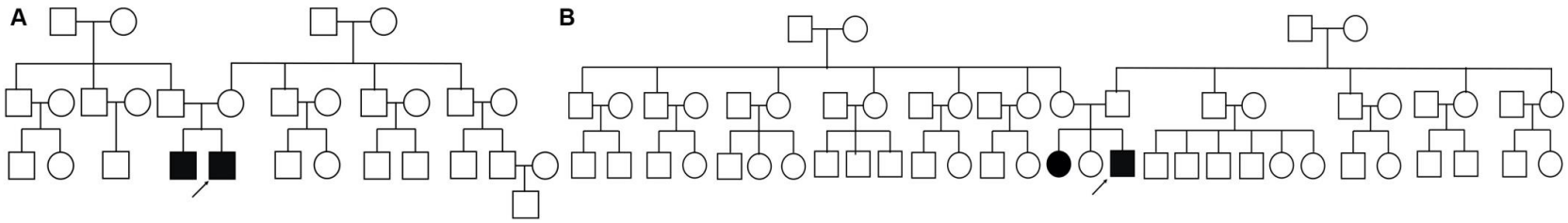

Supplementary Figure 1. Pedigree of families 1 and 2

(A) Pedigree of family 1 with black fill denoting siblings with azoospermia. The two brothers underwent WES. (B) Pedigree of family 2 with black fill denoting azoospermia in male and infertile female patient. The male patient underwent WES. The arrowheads indicate the probands in families 1 and 2.

Supplementary table 1. Primer sequences used for Sanger sequencing.

SHOC1 locus	Forward primers (5'-3')	Reverse primers (5'-3')
SHOC1-528 in family 1	GACCAAGAACCAGTAAACAGAATAA	GAGACAGGACTGACAGCTAAA
SHOC1-78 in family 1	CTTCTCTGTGTCTGTTGGAATTTG	CTTAGAGTTATGCAGGTGGATCTT
SHOC1-400 in family 2	CCAGTGTGGTTCTGTTTATTGTG	ATGGGACCTAGAGAAATCCAAAG
SHOC1-489 in Individual case	CACTCGTTTAGGTGTGGAAGT	AGTCTTTGAGGTGCAAGTCTTAT

Supplementary table 2. Clinical Characteristics of the Chinese Study Subjects at the Clinical Assessment.

Parameters	Proband in family 1	Brother in family 1	Proband in family 2	Individual case
Age (years)	29	32	30	25
Testicular Volume (Left)	10	10	15	10
Testicular Volume (Right)	10	10	20	10
FSH (IU/L)	12.44	11.26	3.9	4.83
LH (IU/L)	5.76	3.03	5.33	4.47
Testosterone (nmol/L)	13.67	13.74	8.5	27.24
Semen Volume (mL)	3	2	4	2.5
Total Sperm Count	0	0	0	0
Karyotype	N	N	N	N
Y Chromosome Microdeletions	N	N	N	N

Abbreviations: AZF, azoospermia factor; FSH, follicle-stimulating hormone; LH, luteinizing hormone; N indicates normal phenotype

Supplementary table 3: Biallelic SHOC1 LoF mutations identified in the subjects with meiotic defects

					gnomAD Dataset			Genotype	
Position	ID	Gene	cDNA mutation	Protein alteration	MAF	Carriers	Case(s)	Father	Mother
Variant identified in two NOA-affected patients in family 1 using WES									
chr9:114489973	rs1004968910	*SHOC1*	c.C1582T	p.R528X	1.8×10^{-5}	Het:5;Hom:0	G/A	G/A	G/G
chr9:114538089	rs777595871	*SHOC1*	c.231_232del	p.L78Sfs*9	2.8×10^{-5}	Het:7;Hom:0	AGT/A	AGT/AGT	AGT/A
Variant identified in NOA-affected proband in family 2 using WES									
chr9:114500590	rs1432616103	*SHOC1*	c.1194delA	p.L400Cfs*7	8.0×10^{-6}	Het:2;Hom:0	AT/A	AT/A	AT/A
Variant identified in NOA-affected Chinses sporadic case subject using WES									
chr9:114490090	NA	*SHOC1*	c.1464delT	p.D489Tfs*13	NA	NA	CA/C	NA	NA