACTTCCCATTTG	CCTGCAGACTCTATGCAACAA	485	60

Gene	Genic position	Forward	Reverse	Amplicon length	Annealing temperature
	Exon 25	CAGTCAACAGTTCAGAATGAAAGAA	TGTTGTTGCAAAGATTGTGAAAA	498	60
	Exon 26	TGCCACTGAATTTTTGAGCA	CGTGATGGCTTGAAAATGTG	483	60
	Exon 27	ATGCATTGCAAGGCCATTA	AGGGAAGCCCAGCCTATATTT	471	60
	Exon 28	GAAACAAATTTAACCTCATCTCTGA	CAAGAAATTTTACATAAAATTATTTAG	537	55
	Exon 29	AACAGTTATGCATTGCACTTAATCAT	AGACTGGATTGTTCCTGCCA	461	60
	Exon 30	CCAAGAGGCTATTATTGCCAAA	CAACCATTTTGGTGGGTTTT	484	60
	Exon 31	TGGATGCATTCTTTGTTGCT	TCACAGAGAAAATACAAGCCGA	451	60
	Exon 31	GCAGCTGCAAGAATCCATTT	CCAAATCAAATTCGGTGTCC	479	60
	Exon 31	TCCCTCCTTTTGGATCACTG	CCTGTGCCATAGTGGGTCTT	492	60
	Exon 31	TCGGCTTGTATTTTCTCTGTGA	ATATTGGAGGATGACCGGTTT	465	60
	Exon 31	GTGGCGTCAAATAGTGGCTT	AAGACAAGCAACAGGCGAAT	445	60
	Exon 33	TAAAAATCATGCTGCCCCA	TGGTGACCTTTTACATAGCGG	494	60
	Exon 34	TTGATCCAAAAGTGAGTCTTCCTA	CCCAGGAGATACCCACAGAT	500	60
	Exon 35	GATCTGTCAGACTCAGGCCC	ATTTACTTATGCACAGGTCTTCCA	452	60
	Exon 36	AAATTGCTTCACTAAAATGGACG	TCTTTTCCACTGTGAACAGCC	499	60
	Exon 37	TGCAATGCAGTTTAGAACTACTGA	TTGGTTTTGTTTTGTTTTTCG	538	60
	Exon 38	CAGAATGTCTAAGTGTGTGACAGG	TCTTGAATTCCTGGGCTCAA	479	60
	Exon 39	GAGGTCAGGATTCAGCAGATG	GGTGTGGTGACTCATGCCTA	584	60
	Exon 39	TGCATATCCCATTATTGTTCCA	CCCTAGATGCATGAAGAAGCA	428	60
	3'UTR	AACGTGAAAGGGCCAAAGTT	CACTGTTCCGCAGTTACTGGT	410	60
	3'UTR	AGCGATGAGATGGAAAGTTGA	GGATATGCAGAAGCACCCTT	490	60
	3'UTR	ACCCACCTACCACTGCATTT	TCTTCATTTTGTTGCAATGTGTT	457	60
	3'UTR	TTGCTATGACTGTGTCAAAACTGA	CATTCCATGGGGAAAGAAGA	483	60
	3'UTR	TGAAGTGCAGTCAAAGAGGTG	TTGATGAGGGCAAGGTTTTT	464	60
	3'UTR	CCAAGACCAAAGACCACGAT	CCCATGATGCAGATCCTTTT	406	60
	3'UTR	ACAGGGATAGTGATGGTGGG	CAGATAAGGGCTGTAAGCTGC	424	60
	3'UTR	TGCTGATATCATTCCTAACCTCTTC	GACATTCATTTTAAACACTACCTCAG	521	60
	3'UTR	TTTCCCCCTTCCAATCTACC	TCTCCATGATTTCACTGACAGG	533	60
	3'UTR	TCTTACCATGCAAAAATAACTGC	TCAAGAACAGATTTCAGCCTCA	539	60

Gene	Genic position	Forward	Reverse	Amplicon length	Annealing temperature
	3'UTR	CACTGCTATTAATTTCACTGGTATGC	CATATCACAACGGCCAAGTG	527	60
	3'UTR	TGCAGCTTTAGGGTTTCTGA	GGAGCTATTTGGAATGAACCA	546	60
	3'UTR	TCAAGGTGAGCAAAACCATTT	CCACAGCATGCATAAATTGC	506	60
	3'UTR	CCCATTTTCCAAATCCCTTT	TTTGGAGGTTCTGGTGTTTG	530	60
Raptor	5'UTR	GAGACCGCTCTGAGGAGGTT	ACTTCAGGCCTCACGATGAC	518	60
	5'UTR	TGAGAGTGGCTGGAGGAGAC	ATTTCGGACTCCATCAGTGG	512	60
	Exon 1	TGAGTAAGGGTCTCCGCACT	GCATTTTCCATTTTCAACCAA	487	60
	Exon 2	GCCTGAGAATAGCCCTTCCT	GAGCGCATCGGAAATTACTC	522	60
	Exon 3	TTCACTTGCCCGTGATGTTA	GGACGCTGTCAAGACAACAC	507	FAIL
	Exon 4	CCTGGTTTTATAGATGGAGAAACTC	GCAGTCATTCCATGAACACG	514	60
	Intron 4	CCAGCTATGGGTCTCACTGC	CGCCTGGTGGTTCATTACAT	542	60
	Exon 5	GCAGCATCAGAGGAGATGTG	CAGCATTGGAGCAGTCGTAG	577	60
	Exon 5	GCCTTTTGTAACGGTGCAGT	CTGAGTGCCTCCCTAGCTCA	401	60
	Exon 6	CCAGGTTTCCAGGCTTACAG	AGAACCCAAACCTCCACCTG	540	60
	Exon 7	TCTCACGCCTTGACAGAATG	TGGCCCATGTTTGGTTATCT	459	60
	Exon 8	GAGCTGTGAGCTCATGGAGA	AGAACCTGGCGTGCAGTAAC	475	60
	Exon 9	GGTTGATTTGGGGTGTTTGT	CACCTGGAGGATCCTGAAAC	591	60
	Exon 9	TCAGACAGGACTTGCTGGTG	CGATCTTGCATGTGCTTCAT	490	60
	Intron 9	GCTCCCAGAGATCTCCACAC	TGCTTGTTAGGACAGCCACA	490	60
	Exon 10	TGACAAAATGGTGAGGATGC	AAACCCTGGTTGACTCGATG	505	60
	Exon 11	GGAGTCATCCTCAGCTGCTC	AGCCCAGTGACTCCTGATTG	541	60
	Exon 12	GGAAAGGAGCACCAGCTCTA	GGCCGACAACACACTCACT	513	60
	Exon 13	CAGTCAAGGCTGGCAGTATG	GGGACAGGACAGGTAGGTGA	539	60
	Exon 14	AGCCAAGGAAAGCATGTCTG	CCGGGTTAACTGAGCTCTTG	496	60
	Exon 15	GCTTCCAAGGAGCAGTCTGT	TCTTGGGAAGCCTTGATTTG	505	60
	Exon 16	GGCTCTGACCCTTCATCTAGC	CTCACCAAGGTCCCACTGTC	532	60
	Exon 17	TAGTGCGAGGAGAGCCTTG	CCAGGCAGGAAGGAGACTAC	543	60
	Exon 18	TAGCAGTGGGGAGGATTCTG	GGCAGGTGCCTAAAAAGATG	528	60
	Exon 19	CGGTTGTTGTTTGTCCAGTG	TCATCACCATGTTGGCAGAT	486	60

Gene	Genic position	Forward	Reverse	Amplicon length	Annealing temperature
	Exon 20	CGTCTCCCAGTAACCTGTGGT	CTGCTGGTGCTGAGGTTTC	598	60
	Exon 20	AAATATGTATCTCAGTCATGCCATT	TCTCACCCGTCTATGGAACA	550	60
	Exon 21	ACCCGTTGCCAGAATAACAA	GTAAATCCAGCACCCTCTGC	487	60
	Exon 22	CAGTGGATGCATGTCCAAGA	CAAGATGGGGAGAGCTCAAG	533	60
	Exon 23	ACCGAGCAGCATGGACTC	GTCTGCAAAGGCAGAGAGTG	532	60
	Exon 24	ACGTGGCTCACACTTCACAG	TCTGCGGTTCTACCTCAACC	488	60
	Exon 25	AAAGCCTTCAGCCTTTGAGA	GGGTGCCCTTACCCTTCTG	538	60
	Exon 25	GTTTCATCGCTGGGCTTAAC	CTCTCAGAGGCCTTCCCTTC	542	60
	Exon 26	GCTCCTGGATCCACTCTTGT	CTGGGGCACCTAGCATCTT	530	60
	Exon 27	CCTTAAGCTTTTGGGACTGG	GGCATCCTTCCCTGATTCTC	479	60
	Intron 27	TAGCATCAACCGATGTCGAA	TCAGGGCTTAGAATGGAGGA	526	60
	Exon 28	GAGATCTGGAACCACGGGTA	CAAGCCAAGGATGAAGGTTG	520	60
	Exon 29	GTCCCTAGCGGGAGCTCATA	TGCAGTTCACCTTCATCCAG	470	60
	Exon 30	CCTTTGATGCTTCCCTGGAT	TGTGCTCCTCCATAGGCTGT	531	60
	Exon 31	AGCTTCCATGTGGCTCAGAC	ATCTTTCTGGCCAGTTCCAG	513	60
	Exon 32	CAGTATGGGTGCATGTCTGG	GGTGGGCAGACATCTTTTGT	464	60
	Exon 33	TTCACTGCTGTGGGGACATA	AGGGGCCCTAGAGATCCTG	535	60*
	Exon 34	CCCTGCAGAGGACTGGTG	CACAGAACAGGAGAGGGACAG	532	FAIL
	3'UTR	CCTGCTACTCGCTTTTGTCTG	GCTTCCTGCTCAGTGATGCT	512	60
	3'UTR	GCAAGCAGGGACATTTCCTA	TTGCCTGTATGTGAGGAACG	498	60
	3'UTR	CTGTGGTCTCCATGCCTGTG	CATCCACCCAGAAGATGCTC	523	60
	3'UTR	GAGAGGTGAAGGAGCCAGGT	GCCTTTTGACAGGTGGTGAT	528	60
	3'UTR	GGTCCGGAAGGTGTAGAGAG	ACAGTGCCTGCCTGGAGAG	627	60
AS160	Exon 1	CGGCAGTTGGCACAGTTT	CTTCTCTCCCACTCCCAAGGT	775	60*
	Exon 2	GACTGCATGGAGAAGTTCAGC	CCTGGAAGAGCATGGTCCTGT	411	60
	Exon 3	TTGAGGCAGCTGATATTTGG	CGGGGTCAAGCCCACTTATTT	350	60
	Exon 4	GCCACTGCATGAGTTTGCTA	CCGCCTTCTTCTTTTCCCATA	383	60
	Exon 5	TCAACAAAGGCTCACATTGC	CGCAGAGATTGACAGCCCACT	382	60
	Exon 6 and 7	CCTGGATCTCTGCTGCTAGAC	CAACCTGAGAGTACAAAGGGAAGAA	598	60

Gene	Genic position	Forward	Reverse	Amplicon length	Annealing temperature
	Exon 8	CATCTTGGAAAGGGATGGTT	CGGAAATCAGTGAGCCTGTTGA	548	60
	Exon 9	GAGACCACCTGGAACTACCTTTT	CCCATATCGGTGCTGACTTTG	217	60
	Exon 10	AGCTCTGTGTTTTGGCAGGT	CCACAATCCACCTTGACTCCA	379	60
	Exon 11	TGTAAGATCCAGCCCTGAGC	CAAAAAGCTTTGAAACTACTGGTTGA	503	60
	Exon 12	TCCTGAATCCTGATTTGGACA	CGCATGCATTCAGGAGACAAA	197	60
	Exon 13	GTGGCCTGGGTGACTAGTGT	CGGAGTAGGCCCAACTACAGG	561	60
	Exon 14	AGCGGCTTTGTGACATTTTT	CTCCAAAACTGAGTGCGGATA	429	60
	Exon 15	AAAGCTTGGGACCCTTTCAT	CGCAAAATCTGAGCTCCACAA	422	60
	Exon 16	TTAGGGTGGCATGCAGTACA	CTCTGCATATGCCAACAGGTG	396	60
	Exon 17	GGGGTGAGAGAGGGAGAGAC	CATTGAGAAGAGGGCCAGGAT	479	60
	Exon 18	GTGTTTGATGCACATGCAGAT	CGGGGCATCATATGGCTAGAA	301	60
	Exon 19	GCGTGCTTTTTACAAGAGTGG	CCAAGCAAAGCTACATTTTTAGGC	321	60
	Exon 20	GAGCTATCTGTTTTTATGTTGACAGTG	CTTTGTAGCCCCAGAGCACTT	532	60
	Exon 21	CATCGTTTCCTGTAGGTTTTCA	CCATGCAGGCTCTTCCTCTCT	441	60
*MAPKAP1***	5'UTR	GAGGCTCATGGCAGAGTCAT	CTCCTCGACAGCCTCTTCAGT	500	60
	5'UTR	GAATTCATCAGCATGACCCC	CTTCCACCTGCATGTTCTCTG	433	56.4
	5'UTR	GCAGGAGGGACACTGAAGAG	CGTCACTTTCCCCTTCAGCAC	442	60
	5'UTR	GCGCTGTAAGGAGAAGAGGA	CATGATGGACCATTCTTTGGG	474	51
	5'UTR	GTGCTGAAGGGGAAAGTGAC	CTTGGCAGTGGCTTTGATAAG	472	60
	5'UTR	AACATAATCAATCCTCCGCC	CAGAAGGAGAAGAAATCCGGG	494	60
	Exon 1	TACCAGGTCAGCTGGCTTCT	CCTACCGCTCCTTGCTTTCAG	569	60
	Exon 2	CCCCATTAAGCCTATTCCAA	CAAGCTCCACAGAACAGGGAT	549	52.5
	Exon 3	CAATCCCTGTGACCCAGAGT	CTGGTATTTGCTTTTCAGGGG	469	60
	Exon 4	TGGAAACAATACTGAACCCAAA	CGTGGGGCTTTCTGCATGTAT	475	50
	Exon 5	GGCCAAAAGACCACAAGAAA	CAGAGAGGCACCCAGTGCTAA	570	60
	Exon 6	GCTTCACACAATGTGCCACT	CTCCTCAATTTGCTTGCTGTG	493	62.7
	Exon 7	CTGGGATCCTTTTCTTCGC	CTTGGCACCTCAGTATGCACTTA	550	60
	Exon 7	CTTCTCACATATGCCGCTGA	CATCTTGCAGTGAGCCGAGAT	497	60
	Exon 8	GCACTCAAGTGGGAAAAAGG	CCCAAGGACCTGCCATAGTGT	532	60

Gene	Genic position	Forward	Reverse	Amplicon length	Annealing temperature
	Exon 9	GGCACCATAATACAATCCCG	CACTTGTGTGTGCCATTTGGA	492	60
	Exon 10	AGGGTTATTGGGATTGGGAG	CTGGGAAGACTAGGTGCTGCT	492	60
	Exon 11	GCATCGCTTATCAAAGCCAC	CGACTCCTTCCACACGACAGC	491	60
	Vega isoform OTTHUMT00000054086, exon 4	GGAAACCTTTACCTTTGTCCC	CAACAATTGGCTCTAAGCTTGC	547	52
	Vega isoform OTTHUMT00000054091, exon 7	AACCAGACCAGTGAGCGTTT	CACTTGGTTTCGGGGAGAACT	429	56.4
GβL	Exon 2	AACCTACCTGCGCTTCTTGTC	CAGCAATCATGCTGCGGTC	500	56.4
	Exon 3	GCAGCACCAGGACTCCGTA	CCACCTGAGGTCCCAGATCCT	464	61.7
	Exon 4	GAATGCCTTGGAGGTCACAC	CAAGGGGAATAACAAGCTGGC	500	54.2
	Exon 5 and 6	AAGAGGCAGGTCCTTCAGC	CTCCAGGAATAAGCCCACCT	500	54.2
	Exon 7	TGGTGCCCTTCTCCTGTCTA	CACGTCCTCCAGATCTTGCAC	576	60
	Exon 8	ACTAAGATCCCTGCCCACAC	CCAGGTTGTCCGAGGAAGC	500	54.2
	Exon 9	ATCGTCACTGGTGAGCCC	CGTCCAGGAGTGTGCAGCC	499	58.9

Primers are M13-tailed. Forward primer sequences are preceded by TGTAAAACGACGGCCAGT and reverse sequences are preceded by AGGAAACAGCTATGACCAT. * 7-deaza-dGTP was added to dNTP mix. ** *MAPKAP1* comprises several isoforms. All exons numbers refer to those in OTTHUMT00000054090 except where specified.

Table A16 *WFS1* variants detected by sequencing 1235 type 2 diabetes cases and 1668 controls

Genic position	Genomic position	Nucleotide substitution (minor|major)	Protein consequence	MAF	SNP ID
Upstream	6316152	C|T		0.0722	rs11726771
Upstream	6316308	T|C		0.0001	
Upstream	6316341	A|C		0.0001	
Upstream	6316361	T|G		0.0063	
Upstream	6316417	A|T		0.0003	
Upstream	6316465	A|G		0.0001	
Upstream	6316475	T|C		0.0006	
Upstream	6316476	A|G		0.0001	
Upstream	6316614	A|G		0.0001	
Upstream	6316637	C|G		0.001	
Upstream	6321827	T|C		0.0004	
Upstream	6321914	G|T		0.0001	
Upstream	6321944	A|T		0.1856	rs4320200
Upstream	6321972	T|C		0.1835	rs13107806
Upstream	6321981	T|C		0.0001	
Upstream	6322051	C|G		0.1856	rs13127445
Upstream	6322203	G|T		0.0003	
Upstream	6322207	A|C		0.0001	
Upstream	6322317	G|T		0.1781	rs4273545
Upstream	6322384	G|A		0.0001	
Upstream	6322420	T|G		0.0001	
Upstream	6322429	A|C		0.0001	
Upstream	6322436	C|A		0.0001	
5'UTR	6322518	T|C		0.002	
5'UTR	6322519	G|A		0.0004	
5'UTR	6322527	T|C		0.0001	
5'UTR	6322576	G|A		0.0001	
5'UTR	6322580	T|C		0.0001	
5'UTR	6322593	G|C		0.0004	
5'UTR	6322609	A|G		0.0001	
Intron 1	6322673	T|G		0.0001	
Intron 1	6322727	T|C		0.0331	rs6830765
Intron 1	6324924	G|A		0.003	
Intron 1	6324986	A|G		0.0001	
Intron 1	6324997	C|T		0.0001	
Intron 1	6325007	G|A		0.0001	
Intron 1	*6325055*	*A|T*		*0.0003*	
Intron 1	*6325091*	*A|G*		*0.0003*	
Intron 1	6325206	G|T		0.0003	
Intron 1	6325258	G|T		0.0003	
Intron 1	6325330	A|G		0.0001	
Intron 1	6325386	A|G		0.0003	rs7657752
Intron 1	6325387	C|T		0.0024	
Intron 1	6325978	T|G		0.0003	
Intron 1	6326039	C|T		0.0003	
Intron 1	6326040	A|G		0.0001	

Genic position	Genomic position	Nucleotide substitution (minor|major)	Protein consequence	MAF	SNP ID
Intron 1	6326063	T|G		0.0003	
Intron 1	6326161	C|G		0.0001	
Intron 1	6326178	G|A		0.0003	
Intron 1	6326225	A|G		0.0001	
Intron 1	6326253	A|G		0.0023	
Intron 1	6326269	A|G		0.0001	
Intron 1	6326276	T|C		0.0001	
Intron 1	6326376	A|C		0.0001	
Intron 1	6329948	G|A		0.0087	rs10937714
Intron 1	6330010	A|G		0.0003	
Exon 2	6330104	A|C	P7P	0.0003	
Exon 2	6330124	C|T	Q14R	0.0001	
Exon 2	6330139	A|G	P19L	0.0003	
Exon 2	6330151	A|G	A23V	0.0001	
Exon 2	6330160	T|C	R26Q	0.0001	
Exon 2	6330207	A|G	R42X	0.0001	
Exon 2	6330212	A|T	A43A	0.0001	
Exon 2	6330213	T|G	P44T	0.0003	
Exon 2	6330215	T|G	P44P	0.0003	
Exon 2	6330314	A|G	T77T	0.0001	
Exon 2	6330315	T|C	G78R	0.0001	
Intron 2	6330358	T|C		0.0003	
Intron 2	6330360	G|A		0.0001	
Intron 2	6330363	A|G		0.0001	
Intron 2	6330375	T|C		0.0001	
Intron 2	6330405	C|T		0.3429	rs28420833
Intron 2	6330455	A|G		0.0001	
Intron 2	6330456	T|C		0.0001	
Intron 2	6330540	T|G		0.0001	
Intron 2	6339429	T|C		0.0003	
Intron 2	6339464	T|C		0.0026	
Intron 2	6339486	C|G		0.0003	
Intron 2	6339501	T|C		0.0001	
Intron 2	6339641	A|G		0.0006	
Intron 2	6339648	A|G		0.0001	
Intron 2	6339692	T|G		0.0001	
Intron 3	6339815	T|C		0.0001	
Intron 3	6339847	T|C		0.0001	
Intron 3	6339866	A|G		0.0001	
Intron 3	6339907	T|C		0.0105	
Intron 3	6339923	T|C		0.0001	
Intron 3	6339931	C|G		0.0001	
Intron 3	6339979	A|C		0.0001	
Intron 3	6340039	A|T		0.002	
Intron 3	6341451	C|T		0.0007	
Intron 3	6341495	A|G		0.3016	rs4688989
Intron 3	6341578	A|G		0.0054	rs4688990
Intron 3	6341579	T|C		0.0006	
Exon 4	6341699	T|C	A134T	0.0004	
Exon 4	6341701	T|C	A134A	0.0003	

Genic position	Genomic position	Nucleotide substitution (minor|major)	Protein consequence	MAF	SNP ID
Intron 4	6341787	A|G		0.0001	rs7688426
Intron 4	6341904	G|C		0.2982	rs4689394
Intron 4	6343810	G|A		0.2139	rs9998519
Intron 4	6343816	T|C		0.1497	rs10010131
Exon 5	6343835	A|G		0.0001	
Exon 5	6343846	T|C	R161Q	0.0001	
Exon 5	6343905	A|G	L181L	0.0001	
Exon 5	6343928	T|G	N188K	0.0001	
Exon 5	6343941	G|T	K193Q	0.0039	
Intron 5	6344085	C|G		0.0001	
Intron 5	6344132	C|T		0.0004	
Intron 5	6344138	G|C		0.3889	rs9998835
Intron 5	6344150	T|C		0.0001	
Intron 5	6344159	G|C		0.0001	
Intron 5	6344161	A|T		0.0001	
Intron 5	6344210	T|C		0.0001	
Intron 5	6344250	A|T		0.001	
Intron 5	6344251	A|G		0.2425	rs10012946
Intron 5	6344302	C|T		0.0001	
Intron 5	6344339	C|G		0.0011	
Intron 5	6344347	A|G		0.1577	rs13101355
Intron 5	6344375	T|C		0.3827	rs13147655
Intron 5	6344439	A|G		0.0001	
Intron 5	6344454	A|G		0.0001	
Intron 5	6344456	C|G		0.0001	
Intron 5	6344495	A|G		0.0001	
Exon 6	6344567	A|G	P218P	0.0001	
Exon 6	6344596	T|C	R228H	0.0009	
Exon 6	6344597	G|C	R228R	0.3081	rs7672995
Exon 6	6344608	T|C	R232H	0.0001	
Intron 6	6344641	T|C		0.0017	
Intron 6	6344703	G|A		0.112	rs7655482
Intron 6	6344717	A|G		0.0001	
Intron 6	6344730	T|C		0.0024	
Intron 6	6344739	T|C		0.0328	rs11729672
Intron 6	6344756	G|A		0.0641	rs11725494
Intron 6	6344806	A|G		0.0001	
Intron 6	6344820	G|C		0.0232	rs11725500
Intron 6	6344868	C|T		0.0082	rs4416547
Intron 6	6347348	A|C		0.0115	rs12511742
Intron 6	6347438	T|C		0.0066	
Intron 6	6347519	T|C		0.0001	
Intron 6	6347522	T|C		0.0004	
Intron 6	6347524	C|G		0.0001	
Intron 6	6347582	T|C		0.007	
Intron 6	6347602	T|C		0.0001	
Intron 6	6347603	A|G		0.0001	
Exon 7	6347682	A|G	I242I	0.0001	
Exon 7	6347684	A|G	A243V	0.0003	
Exon 7	6347685	T|C	A243A	0.0001	

Genic position	Genomic position	Nucleotide substitution (minor|major)	Protein consequence	MAF	SNP ID
Exon 7	6347755	T|C	D267N	0.0007	
Exon 7	6347810	T|C	R285H	0.0004	
Exon 7	6347813	G|A	L286P	0.0001	
Intron 7	6347845	T|C		0.0003	
Intron 7	6347856	A|G		0.0003	
Exon 8	6353353	G|A	A310A	0.0001	
Exon 8	6353386	C|G	T321T	0.0001	
Exon 8	6353398	T|C	N325N	0.0003	
Exon 8	6353400	T|C	A326V	0.0004	
Exon 8	6353402	T|C	L327F	0.0001	
Exon 8	6353408	A|T	F329I	0.0003	
Exon 8	6353420	G|A	V333I	0.2613	rs1801212
Exon 8	6353431	G|C	L336L	0.0001	
Exon 8	6353446	T|C	F341F	0.0718	
Exon 8	6353447	A|G	A342T	0.0003	
Exon 8	6353487	G|T	I355S	0.0001	
Exon 8	6353502	A|G	C360Y	0.0004	
Exon 8	6353548	T|C	R375R	0.0001	
Exon 8	6353557	A|C	T378T	0.0004	
Exon 8	6353571	A|G	R383H	0.0001	
Exon 8	6353576	A|G	E385K	0.0004	
Exon 8	6353608	C|T	V395V	0.3809	rs1801206
Exon 8	6353614	T|C	F397F	0.0003	
Exon 8	6353669	G|A	I416V	0.0001	
Exon 8	6353688	T|C	A422V	0.0004	
Exon 8	6353698	T|C	D425D	0.0001	
Exon 8	6353700	A|G	C426Y	0.0007	rs35218685
Exon 8	6353717	G|C	L432V	0.005	rs35031397
Exon 8	6353720	A|G	A433T	0.0001	
Exon 8	6353725	A|C	V434V	0.0001	
Exon 8	6353731	T|C	T436T	0.0083	
Exon 8	6353739	G|T	F439C	0.0003	
Exon 8	6353782	T|C	P453P	0.0001	
Exon 8	6353788	A|G	T455T	0.0001	
Exon 8	6353790	A|G	R456H	0.0417	rs1801208
Exon 8	6353794	T|G	R457S	0.0004	
Exon 8	6353795	C|G	A458P	0.0004	
Exon 8	6353815	T|C	T464T	0.0001	
Exon 8	6353822	T|C	L467L	0.0004	
Exon 8	6353893	T|C	T490T	0.0001	
Exon 8	6353903	A|G	G494S	0.0001	
Exon 8	6353918	T|C	L499F	0.0007	
Exon 8	6353923	C|T	N500N	0.3161	rs1801214
Exon 8	6353929	T|C	S502S	0.0001	
Exon 8	6353975	G|A	M518V	0.0001	
Exon 8	6354003	T|C	T527I	0.0001	
Exon 8	6354020	T|C	P533S	0.0014	
Exon 8	6354031	A|G	V536V	0.0001	
Exon 8	6354055	T|C	S544S	0.0001	
Exon 8	6354056	A|G	V545M	0.0001	

Genic position	Genomic position	Nucleotide substitution (minor|major)	Protein consequence	MAF	SNP ID
Exon 8	6354067	A|G	L548L	0.0001	
Exon 8	6354068	A|C	L549L	0.0001	rs1801211
Exon 8	6354080	A|G	G553S	0.0003	
Exon 8	6354096	A|G	R558H	0.0001	
Exon 8	6354098	A|G	A559T	0.0037	
Exon 8	6354105	G|T	I561S	0.0004	
Exon 8	6354106	T|C	I561I	0.0006	
Exon 8	6354107	A|G	G562S	0.0001	
Exon 8	6354148	T|C	A575A	0.0562	rs2230719
Exon 8	6354149	A|G	G576S	0.0006	rs1805069
Exon 8	6354151	A|C	G576G	0.0001	
Exon 8	6354166	T|C	G581G	0.0001	
Exon 8	6354183	A|G	R587Q	0.0001	
Exon 8	6354199	T|G	L592L	0.0001	
Exon 8	6354220	A|C	V599V	0.0004	
Exon 8	6354223	T|C	T600T	0.0001	
Exon 8	6354228	T|C	A602V	0.0004	rs2230720
Exon 8	6354229	A|G	A602A	0.0003	
Exon 8	6354255	G|A	R611H	0.2863	rs734312
Exon 8	6354262	A|G	W613X	0.0001	
Exon 8	6354306	T|C	T628M	0.0001	
Exon 8	6354308	T|C	R629W	0.0001	
Exon 8	6354346	A|G	T641T	0.0001	
Exon 8	6354435	T|C	A671V	0.0006	
Exon 8	6354436	A|G	A671A	0.0001	
Exon 8	6354442	T|C	C673C	0.0006	
Exon 8	6354443	A|G	G674R	0.0001	
Exon 8	6354449	T|C	R676C	0.0006	
Exon 8	6354450	A|G	R676H	0.0003	
Exon 8	6354451	T|C	R676R	0.0001	
Exon 8	6354475	A|G	A684A	0.0026	
Exon 8	6354476	T|C	R685C	0.0001	
Exon 8	6354477	A|G	R685H	0.0003	
Exon 8	6354522	A|G	W700X	0.0001	
Exon 8	6354530	T|C	R703C	0.0003	
Exon 8	6354545	T|C	R708C	0.0003	
Exon 8	6354547	T|C	R708R	0.0011	
Exon 8	6354560	A|G	D713N	0.0001	
Exon 8	6354571	T|C	A716A	0.0001	
Exon 8	6354572	A|G	E717K	0.0001	
Exon 8	6354580	T|C	A719A	0.0004	
Exon 8	6354581	G|A	I720V	0.0006	rs1805070
Exon 8	6354607	T|C	G728G	0.0006	
Exon 8	6354628	T|C	Y735Y	0.0001	
Exon 8	6354631	T|C	G736G	0.0003	
Exon 8	6354636	A|C	A738D	0.0001	
Exon 8	6354661	A|C	N746K	0.0003	
Exon 8	6354709	A|G	K762K	0.0001	
Exon 8	6354720	C|A	H766P	0.0001	
Exon 8	6354733	T|C	F770F	0.0001	rs34384569

Genic position	Genomic position	Nucleotide substitution (minor|major)	Protein consequence	MAF	SNP ID
Exon 8	6354737	T|C	R772C	0.0004	
Exon 8	6354745	A|G	K774K	0.0669	rs2230721
Exon 8	6354750	T|A	E776V	0.0046	
Exon 8	6354758	A|G	V779M	0.0006	
Exon 8	6354782	A|G	A787T	0.0001	
Exon 8	6354792	G|C	S790W	0.0001	
Exon 8	6354793	A|G	S790S	0.0003	
Exon 8	6354808	C|G	E795D	0.0001	
Exon 8	6354815	A|G	V798I	0.0001	
Exon 8	6354847	T|C	S808S	0.0003	
Exon 8	6354856	G|A	K811K	0.3936	rs1046314
Exon 8	6354875	T|C	R818C	0.0046	rs35932623
Exon 8	6354889	T|C	L822L	0.0001	
Exon 8	6354892	T|C	I823I	0.0006	rs1801215
Exon 8	6354917	T|C	R832C	0.0003	
Exon 8	6354923	A|G	G834S	0.0003	
Exon 8	6354986	C|T	S855P	0.0001	
Exon 8	6354987	T|C	S855L	0.0003	
Exon 8	6354988	A|G	S855S	0.2484	rs1046316
Exon 8	6355004	A|G	V861M	0.0001	
Exon 8	6355012	T|C	I863I	0.0011	
Exon 8	6355019	A|G	D866N	0.0001	rs3821945
Exon 8	6355034	A|G	V871M	0.0129	
Exon 8	6355045	T|C	A874A	0.0001	
Exon 8	6355046	A|G	V875M	0.0003	
Exon 8	6355054	T|C	F877F	0.0001	
Exon 8	6355061	A|G	D880N	0.0001	
Exon 8	6355077	T|C	P885L	0.0001	
Exon 8	6355089	T|C	A889V	0.0001	
3'UTR	6355100	G|T		0.0001	
3'UTR	6355126	T|C		0.0001	
3'UTR	6355143	T|C		0.3131	rs1046317
3'UTR	6355150	G|A		0.0003	
3'UTR	6355165	T|C		0.0001	
3'UTR	6355178	G|C		0.0004	
3'UTR	6355186	A|G		0.0688	rs1802453
3'UTR	6355187	C|T		0.2659	rs1046319
3'UTR	6355218	A|G		0.0001	
3'UTR	6355227	T|C		0.0199	
3'UTR	6355245	G|A		0.303	rs1046320
3'UTR	6355321	G|A		0.0003	
3'UTR	6355337	C|T		0.0006	
3'UTR	6355349	A|G		0.051	rs1046322
3'UTR	6355370	G|A		0.0526	rs1046325

Genomic coordinates are NCBI build 36 (B36). Intron 1 SNPs in italics are conserved and within highly conserved regions.

Table A17 Rare (MAF<0.01) nonsense and missense changes detected in *WFS1* by sequencing 1235 type 2 diabetes cases and 1668 controls

Chr:base	Variant	rs ID	Biochemical/genetic evidence	Pdel*	SIFT	PolyPhen	MAF in cases	MAF in controls	Conservation
4:6330124	Q14R		Unknown	0.16483	affects protein	benign	0.000405	0	Low
4:6330151	A23V		Novel	0.43978	affects protein	benign	0	0.0003	Low
4:6330160	R26Q		Novel	0.32096	affects protein	benign	0	0.0003	Low
4:6330207	R42X		Novel				0	0.0003	STOP
4:6330213	P44T		Novel	0.40278	affects protein	benign	0	0.0003	Low
4:6330315	G78R		Novel	0.46078	affects protein	benign	0.000405	0	Low
4:6341699	A134T		Novel	0.30575	affects protein	possibly damaging	0	0.0006	Complete
4:6343846	R161Q		Associated with hearing loss	0.12633	tolerated	possibly damaging	0	0	Low
4:6343928	N188K		Novel	0.43033	affects protein	possibly damaging	0	0	Low
4:6343941	K193Q		Associated with earing loss (HL)	0.30458	tolerated	possibly damaging	0.003239	0.003297	Complete
4:6344596	R228H		Novel	0.50948	tolerated	possibly damaging	0.000405	0.000899	Low
4:6344608	R232H		Novel	0.55092	tolerated	possibly damaging	0.000405	0	Low
4:6347684	A243V		Novel	0.3299	tolerated	benign	0	0.0003	Low
4:6347755	D267N		Novel	0.3789	tolerated	benign	0	0.001499	Low
4:6347810	R285H		Novel	0.50948	tolerated	benign	0.000405	0.0006	Low
4:6347813	L286P		Novel	0.31806	tolerated	benign	0	0	Low
4:6353400	A326V		Associated with psychiatric disorders	0.40939	affects protein	benign	0.000405	0	Vertebrate
4:6353402	L327F		Novel	0.55726	affects protein	possibly damaging	0.000405	0	Vertebrate

Chr:base	Variant	rs ID	Biochemical/genetic evidence	Pdel*	SIFT	PolyPhen	MAF in cases	MAF in controls	Conservation
4:6353408	F329I		Novel	0.2868	affects protein	probably damaging	0.00081	0	Vertebrate
4:6353447	A342T		Novel	0.67015	tolerated	benign	0.000405	0.0003	Vertebrate
4:6353487	I355S		Novel	0.41045	tolerated	probably damaging	0	0.0003	Low
4:6353502	C360Y		Novel	0.77597	affects protein	probably damaging	0.00081	0	Vertebrate
4:6353571	R383H		Novel	0.59158	tolerated	benign	0	0	Low
4:6353576	E385K		Polymorphism	0.36132	affects protein	possibly damaging	0.000405	0.0003	Vertebrate
4:6353669	I416V		Novel	0.12475	tolerated	benign	0.000405	0	Vertebrate
4:6353688	A422V		Novel	0.54956	tolerated	benign	0.000405	0.0006	Low
4:6353700	C426Y	rs35218685	Associated with psychiatric disorders	0.63713	tolerated	probably damaging	0.00081	0.0003	Low
4:6353717	L432V	rs35031397	Associated with psychiatric disorders and HL	0.42538	affects protein	benign	0.005263	0.005396	Complete
4:6353720	A433T		Novel	0.42841	tolerated	benign	0	0.0003	Low
4:6353739	F439C		Novel	0.59591	affects protein	probably damaging	0.000405	0.0003	Vertebrate
4:6353794	R457S		Unknown	0.56262	tolerated	possibly damaging	0.000405	0.0003	Low
4:6353795	A458P		Novel	0.53226	tolerated	possibly damaging	0.000405	0.0006	Vertebrate
4:6353903	G494S		Novel	0.40523	affects protein	possibly damaging	0	0.0003	Complete
4:6353918	L499F		Suicide	0.6993	tolerated	benign	0.000405	0.001199	Low
4:6353975	M518V		Novel	0.31466	affects protein	probably damaging	0	0	Complete
4:6354003	T527I		Novel	0.37986	tolerated	possibly damaging	0	0.0003	Low
4:6354020	P533S		Suicide	0.12494	affects protein	probably damaging	0.00081	0.002098	Complete
4:6354056	V545M		Novel	0.72875	tolerated	benign	0	0.0003	Vertebrate

Chr:base	Variant	rs ID	Biochemical/genetic evidence	Pdel*	SIFT	PolyPhen	MAF in cases	MAF in controls	Conservation
4:6354080	G553S		Novel	0.40523	tolerated	benign	0.000405	0.0003	Low
4:6354096	R558H		WS	0.61604	affects protein	possibly damaging	0.000405	0	Complete
4:6354098	A559T		WS and psychiatric disorders	0.34899	tolerated	benign	0.004858	0.002998	Low
4:6354105	I561S		Novel	0.44338	affects protein	possibly damaging	0	0.0006	Low
4:6354107	G562S		Unknown	0.28935	tolerated	benign	0.000405	0	Low
4:6354149	G576S	rs1805069	Associated with psychiatric disorders	0.38948	tolerated	benign	0.00081	0.0003	Vertebrate
4:6354183	R587Q		Novel	0.42654	tolerated	benign	0.000405	0	Low
4:6354228	A602V	rs2230720	Polymorphism	0.10473	tolerated	benign	0.00081	0.0003	Low
4:6354262	W613X		WS				0	0	STOP
4:6354306	T628M		Novel	0.74256	affects protein	possibly damaging	0.000405	0	Vertebrate
4:6354308	R629W		WS & reduces half-life of wolframin	0.87775	affects protein	probably damaging	0.000405	0	Low
4:6354435	A671V		WS and psychiatric disorders	0.21811	tolerated	benign	0.001215	0	Low
4:6354443	G674R		Polymorphism	0.60981	affects protein	probably damaging	0.000405	0	Vertebrate
4:6354449	R676C		Novel	0.75663	affects protein	probably damaging	0.000405	0.0003	Low
4:6354450	R676H		Novel	0.54195	tolerated	possibly damaging	0	0.0003	Low
4:6354476	R685C		Polymorphism	0.84434	affects protein	probably damaging	0.000405	0	Low
4:6354477	R685H		Novel	0.68545	affects protein	benign	0.000405	0	Low
4:6354522	W700X		WS & reduces half-life of wolframin				0	0	
4:6354530	R703C		Novel	0.10889	affects protein	probably damaging	0	0.0006	Low
4:6354545	R708C		WS	0.77798	affects protein	probably damaging	0	0.0006	Vertebrate
4:6354560	D713N		Novel	0.41237	tolerated	benign	0.000405	0	Low

Chr:base	Variant	rs ID	Biochemical/genetic evidence	Pdel*	SIFT	PolyPhen	MAF in cases	MAF in controls	Conservation
4:6354572	E717K		WS and psychiatric disorders	0.32653	tolerated	benign	0	0.0003	Low
4:6354581	I720V	rs1805070	Association with T1D	0.12475	tolerated	benign	0.001215	0.0003	Complete
4:6354636	A738D		Novel	0.49144	tolerated	benign	0	0	Low
4:6354661	N746K		Novel	0.24923	tolerated	possibly damaging	0.000405	0	Vertebrate
4:6354720	H766P		Novel	0.53195	tolerated	probably damaging	0.000405	0	Vertebrate
4:6354737	R772C		Associated with psychiatric disorders	0.91098	affects protein	probably damaging	0	0.000899	Low
4:6354750	E776V		WS	0.49302	affects protein	probably damaging	0.004049	0.006295	Complete
4:6354758	V779M		Associated with HL	0.65969	tolerated	benign	0.000405	0.000899	Low
4:6354782	A787T		Unknown	0.17367	tolerated	benign	0	0.0003	Low
4:6354792	S790W		Novel	0.71718	affects protein	possibly damaging	0	0.0003	Low
4:6354808	E795D		Novel	0.31774	tolerated	benign	0	0.0003	Low
4:6354815	V798I		Novel	0.13624	tolerated	benign	0.000405	0	Low
4:6354875	R818C	rs35932623	WS and psychiatric disorders	0.68043	affects protein	possibly damaging	0.004858	0.005396	Low
4:6354917	R832C		Novel	0.75614	affects protein	probably damaging	0.000405	0.0003	Low
4:6354923	G834S		Novel	0.44336	tolerated	possibly damaging	0.000405	0.0003	Vertebrate
4:6354986	S855P		Novel	0.69611	tolerated	benign	0	0.0003	Low
4:6354987	S855L		Novel	0.47396	tolerated	benign	0.000405	0.0003	Low
4:6355004	V861M		Novel	0.17054	affects protein	benign	0.000405	0	Vertebrate
4:6355019	D866N	rs3821945	Associated with psychiatric disorders	0.39863	tolerated	benign	0.000405	0	Low
4:6355046	V875M		Novel	0.3126	tolerated	benign	0.000405	0.0003	Low
4:6355061	D880N		Novel	0.49211	affects protein	possibly damaging	0	0.0003	Vertebrate

Chr:base	Variant	rs ID	Biochemical/genetic evidence	Pdel*	SIFT	PolyPhen	MAF in cases	MAF in controls	Conservation
4:6355077	P885L		WS & reduces half-life of wolframin	0.54691	affects protein	probably damaging	0	0.0003	Complete
4:6355089	A889V		Novel		affects protein	benign	0	0.0003	Vertebrate

Genomic coordinates are NCBI build 36 (B36). * = Probability that the variation is deleterious according to PANTHER.

Table A18 *WFS1* PCR primer sequences

Gene	Genic position	Forward	Reverse	Amplicon length	Annealing temperature
WFS1	Upstream	ATAGACCCTCGCCCTCATCT	CGCCAATCACTCCACATCTCC	604	
WFS1	Exon 1*	GGAGTGGTGGATGAAAGGTG	CACAGTGCCTCAGGGCTCTT		50
WFS1	Intron 1	GGACCTACCCTCAGTCCACA	CGGCAGGATTTAAAGCAGCAG	576	61.7
WFS1	Intron 1	GTCTGAGTGGATGCATGGTG	CCTGGTCATGGTTGGTCACAG	557	58.9-62.7
WFS1	Exon 2	TGCAAATGGAAGCTGTGAAG	CGTGAGTGTCCTCCCATGGTT	718	61.7
WFS1	Exon 3	AGTGTATGTTGAGGGGTGGC	CGGTCATGGGCTGTCAAACTT	790	58.9-64.7
WFS1	Exon 4	ACCCAGGATCGATGTCTCAG	CTACCAGTACCAGTCGGAGCC	703	54.2
WFS1	Exon 5	TCCTACGTTCTGGGGATCAT	CTCCCTGGTAACCAAGTCCTG	743	50
WFS1	Exon 6	CTGGCACATCATGGTTTCTG	CGCCCTAGTGGTGAGGTGTGT	728	50-54.2
WFS1	Exon 7	CCCGAGGACACATCCTTATG	CGCTGAAGAGGAGGGTGTCAG	703	54.2-56.4
WFS1	Exon 8	CTCGTTCCCACGTACCATCT	CCTTCAGGTAGGGCCAATTCA	614	61.7
WFS1	Exon 8	TCATCACCGGCTTCTTTACC	CACACCAGGATGAGCTTGACC	615	52.5
WFS1	Exon 8	GTCTGTAGTGTGCCCCTGCT	CGATGGTGCTGAACTCGATGA	678	50
WFS1	Exon 8	TTCGACCGCTACAAGTTTGA	CGGGAGGAGAGGGAATCTCAT	698	54.2

Primers are M13-tailed. Forward primer sequences are preceded by TGTAAAACGACGGCCAGT and reverse sequences are preceded by AGGAAACAGCTATGACCAT. * 7-deaza-dGTP was added to dNTP mix.

Table A19 *WFS1* Sequenom primer sequences

Plex	SNP_ID	2nd PCR primer	1st PCR primer	MassEXTEND primer
iPLEX1	WFS1_S855P	ACGTTGGATGTTCGAGCTCAAGGCCATCAG	ACGTTGGATGCCAGTCGTGCTCGATCTTCA	CTGCATGGCCCAGCTC
iPLEX1	rs4688989	ACGTTGGATGTAGCCTAGTGGACATGCCTG	ACGTTGGATGTTCACCCACTTCCCTTTGTC	GGTGTGACCCCATTTC
iPLEX1	VAR_010602	ACGTTGGATGAAGAGGAAGAGGAAGTAGCC	ACGTTGGATGTGTGCTTCATGTGGTGTGAG	AAGTAGCCGATGGAGG
iPLEX1	rs13101355	ACGTTGGATGACTGTGTCCATCACCAAGTG	ACGTTGGATGCACCTCTGTACCTCCAGTAG	GGGAGCACGCTACGTGG
iPLEX1	rs2230719	ACGTTGGATGACTTCCTCTTCCTCTTTGCC	ACGTTGGATGTGAGCTCCAGAGACGTGAAC	CCTCCCCATCCTGGTGGC
iPLEX1	WFS1_K800E	ACGTTGGATGCTCTTGAACTCGCTGCTGG	ACGTTGGATGTGGGCATGCCATTCAGCAG	CCCGCAGCACGATGTCCT
iPLEX1	rs7672995	ACGTTGGATGTGCCCAAGTCCCTGCAGAAG	ACGTTGGATGACTCACACTCGCTGCTGAC	CCTGCAGAAGCAGAGGCG
iPLEX1	WFS1_A684A	ACGTTGGATGACTGACCTGGCAGCAGTATG	ACGTTGGATGTCCAGGTGGCTGCAGAGGAT	GGAAGGAGACCAACATGGC
iPLEX1	rs12642481	ACGTTGGATGAAGGCCAAGAGCTCTTTCTG	ACGTTGGATGATAGTCAAGGCCAAAGGCTG	GAGCTCTTTCTGACCTTAGC
iPLEX1	WFS1_W648R	ACGTTGGATGTTGTAGACCTTCATGCCCTC	ACGTTGGATGTCAAGCTCATCCTGGTGTGG	TGAGCGGTACACATAGAACC
iPLEX1	rs12511742	ACGTTGGATGTCTCTGCCTAGCTCCTTATG	ACGTTGGATGTGGAGGTGCATGTTGTAAGG	CACACCTTTCACCCAGTTCAC
iPLEX1	rs1046322	ACGTTGGATGAATTGCATGCCATCTCCACC	ACGTTGGATGAAAGTGCTTTTCCAGACACC	ACCCTGAGCCTGACCTTTCTGA
iPLEX1	rs5018648	ACGTTGGATGTGGAAGGCTTTCGAGCGAGA	ACGTTGGATGCTTGAGTCAGATGTCCATGC	AGGCTTTCGAGCGAGACTCATA
iPLEX1	VAR_014995	ACGTTGGATGCATGTACTGGAAGCTCAACC	ACGTTGGATGTTGACCTGGCCGACATTCTC	GGAAGCTCAACCCCAAGAAGAAG
iPLEX1	WFS1_19903	ACGTTGGATGAACCTGTACCAGTACCAGTC	ACGTTGGATGTCAAAACACGGTGAGGAAGG	TACCAGTACCAGTCGGAGCCCGTGT
iPLEX1	rs13107806	ACGTTGGATGGCACTGATAGCAGTACTTGG	ACGTTGGATGTCCCCTAATAACAGCAGCAG	GCAGTACTTGGCTGCAGACACTCGTT
iPLEX2	rs1802453	ACGTTGGATGCAGTGCATGTTGCCATGAGG	ACGTTGGATGTGCAGCCACAGTCTGCACAC	GGCATGCACCAGTGCC
iPLEX2	rs4416547	ACGTTGGATGTTCTGTCCTGGGTCTGCCTT	ACGTTGGATGTGTTCTGGACGCTGGGAGTA	CTGCCTTCCCTCCAGTG
iPLEX2	WFS1_33750	ACGTTGGATGTGCAGACTGTGGCTGCAGA	ACGTTGGATGTTCAGAGTGGACAGAGCTAC	TGGCTGCAGAGACCTTG
iPLEX2	rs10937714	ACGTTGGATGAGCTGCCAGACACTCCATAC	ACGTTGGATGTTTCCTCCCTGGAAGCGGTG	TCCATACAGGATCTCGCT
iPLEX2	rs7655482	ACGTTGGATGTCTCACCCATGCCTCCCAG	ACGTTGGATGACGGGTGAGATAGGGCAGG	CCTCTCCTTCCTGTGCGAC
iPLEX2	rs35031397	ACGTTGGATGAGCTGGTCACGGTAAAGAAG	ACGTTGGATGTCATCTTCTCCTTCCCCATC	AAGCCGGTGATGACAGCCA
iPLEX2	rs1801208	ACGTTGGATGAGCCTGAGCACCCATGCAGA	ACGTTGGATGCAATTCAAGGGCATGGAGGG	CCATGCAGAGCCCTACACGC
iPLEX2	rs1046316	ACGTTGGATGTGCTCGATCTTCACGTGCC	ACGTTGGATGTCTTCGAGCTCAAGGCCATC	CTTCACGTGCCGCCTGGTGGG
iPLEX3	VAR_014996	ACGTTGGATGGTGAAGATCGAGCACGACTG	ACGTTGGATGAAGTCGAAGGCGAACTTCAC	CGACTGGCGCAGCACC
iPLEX3	rs35932623	ACGTTGGATGGGGCCAGCAGCGAGTTCAA	ACGTTGGATGTCCAGGATGGTGCTGAACTC	GCGTGCTGCTCAGCCTG
iPLEX3	WFS1_33768	ACGTTGGATGGTATTCAGAGTGGACAGAGC	ACGTTGGATGAGAGACCTTGCGACCATGTG	TCCCTTTGTCGGGGTCCA