

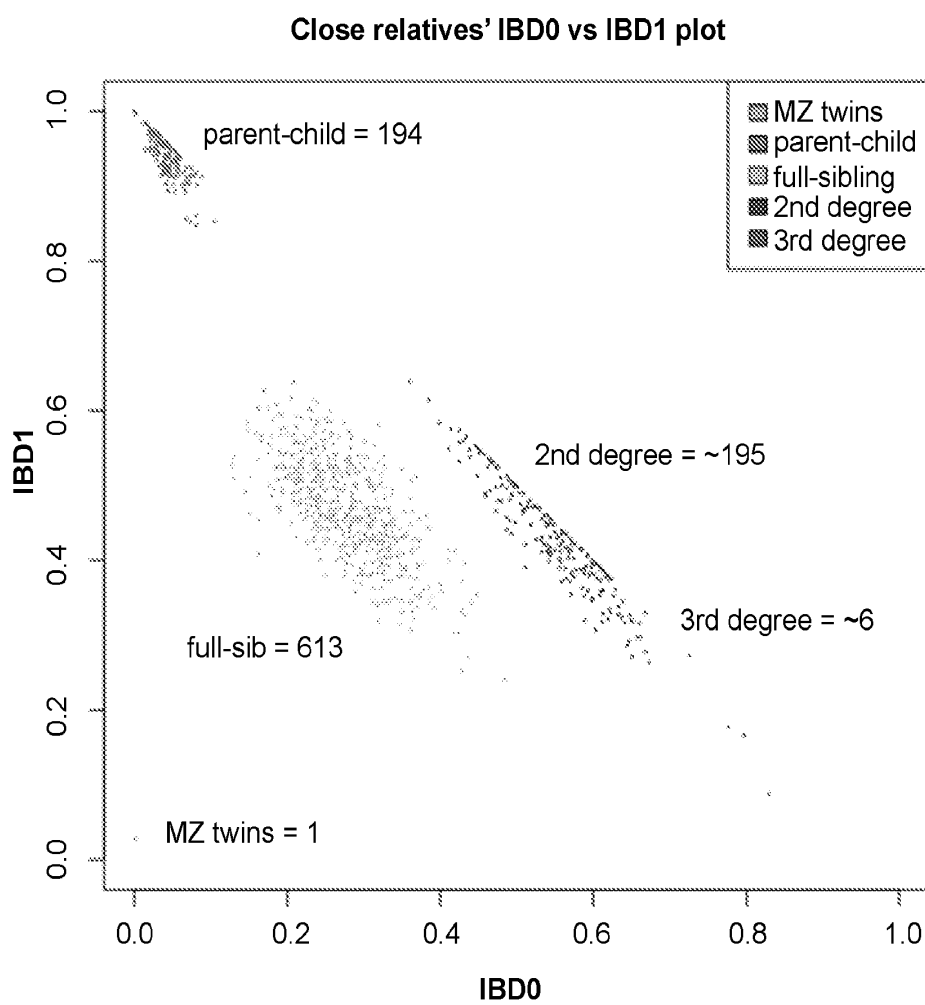


Figure 1

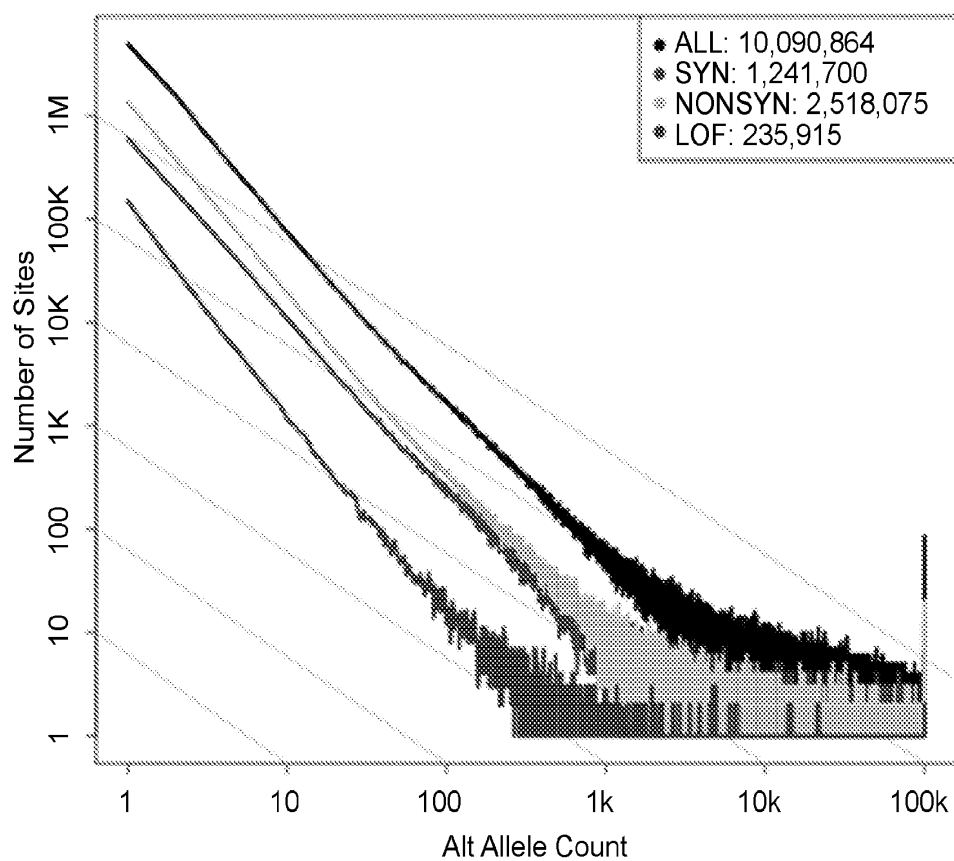


Figure 2

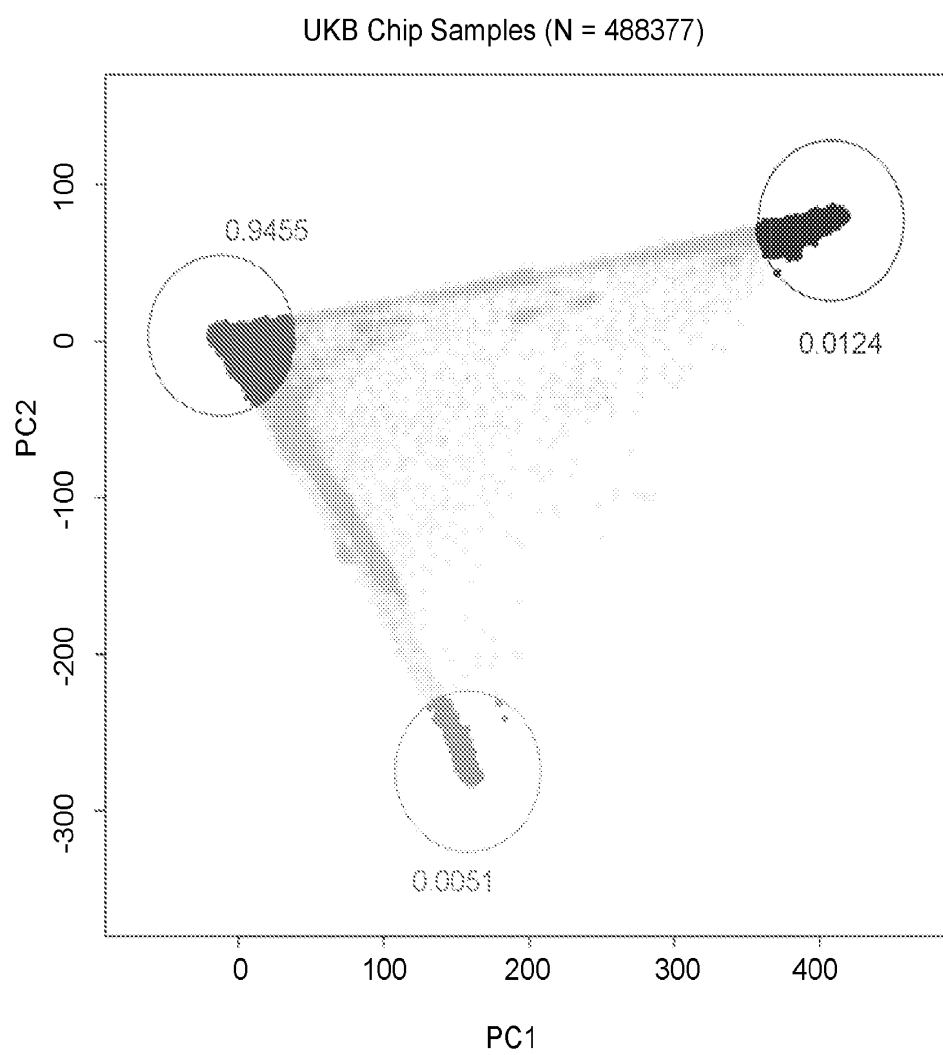


Figure 3

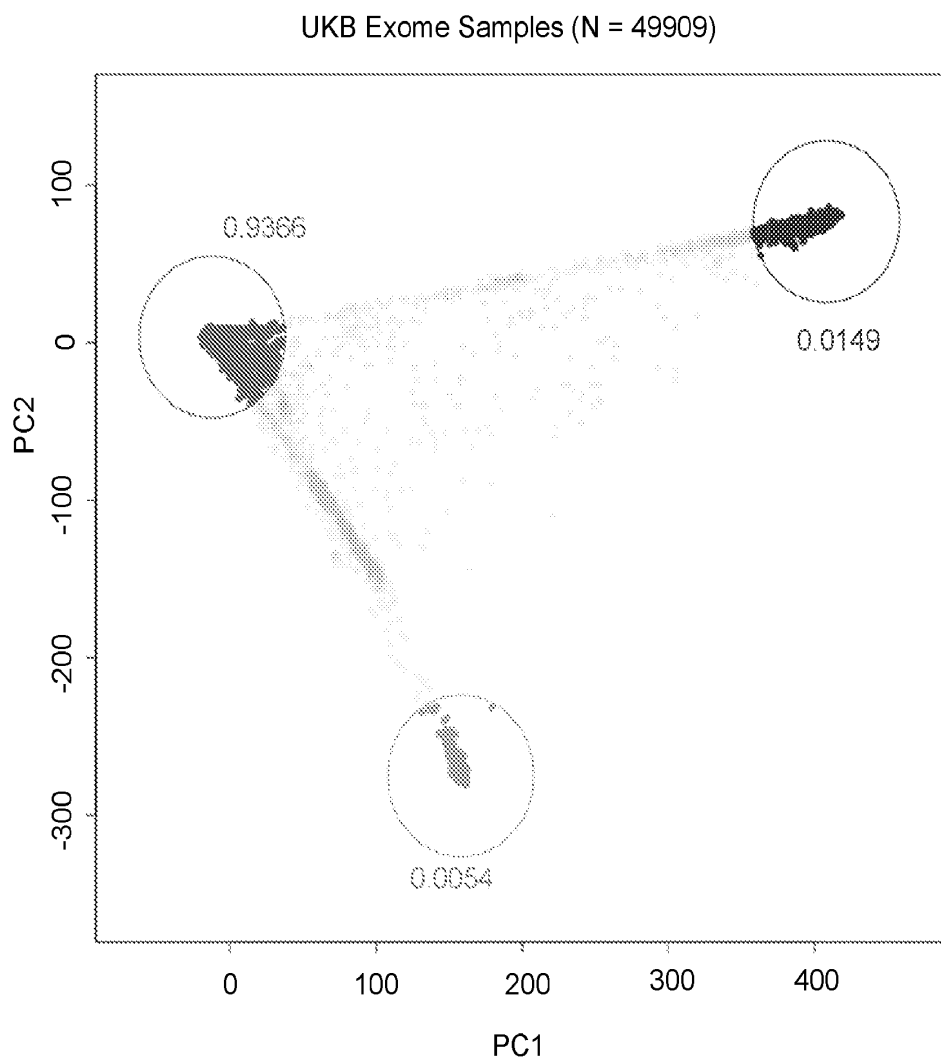


Figure 3 (cont.)

Single-point and aggregate results for the 65 pLOF variants in PIEZO1

Single-point analysis										Drop one - RVT	
SNPID (rsID)	Ref	Alt	AAF	Beta	SE	P-Val	MAC	Functional Effect		P-Val LOO Burden	Delta P-Val Burden
16:88727163:G:A	G	A	6.9E-05	2.81	0.88	0.001	6	stop gained		1.06E-06	33.91
16:88721586:G:C	G	C	1.1E-05	14.63	119.5	0.903	1	stop gained		2.63E-07	8.45
16:88716874:G:A	G	A	1.1E-05	14.40	119.5	0.904	1	stop gained		2.59E-07	8.31
16:88738735:D:1	TC	T	1.1E-05	13.95	119.5	0.907	1	frameshift		2.45E-07	7.86
16:88721268:D:1	CT	C	1.1E-05	13.90	119.5	0.907	1	frameshift		2.43E-07	7.80
16:88726546:C:T	C	T	1.1E-05	13.67	119.5	0.909	1	splice donor		2.34E-07	7.49
16:88723253:G:A (rs368895635)	G	A	2.3E-05	3.77	1.42	0.008	2	stop gained		2.31E-07	7.42
16:88719588:G:A	G	A	1.1E-05	13.57	119.5	0.910	1	stop gained		2.29E-07	7.35
16:88720229:C:A	C	A	1.1E-05	13.55	119.5	0.910	1	stop gained		2.28E-07	7.32
16:88727072:D:1	TC	T	2.3E-05	3.59	1.42	0.011	2	frameshift		2.22E-07	7.12
16:88716359:A:G (rs776709730)	A	G	2.3E-05	3.15	1.42	0.026	2	splice donor		1.94E-07	6.23
16:88736324:G:A	G	A	4.6E-05	2.48	1.18	0.035	4	stop gained		1.67E-07	5.35

Figure 4

LD Assessment for PIEZO1

Variant	LD(R ²)	1	2	3	4	5	6	7	8	9	10	11	12
1	16:88716359:A:G	1	1.1E-9	1.1E-9	1.1E-9	1.1E-9	2.1E-9	1.1E-9	2.1E-9	6.3E-9	4.2E-9	1.1E-9	2.1E-4
2	16:88716874:G:A		1	5.2E-10	5.2E-10	5.2E-10	1.1E-9	5.2E-10	1.1E-9	3.1E-9	2.1E-9	5.2E-10	2.1E-5
3	16:88719588:G:A			1	5.2E-10	5.2E-10	1.1E-9	5.2E-10	1.1E-9	3.1E-9	2.1E-9	5.2E-10	8.1E-6
4	16:88721268:D:1				1	5.2E-10	1.1E-9	5.2E-10	1.1E-9	3.1E-9	2.1E-9	5.2E-10	8.1E-6
5	16:88721586:G:C					1	1.1E-9	5.2E-10	1.1E-9	3.1E-9	2.1E-9	5.2E-10	8.1E-6
6	16:88723253:G:A						1	1.1E-9	2.1E-9	6.3E-9	4.2E-9	1.1E-9	1.6E-5
7	16:88726546:C:T							1	1.1E-9	3.1E-9	2.1E-9	5.2E-10	2.1E-5
8	16:88727072:D:1								1	6.3E-9	4.2E-9	1.1E-9	1.6E-5
9	16:88727163:G:A									1	1.3E-8	3.1E-9	2.8E-5
10	16:88736324:G:A										1	2.1E-9	8.6E-5
11	16:88738735:D:1											1	8.1E-6
12	16:88835545:G:A												1

Figure 5