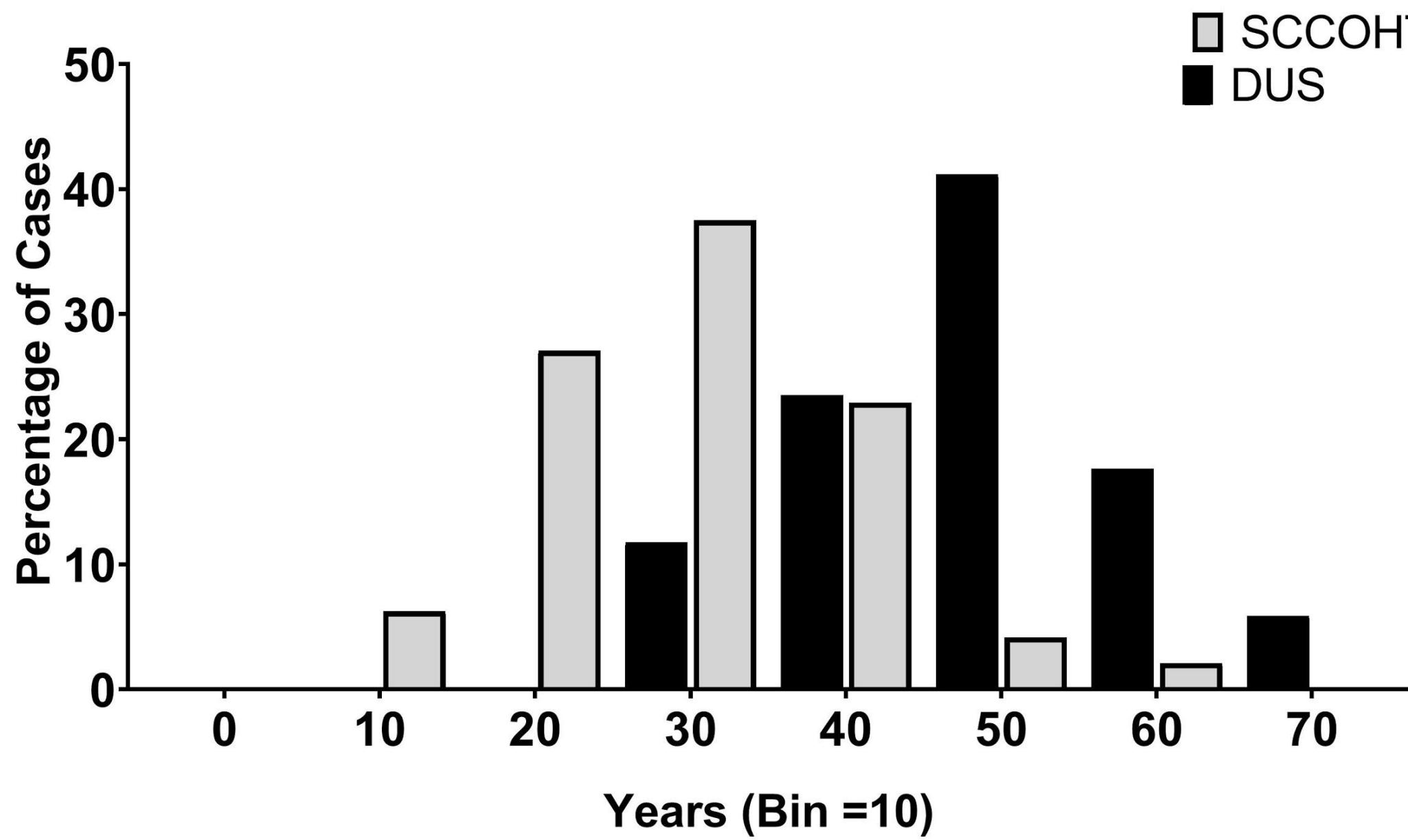
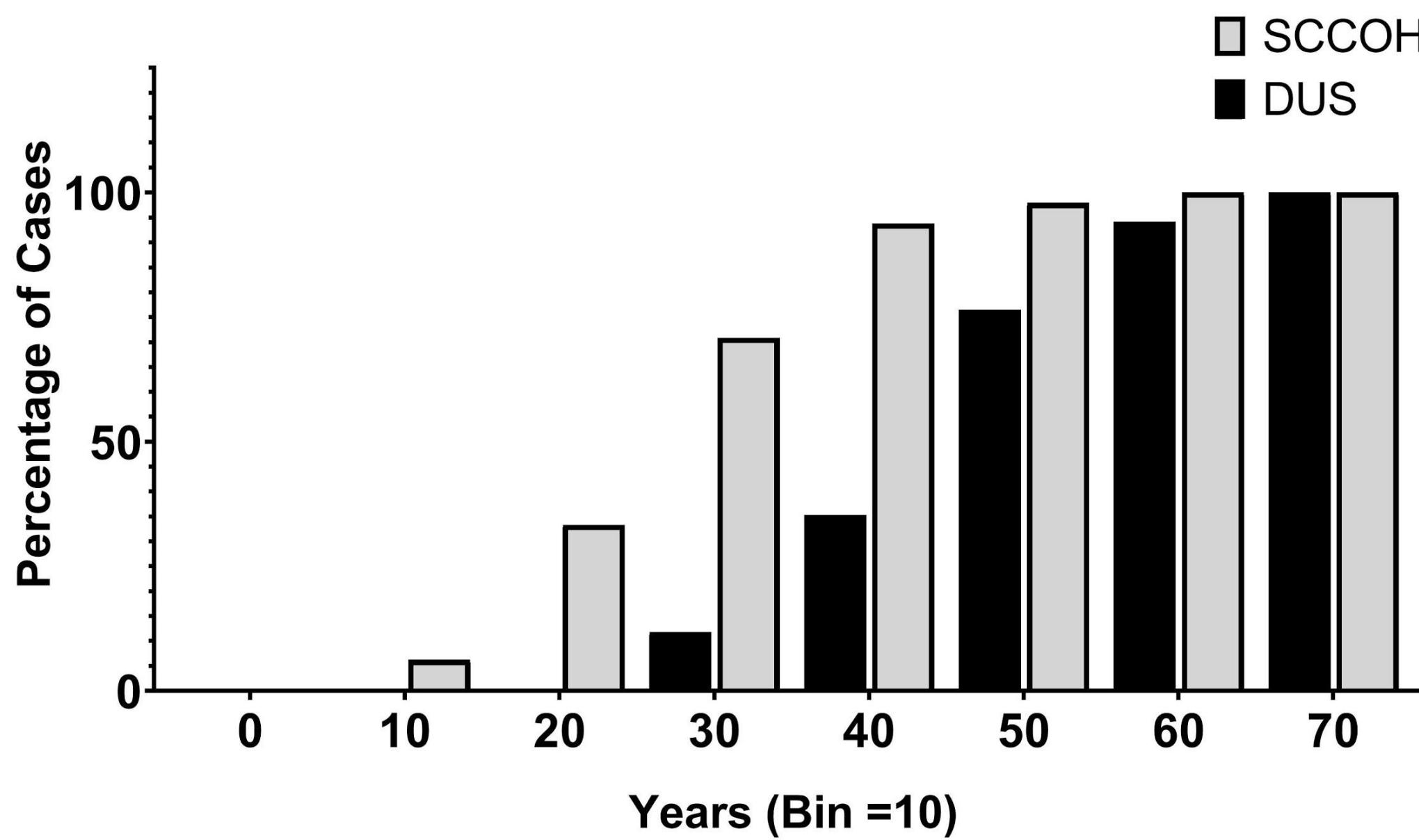


Supplementary Figure 1: Three generation family pedigree

a.



b.



Supplementary Figure 2: Histogram of SMARCA4 mutations

Age	Parity	Presenting Symptoms	Diagnosis	Stage	Tumor Size	Notable Labs	Surgery	Chemotherapy	Overall survival	Immunohistochemistry Characterization
31	0	Pelvic Pain and fullness	SCCOHT	IIIB	19cm	CA-125: 133, LDH: 621, normal serum calcium	Ex-lap, LSO, omentectomy, 3, LND	Cisplatin/etoposide x 3, Carboplatin/Paclitaxel x 1, Pembrolizumab x 2 cycles	175 Days	focally positive: cytokeratin cocktail, Sall4, Glyican 3, GATA3, calretinin negative: AFP, CD30, HCG, Oct3/4, Inhibin, EMA, Pax-8
55	1	Vaginal bleeding	DUS	IVB	9cm	CA-125: 100.7	Ex-lap, TAH, BSO, omentectomy	AIM 75/9 x 4 (20% dose reduction after cycle 2)	267 Days	positive: p16, WT-1, intact mismatch repair proteins negative: CD10, pan-keratin, ER, PR, synaptophysin, chromogranin, inhibin, HMB45, MART1, actin, desmin, and CD117, BCOR, cytokeratin cocktail (AE1/3&CAM5.2), EMA, BCL-1, BRG-1

Supplementary Table 1: Clinical Data Summary

Gene	Variants
CIC	A582V
DNM2	V64I
DOT1L	L1306F
FAM123B	H104R
FOXP1	P168H
HIST1H2AL	S2L
JAK2	T514M
JARID2	N226S
MYO18A	R1059C
PLCG2	A136T

Supplementary Table 2. Detected variations of unknown significance

SMARCA4 Alteration	SGZ prediction algorithm	Case	Mutation	Protein change	Mutation type	Protein Expression	Origin	Inheritance of germline mutations
p.Arg1077* and splice site c.2438+1G>A	Somatic	Le Loarer (2015)_SCCOHT7	c.3229C>T	p.Arg1077*	Stop	Loss	Unknown	
		Le Loarer (2015)_SCCOHT9	c.3229C>T	p.Arg1077*	Stop	Loss	Unknown	
		Unpublished_NF13	c.3229C>T	p.Arg1077*	Stop	Loss	Germline	Unknown
		SCCOHT-1 (Cell line)	c.3229C>T	p.Arg1077*	Stop	Loss	Unknown	
		BIN-67 (Cell line)	c.2438+1 G>A	NA	Splice	Loss	Unknown	
		Jelinic (2014)_107	c.3896_3907 del12	p.Glu1300_Asn1303del	In-frame deletion	Unknown	Somatic	
splice site 1119-1G>C and homozygous loss	Germline	Unpublished_Case27	c.1119-1G>C	NA	Splice	Loss	Somatic	
splice site 2859+1G>C	Germline	Unpublished_NF21	c.2859+1G>C	NA	Splice	Loss	Somatic	
		Unpublished_FA7a	c.196C>T	p.Gln66*	Stop	Loss	Germline	Inherited from father
		Unpublished_FA7b	c.2859+1G>C	NA	Splice	Loss	Germline	Inherited from father
p.Lys585fs*27 and p.Arg1093*	Germline	Kupryjanczyk (2014)_PJK1233	c.3277C>T	p.Arg1093*	Stop	Loss	Unknown	
		Unpublished_NF10	c.2184_2206del	p.Gln729fs	Frameshift	Loss	Germline	Unknown
		Ramos (2014)_DAH457	c.3277C>T	p.Arg1093*	Stop	Unknown	Unknown	

		Jelinic (2014)_106	c.1165_1165d delC	p.Ser391Profs*20	Frameshift	Loss	Somatic
			c.3277C>T	p.Arg1093*	Stop	Loss	
p.Arg979*	Somatic	Ramos (2014)_SCCO-008	c.2935C>T	p.Arg979*	Stop	Unknown	Germline
p.Arg381* and p.Ala573fs*40	Germline	Witkowski (2014)_UN7	c.1141C>T	p.Arg381*	Stop	Loss	Germline
splice site 2859+1G>A	Somatic	Jelinic (2014)_112	c.2859+1G>A	NA	Splice	Equivocal	Somatic
		Jelinic (2014)_106	c.1165_1165 delC	p.Ser391Profs*20	Frameshift	Loss	Somatic
			c.3277C>T	p.Arg1093*	Stop	Loss	
p.Arg1093*	Somatic	Ramos (2014)_DAH457	c.3277C>T	p.Arg1093*	Stop	Unknown	Unknown
		Unpublished_NF10	c.3277C>T	p.Arg1093*	Stop	Loss	Germline
		Kupryjanczyk (2014)_PJK1233	c.3277C>T	p.Arg1093*	Stop	Loss	Unknown
			c.2184_2206 del	p.Gln729fs	Frameshift	Loss	
		Unpublished_NF21	c.2859+1G>C	NA	Splice	Loss	Somatic
p.Gln66* and splice site 2859+1G>C	Somatic	Unpublished_FA7a	c.196C>T	p.Gln66*	Stop	Loss	
		Unpublished_FA7b	c.2859+1G>C	NA	Splice	Loss	Germline
			c.2859+1G>C	NA	Splice	Loss	Inherited from father
		Jelinic (2014)_106	c.1165_1165 delC	p.Ser391Profs*20	Frameshift	Loss	Somatic
p.Arg1093*	Indeterminate		c.3277C>T	p.Arg1093*	Stop	Loss	
		Ramos	c.3277C>T	p.Arg1093*	Stop	Unknown	Unknown

(2014)_DAH457

Unpublished_NF10	c.3277C>T	p.Arg1093*	Stop	Loss	Germline	Unknown
Kupryjanczyk (2014)_PJK1233	c.3277C>T	p.Arg1093*	Stop	Loss	Unknown	
	c.2184_2206 del	p.Gln729fs	Frameshift	Loss		

Supplementary Table 3. Previously reported mutations