Supplementary Table 1: Sequences of obscurin fragments obtained from non-transformed (MCF10A) and cancer (MCF7) breast epithelial cell lines were aligned to the mRNA entries coding for human obscurin-A (accession number NM_052843.2) and obscurin-B (accession number NM_001098623). In addition to the seven previously identified and twelve novel mutations detected in obscurin transcripts derived from MCF7 cells, we also found a novel polymorphism (T→G) in position 23,091 of the obscurin-B transcript that results in a missense mutation (Phe→Val) within the SKI kinase domain in both MCF10A and MCF7 cells. NR = Not Reported in dbSNP database.

Nucleotide	Base Change	Domain	Mutation	Codon position	Amino acid	dbSNP accession number
13969	C→G	Ig47	Missense	2	Ser→Cys	rs1188729
14000	T→C	Ig47	Synonymous	3	Gly→Gly	rs1188728
14929	A→G	Ig48	Missense	2	Asp→Gly	rs373610
15371	T→C	Linker Ig48-Ig49	Synonymous	3	Pro→Pro	NR
15728	T→C	Linker Ig49-Ig50	Synonymous	3	Asp→Asp	NR
15849	C→G	Ig50	Missense	1	Leu→Val	rs369909
16721	C→A	Linker Ig51-SH3	Synonymous	3	Pro→Pro	rs3795808
17240	T→C	RhoGEF	Synonymous	3	Ala→Ala	rs505629
17513	A→G	RhoGEF	Synonymous	3	Pro→Pro	NR
17671	A→G	Linker RhoGEF-PH	Missense	2	Asn→Ser	NR
17715	C→G	PH	Missense	1	Gln→Glu	rs1188710
17835	A→G	PH	Missense	1	Ile→Val	NR
17960	G→A	PH	Nonsense	3	Trp→STOP	NR
19879	A→G	SKII	Missense	2	Gln→Arg	NR
20008	*Insertion of 38 nts	SKII	Insertion/ Nonsense			NR
20245	A→G	SKII	Missense	2	Asn→Ser	NR
20403	G→A	Linker SKII-Ig55	Missense	1	Gly→Ser	NR
22377	A→G	Linker SKII-Ig55	Missense	1	Lys→Glu	NR
22452	T→C	Ig55	Missense	1	Ser→Pro	NR

***:** The following nucleotides are inserted after position 20,008: 5' CAGGGCCTCATCTGAGGGCTGG ACCCTCCCTCTGTCTT 3', leading to a frameshift mutation and premature termination of Obscurin-B.