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SWI/SNF Chromatin Remodeling Complexes and Cancer

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Abstract

The identification of mutations and deletions in the *SMARCB1* locus in chromosome band 22q11.2 in pediatric rhabdoid tumors provided the first evidence for the involvement of the SWI/SNF chromatin remodeling complex in cancer. Over the last 15 years, alterations in more than 20 members of the complex have been reported in a variety of human tumors. These include germline mutations and copy number alterations in *SMARCB1*, *SMARCA4*, *SMARCE1*, and *PBRM1* that predispose carriers to both benign and malignant neoplasms. Somatic mutations, structural abnormalities, or epigenetic modifications that lead to reduced or aberrant expression of complex members have now been reported in more than twenty percent of malignancies, including both solid tumors and hematologic disorders in both children and adults. In this review, we will highlight the role of *SMARCB1* in cancer as a paradigm for other tumors with alterations in SWI/SNF complex members and demonstrate the broad spectrum of mutations observed in complex members in different tumor types.

Keywords

SWI/SNF; *SMARCB1*; *SMARCA4*; rhabdoid tumor

Recent studies have established that cancer development depends on epigenetic alterations as well as genomic changes [Choi and Lee, 2013; Feinberg, 2014]. Multiple reports have demonstrated roles for altered DNA methylation, histone modifications and microRNA expression in the etiology of a wide variety of human cancers [Sarkar et al., 2013; Waldmann and Schneider, 2013]. The perturbation of SWI/SNF chromatin remodeling complexes is an emerging theme in cancer initiation and progression [Narlikar et al., 2013] and it is now postulated that at least 20% of all human tumors contain mutations in at least one member of the SWI/SNF complex [Kadoch et al., 2013]. In this review, we will

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specifically highlight those subunits that demonstrate germline alterations and predispose individuals to benign and malignant neoplasms.

SWI/SNF chromatin remodeling complexes regulate critical cellular processes including cell cycle progression, programmed cell death, differentiation, genomic instability and DNA repair [Narlikar et al., 2013]. The SWI/SNF complex, first discovered in *S. cerevisiae*, shows strong conservation from yeast to *Drosophila* to mammals and contains approximately 12–28 components [Kadoch et al., 2013; Yaniv, 2014]. The complex, containing one of 2 mutually exclusive ATPase subunits, BRG1/SMARCA4 or BRM/SMARCA2, physically alters nucleosome positioning using energy generated by ATP hydrolysis. The complexes can be subdivided into two broad categories, BAF or PBAF (Figure 1), based upon the presence of the ARID1A/B subunits, or ARID2 and PBRM1 subunits, respectively [Kadoch et al., 2013; Wei and Weissman, 2014]. Depending upon the configuration of complex components, SWI/SNF complexes can carry out a variety of cellular functions including neural differentiation [Lessard et al., 2007], embryonic stem cell differentiation [Ho et al., 2009], hepatic lipid metabolism [Li et al., 2008], glucose metabolism [Meng et al., 2013] and brain development [Tuoc et al., 2013]. However, the mechanisms that determine the types of cancers associated with inactivation of different complex members and how their losses fuel transformation remain undetermined.

SMARCB1 alterations and rhabdoid tumor

Rhabdoid tumors are rare pediatric malignancies that most often arise in the brain, kidney and soft tissues. The peak incidence is in the first several years of life, although we have studied congenital tumors that arise during fetal development and tumors that have presented in late adulthood. Regardless of the anatomic location, rhabdoid tumors are highly aggressive malignancies, and combined approaches using surgery, chemotherapy and/or radiation are required for treatment [Hilden et al., 2004]. Although the tumors may be rapidly fatal, individual case reports have described patients who have survived for up to 26 years with multiple recurrences [Takahashi-Fujigasaki et al., 2012]. The identification of deletions and mutations in the *SMARCB1/INI1/BAF47/hSNF5* locus, hereafter referred to as *SMARCB1*, in pediatric rhabdoid tumors [Versteege et al., 1998] served as a paradigm for how genetic alterations in chromatin-remodeling complex subunits might play a role in tumor development. The identification of germline mutations in *SMARCB1* [Biegel et al., 1999], accompanied by loss of the normal allele, confirmed that rhabdoid tumorigenesis followed the classic two-hit model for a tumor suppressor gene.

Germline and acquired alterations of *SMARCB1* and rhabdoid tumor

The *SMARCB1* locus is located in chromosome band 22q11.2, distal to the region that is typically deleted in DiGeorge and Velo-cardio-facial syndromes. The alterations in *SMARCB1* include intragenic and whole gene deletions, intragenic duplications, and mutations. The molecular characterization of two large cohorts of patients with rhabdoid tumors has been reported [Eaton et al., 2011; Bourdeaut et al., 2011] which revealed an overall incidence of germline alterations of *SMARCB1* in 35% of patients. Virtually all patients with two primary tumors have a germline mutation or deletion of *SMARCB1*,

however the incidence is also high in children with primary brain tumors (atypical teratoid/rhabdoid tumor; AT/RT) and renal tumors. In contrast, extra-renal rhabdoid tumors are typically associated with bi-allelic, somatic deletions of *SMARCB1*. A summary of the germline alterations in *SMARCB1* in 70 rhabdoid tumor patients studied from one of the author's laboratory is shown in Figure 2. The percentages of patients with whole gene deletions (22%), intragenic deletions and duplications (24%), nonsense mutations (24%) and frameshift mutations (20%) were similar. However, unlike other tumor suppressor genes, such as *TP53* and *WT1*, there were no missense mutations, and only 10% of the patients had splice-site mutations. The second inactivating event in the tumors from these individuals is typically a deletion of the remaining copy of *SMARCB1*, or a copy number neutral loss of heterozygosity event that results in duplication of the mutated allele. The types of mutations in sporadic tumors are similar to those observed in the germline; essentially all copy number alterations or truncating mutations. Missense mutations are virtually absent and splice site mutations are uncommon.

The vast majority of germline deletions and mutations appear de novo, due in part to the fatal nature of the disease. Several families have been reported with two or more affected siblings in which there is presumed gonadal mosaicism, and two of our patients demonstrated germline mosaicism for a single exon deletion. While a few multi-generation families have been described with an appearance of some reduced penetrance, no established risk estimates for cancer in *SMARCB1* mutation carriers exists. This has become an increasingly difficult issue in providing genetic counseling for families in which an individual is found to have a deletion in *SMARCB1* as part of a contiguous 22q11.2 deletion syndrome, but who does not have a tumor. As whole exome and ultimately whole genome sequencing studies become established as clinical diagnostic tests, the identification of *SMARCB1* mutations as an incidental finding will increase. Without established risk estimates for cancer in unaffected carriers, it will be challenging to provide guidance for cancer surveillance over an individual's lifetime. Furthermore, as presented in the accompanying articles in this special issue, and as shown in Figure 1, the number of genes in the SWI/SNF and other chromatin remodeling complexes mutated in genetic disorders such as Coffin-Siris or Nicolaides-Baraitser syndrome, autism spectrum disorders and intellectual disability is also increasing. The current inability to predict the phenotype associated with mutations or deletions in these genes will be particularly challenging in a prenatal or neonatal setting.

SMARCB1 alterations and schwannomatosis

As shown in Table 1 and reviewed by Smith et al. [Smith et al., 2014], germline mutations in *SMARCB1* are also seen in association with schwannomatosis, in which affected individuals develop multiple, benign nerve sheath tumors (schwannomas). Approximately 45% of patients with familial schwannomatosis and 9% of patients with apparently sporadic schwannomas have germline mutations in *SMARCB1*. However, in contrast to the whole gene deletions and truncating mutations observed in in rhabdoid tumor patients, the mutations in schwannomatosis are primarily splice site mutations and missense mutations in exons 1 and the 3'UTR [Hulsebos et al., 2007; Hadfield et al., 2008; Rousseau et al., 2011; Smith et al., 2012; Smith et al., 2014]. To date, one patient with a missense mutation in exon

9 of *SMARCB1*, who has both schwannomatosis and Coffin-Siris syndrome (Gossai et al, in preparation), has been reported. These studies suggest that loss of function truncating mutations and whole gene deletions are associated with more aggressive tumors, including rhabdoid tumors and malignant peripheral nerve sheath tumors. These tumors most often arise during infancy and early childhood. In contrast, germline missense mutations are more often associated with developmental disorders and late onset, typically benign tumors. We have described several families [Eaton et al., 2011; Carter et al., 2012] in which the identical truncating mutation led to a rhabdoid tumor in a young child, and the development of schwannomas in the adult carrier(s) in the previous generation. This led to the hypothesis that an early developmental window occurs in which the risk for rhabdoid tumors is highest, consistent with the peak incidence at 6 months of age in germline mutation carriers. After three years of age, the incidence dramatically decreases [Eaton et al., 2011]. Therefore, the cells of origin for rhabdoid tumor and schwannoma may differ, with the nature of the SWI/SNF complexes in those cells dictating the morphology and clinical behavior of the resulting neoplasms.

In addition to the germline mutations reported in rhabdoid tumors and schwannomatosis, Van den Munckhof et al. [van den Munckhof et al., 2012] identified *SMARCB1* mutations in families with multiple meningiomas of the falx cerebri of the cranium. Loss of function mutations in the *SMARCE1* gene have recently been described in several families with multiple meningiomas of the spine [Smith et al., 2013], suggesting that a genotype-phenotype correlation exists between histology, anatomic location and gene mutation. Reports of *SMARCE1* mutations in breast cancer cell lines [Kiskinis et al., 2006], as well as in a primary breast tumor [Villaronga et al., 2011], make it likely that germline mutations in *SMARCE1* will ultimately arise in other tumor types.

Cancer predisposition associated with *SMARCA4* mutations

SMARCB1 is the primary gene associated with rhabdoid tumors of the brain, kidney and extra-renal sites. In fact, homozygous inactivation of this locus appears to be sufficient for tumorigenesis, as whole exome sequencing of primary tumors failed to identify any additional non-random coding sequence mutations [Lee et al., 2012]. A second rhabdoid tumor locus was identified when germline and somatic mutations in *SMARCA4* were found in a small number of patients with rhabdoid tumors who did not have *SMARCB1* loss [Schneppenheim et al., 2010; Hasselblatt et al., 2011; Witkowski et al., 2013]. As shown in Table 1, a wide variety of solid tumors demonstrate missense and loss of function alterations in *SMARCA4*. To date, the only other tumor type to demonstrate bi-allelic inactivation of *SMARCA4*, consistent with a cancer predisposing germline mutation and second somatic alteration, is small cell carcinoma of the ovary, hypercalcemic type (SCCOHT) [Ramos et al., 2014; Witkowski et al., 2014; Jelinic et al., 2014]. Based on the relatively early age at presentation, and the presence of rhabdoid appearing cells by histology, it has been proposed that SCCOHT represents another type of extra-renal rhabdoid tumor [Foulkes et al., 2014]. The germline mutations in SCCOHT included both missense and truncating mutations, typically with loss of the wild-type allele as the second inactivating event in the tumor. In contrast to the typical findings in tumors, and similar to *SMARCB1* mutations, the

SMARCA4 mutations in developmental disorders are all missense, non-truncating mutations, or in-frame deletions, as described in the accompanying papers in this issue.

Germline mutations in *PBRM1* associated with clear cell sarcoma of the kidney

Varela et al. [Varela et al., 2011] were the first to describe *PBRM1* mutations in renal cell carcinoma, which were highly associated with the clear cell subtype. Patients with and without germline *VHL* mutations, which characterize the majority of renal cell carcinomas, demonstrated somatic *PBRM1* mutations, initially suggesting that *PBRM1* mutations may have been a later genetic event in tumor development. Both genes map to 3p, thus deletions of 3p in tumors would unmask recessive mutations in both *VHL* and *PBRM1*. A subsequent report by Pena-Llopis [Pena-Llopis et al., 2012] described germline mutations of *PBRM1* in five patients with clear cell renal cell carcinoma, including four frameshift and one missense mutation. Intriguingly, three of these patients also had germline mutations in *VHL*. The occurrence of germline mutations in two different cancer predisposition genes is highly unusual. The identification of somatic *BAP1* mutations in renal cell carcinoma [Pena-Llopis et al., 2012], also in tumors with *PBRM1* or *VHL* mutations, was also interesting given that *BAP1* is a cancer predisposition gene. *BAP1* and *PBRM1* mutations were most often mutually exclusive, however those tumors with *PBRM1* and *BAP1* mutations had rhabdoid features, and *BAP1* loss was associated with a higher grade, compared with those tumors with *PBRM1* loss. As shown in Table 1, mutations in *PBRM1* have been described in a variety of other carcinomas, raising the possibility that germline alterations could contribute to a broader spectrum of cancers.

Somatic mutations in SWI/SNF complex components

A shown in Table 1 and Figure 1, mutations in multiple members of the SWI/SNF complex have been found in human cancers including non-small cell lung cancer (NSCLC), ovarian carcinomas and renal cell carcinomas [Fukuoka et al., 2004; Medina et al., 2004; Medina et al., 2008; Reisman et al., 2003; Rodriguez-Nieto et al., 2011; Weissman and Knudsen, 2009; Wilson and Roberts, 2011]. Indeed, Kadoch et al. and Wang et al. recently reported that ~20% of all human tumors show mutations in SWI/SNF components [Kadoch et al., 2013; Wang et al., 2014b]. Table 1 provides an overview of the range of mutations found in primary tumors, not including cell lines and xenografts, originating in a broad spectrum of human tissues.

The ubiquitous presence of SWI/SNF complex mutations in human cancer raises several fundamental questions about their roles in human tumor development. For example, in yeast, loss of any complex component yields similar phenotypes [Laurent et al., 1991; Peterson and Herskowitz, 1992; Wang et al., 1996]. However, it seems clear that loss of different SWI/SNF complex members in humans gives rise to different spectra of tumors. Therefore, how does the loss of each individual complex member affect the activity of the remaining SWI/SNF complexes? Potential mechanisms include altered nucleosome positioning induced by absent or aberrant SWI/SNF complex activity that lead to changes in DNA accessibility for RNA polymerase II or transcription factors [Kuwahara et al., 2013;

Tolstorukov et al., 2013], secondary effects on histone modifications through altered interactions with histone mark readers, writers and erasers [Hargreaves and Crabtree, 2011], and direct or indirect effects on DNA methylation [Berdasco and Esteller, 2013; Banine et al., 2005]. Once we have a better understanding of these mechanisms, we can address a second important issue- the different effects of complex activities between complete loss of a complex member versus the consequences of missense mutations. Whether the missense mutations in complex members found in developmental disorders represent another mechanism for complete loss of activity or the acquisition of new SWI/SNF functions remains an exciting and open question.

Genetically engineered mouse models of *Smarcb1/Snf5* and other complex member loss

Murine models in which *Smarcb1/Snf5* was knocked out in various cell types confirmed that homozygous loss of the locus results in embryonic lethality, and that heterozygous carriers developed tumors following somatic loss of the wild-type allele [Guidi et al., 2001; Klochendler-Yeivin et al., 2000; Roberts et al., 2000; Roberts et al., 2002]. Although the tumors formed in the *Smarcb1*$^{+/-}$ mice resembled the histopathology of human rhabdoid tumors, they occurred after a long latency period (>7 months) with a low penetrance (10–30%) [Guidi et al., 2001; Klochendler-Yeivin et al., 2000; Roberts et al., 2000]. Considering the absence of mutations of well-established oncogenes and tumor suppressor genes in human rhabdoid tumors, additional studies have examined the effects of *Smarcb1* inactivation in tandem with other cancer-driving genes [Lee et al., 2012]. While *Smarcb1* loss in the absence of the *Tp53* tumor suppressor gene resulted in a dramatic acceleration of tumor development, concomitant loss of *Smarcb1* and *Rb* or *p16*^{INK4A} did not affect MRT development [Isakoff et al., 2005; Klochendler-Yeivin et al., 2006]. However, inactivation of the Rb family of genes gene through the expression of a truncated form of T antigen resulted in an increased tumor penetrance of neural system tumors in spinal cords or brains depending upon the genetic background [Chai et al., 2007; Kuwahara et al., 2012]. In contrast, simultaneous loss of *Smarcb1* with *Ccnd1*, *Ezh2* or *Smarca4/Brg1* either suppressed or eliminated tumor development in mouse models [Tsikitis et al., 2005; Wang et al., 2009; Wilson et al., 2010]. The virtual loss of tumor development in the absence of EZH2 emphasizes the strong interactions between the Polycomb and SWI/SNF complexes while the effects of SMARCA4 loss suggests that the residual SWI/SNF complexes lacking SMARCB1 may gain oncogenic activity.

Similar to the *Smarcb1* genetically engineered mouse model (GEMMs), homozygous knockout of *Smarca4, Srg3/Smarcc1/Baf155* and *Pbrm1/Baf180* mice show embryonic lethality [Bultman et al., 2000; Han et al., 2008; Wang et al., 1999]. However, rhabdoid tumors have not appeared in knockouts of other family members including *Smarca4* [Bultman et al., 2000; Bultman et al., 2007], *Brm/Smarca2* [Reyes et al., 1998] or *Baf155/Smarcc1/Srg3* [Han et al., 2008]. Instead, loss of *Smarca4* in GEMMs contributes to the development of mammary, lung and uterine tumors as well as ovarian cysts [Bultman et al., 2007; Glaros et al., 2008; Serber et al., 2012; von Figura et al., 2014]. Loss of *Smarca2* has been associated with lung and prostate tumor development while *Smarcc1* inactivation

results in mainly sarcomas [Ahn et al., 2010; Glaros et al., 2007; Shen et al., 2008]. A tumor phenotype has not been reported for GEMMs involving *Pbrm1/Baf180* inactivation. The reports that heterozygous knockout mice of different SWI/SNF complexes develop divergent tumors appear consistent with the observed differences in tumor specificity found in humans with germline mutations in complex members (see above). The mechanisms that drive these differences remain unknown but may result from the different types of SWI/SNF complexes remaining in the absence of each unique component. The association between complex member loss and the appearance of specific tumors may also reflect their individual roles in the development of specific tissues. This notion would potentially link tumor development with the appearance of developmental disorders found in individuals with germline missense mutations in complex components.

With the development of a GEMM expressing a conditional knockout allele of *Smarcb1*, attempts were made to define a cell of origin for rhabdoid tumors. However, GEMMs with tissue-restricted inactivation of *Smarcb1* either developed aggressive lymphomas [Roberts et al., 2002; Isakoff et al., 2005] or led to developmental blocks [Gresh et al., 2005]. Although these approaches have not yet proved successful, based upon the complex histology and immunophenotypic profiles of the tumors, our current hypothesis proposes that rhabdoid tumors arise from a primitive stem cell.

Future perspectives

The large volume of high impact publications during the past 15 years emphasize that mutations in the SWI/SNF complex play a key role in a broad spectrum of human diseases. Not surprisingly, a complex that regulates central and essential features of cellular functions, including chromatin organization, RNA transcription, DNA damage response and meiosis, may provide a significant challenge for understanding its roles in developmental disorders and cancer. The fact that we have not fully identified the number and composition of SWI/SNF complexes in normal human tissues further exacerbates this problem. However, the studies discussed in this special issue can provide a framework in which to tackle these issues. As more high-throughput gene sequencing and gene expression studies become available along with the identification of protein constituents of individual complexes through proteomic advances, we should gain new insights in the complex's normal and aberrant activities. Combined with better cell culture models and expansion of GEMMs, we can use these data to develop rational treatments for these important human diseases.

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The authors are investigators in the Departments of Pathology and Pediatrics, and have investigated the role of several members of the SWI/SNF complex in primary tumors, cell lines, and mouse models with respect to tumor development.

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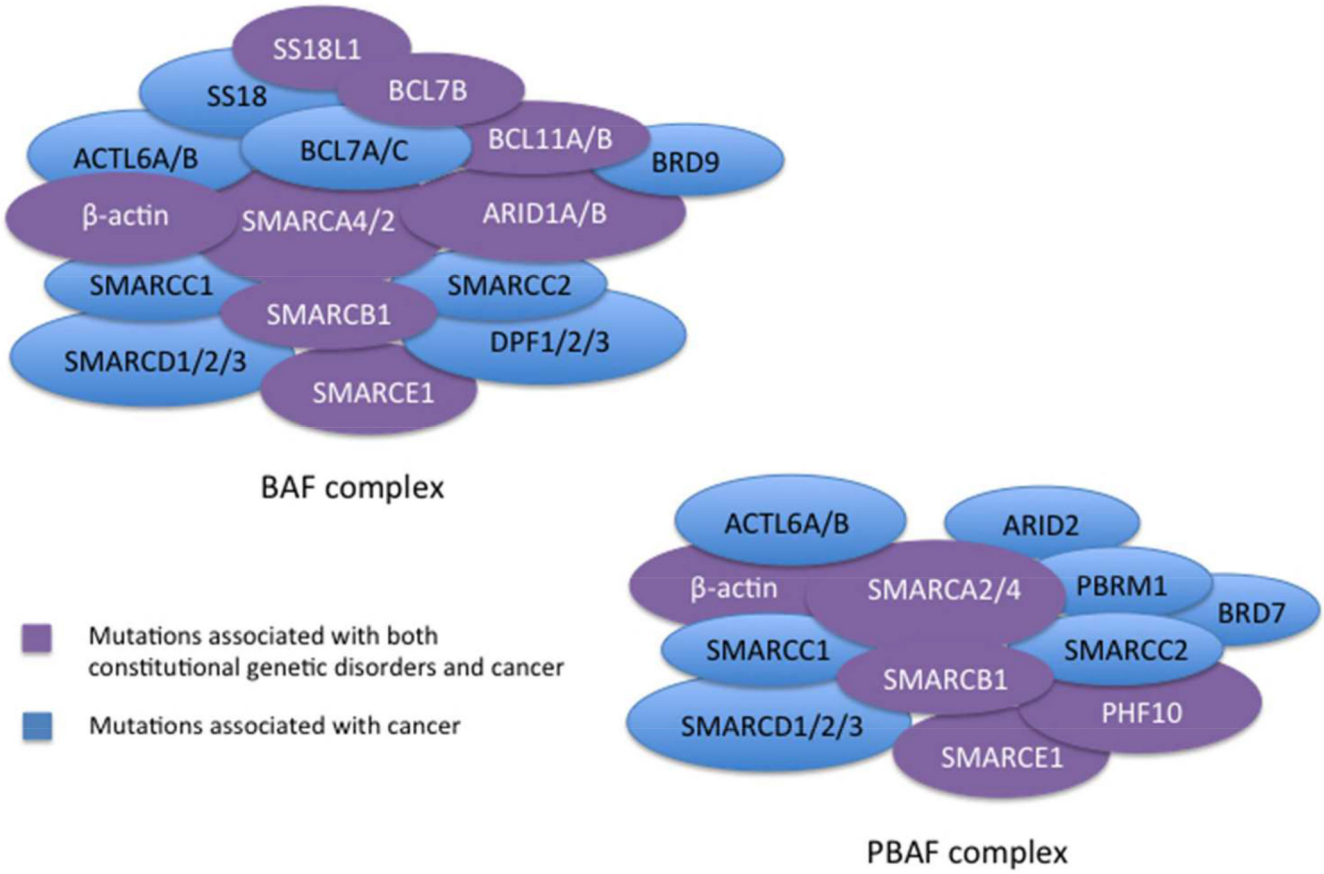

Figure 1.

BAF and PBAF complexes. The subunits specific to the BAF complex including ARID1A and ARID1B and to the PBAF complex including ARID2 and PBRM1 are demonstrated in the cartoon rendition of the complexes. Mutations in subunits colored in blue have been demonstrated in the literature to be associated with cancer, and subunits with mutations that have been associated with either cancer and/or known genetic disorders (i.e. Coffin-Siris syndrome) are colored in purple.

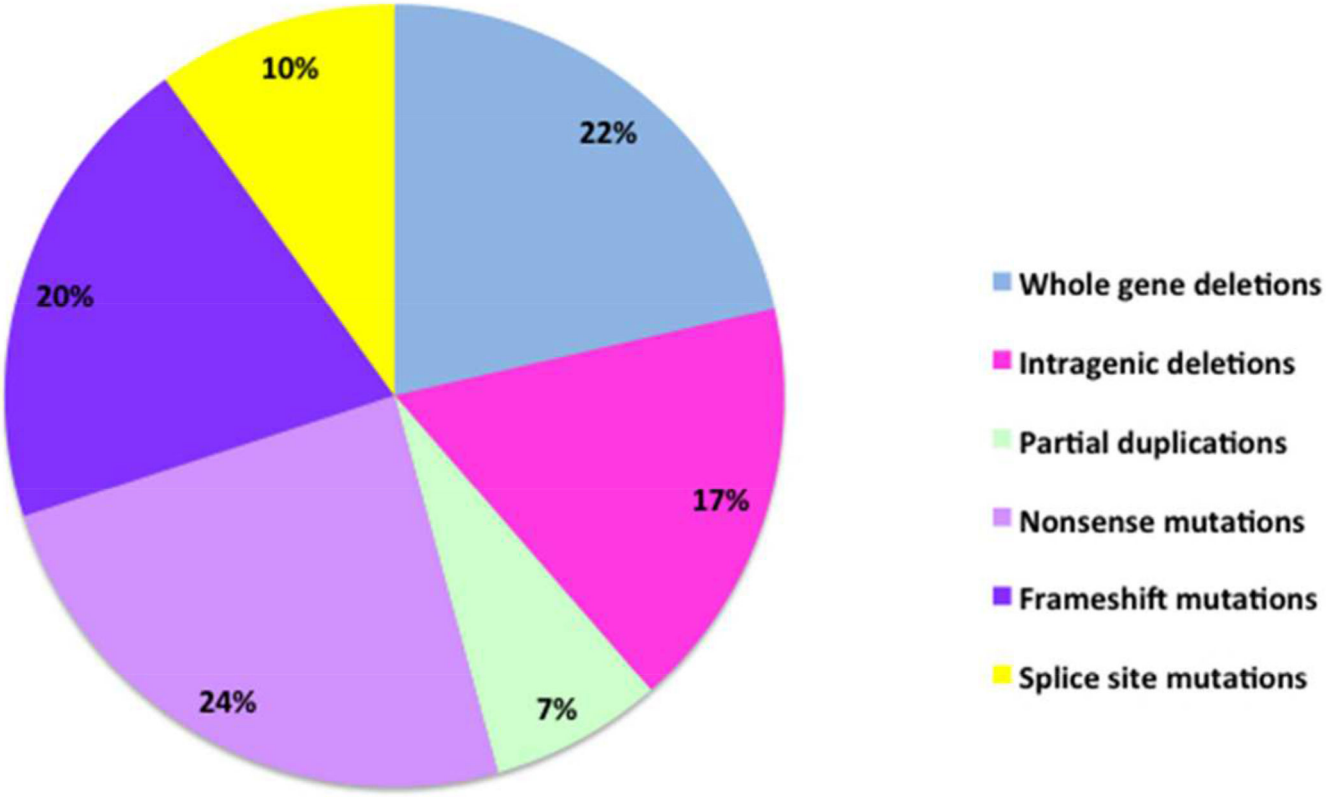

Figure 2.

Germline *SMARCB1* alterations in patients with rhabdoid tumor. Deletions of *SMARCB1*, both whole gene (22%) and intragenic (24%), encompass the majority of the germline alterations observed in rhabdoid tumors. A large percentage of the remaining germline alterations observed in *SMARCB1* are frameshift mutations (20%) followed by nonsense mutations (17%). A smaller percentage of germline SMARCB1 alterations result from splice site mutations (10%) and partial gene duplications (7%).

Am J Med Genet C Semin Med Genet. Author manuscript; available in PMC 2015 September 01.

Table 1

Alterations in SWI/SNF components reported in primary human tumors*

Subunit	Gene	Cancer Types	Alteration	Germline	*Missense*	*Truncating*	*Loss*	*Gene fusion*	Reference(s)
Core complex subunits									
BRM	*SMARCA2*	Adenoid cystic carcinoma	Mutation, deletion, and amplification						[Ho et al., 2013]
		Non-melanoma skin cancer	Mutation						[Moloney et al., 2009; Bock et al., 2011]
		Hepatocellular carcinoma	Deletion						[Endo et al., 2013]
		Head and neck squamous cell carcinoma	Deletion						[Gunduz et al., 2009; Wang et al., 2013]
		Gastric cancer	Decreased expression						[Yamamichi et al., 2007]
		Clear cell renal cell carcinoma	Decreased expression						[Xia et al., 2014]
		Prostate cancer	Decreased expression						[Shen et al., 2008]
		Lung cancer (adenocarcinoma and squamous cell carcinoma)	Decreased expression						[Reisman et al., 2003]
BRG1	*SMARCA4*	Small-cell carcinoma of the ovary, hypercalcemic type (malignant rhabdoid tumor of the ovary)	Biallelic inactivation	Yes					[Jelinic et al., 2014; Ramos et al., 2014; Kupryjanczyk et al., 2013; Witkowski et al., 2014]
		Rhabdoid tumor	Biallelic inactivation	Yes					[Schneppenheim et al., 2010; Hasselblatt et al., 2011; Witkowski et al., 2013]
		Medulloblastoma	Mutation						[Parsons et al., 2011; Robinson et al., 2012; Pugh et al., 2012; Jones et al., 2012a]
		Lung adenocarcinoma	Mutation and deletion	Rare					[Imielinski et al., 2012; Seo et al., 2012; Medina et al., 2004; Rodriguez-Nieto et al., 2011]

Subunit	Gene	Cancer Types	Alteration	Germline	*Missense*	*Truncating*	*Loss*	*Gene fusion*	Reference(s)
		Mantle cell lymphoma	Mutation						[Zhang et al., 2014]
		Burkitt lymphoma	Mutation						[Love et al., 2012]
		Hepatocellular carcinoma	Mutation and deletion						[Endo et al., 2013]
		Esophageal adenocarcinoma	Mutation and translocation						[Dulak et al., 2013]
		Melanoma	Mutation						[Hodis et al., 2012]
		Non-melanoma skin cancer	Decreased expression						[Bock et al., 2011]
		Intraductal papillary mucinous neoplasms of the pancreas	Decreased expression						[Dal Molin et al., 2012]
BAF47	*SMARCB1*	Rhabdoid tumor	Biallelic inactivation	Yes					[Versteege et al., 1998; Biegel et al., 1999]
		Schwannoma	Biallelic inactivation	Yes					[Hulsebos et al., 2007; Hadfield et al., 2008; Rousseau et al., 2011; Smith et al., 2012]
		Meningioma	Biallelic inactivation	Rare					[van den Munckhof et al., 2012; Christiaans et al., 2011; Schmitz et al., 2001]
		Epithelioid sarcoma	Biallelic inactivation	Rare					[Le Loarer et al., 2014; Sullivan et al., 2013]
		Cribriform neuroepithelial tumor	Biallelic inactivation						[Arnold et al., 2013; Hasselblatt et al., 2009]
		Renal medullary carcinoma	Deletion						[Liu et al., 2013; Calderaro et al., 2012]
BAF155	*SMARCC1*	Colorectal carcinoma	Increased expression						[Andersen et al., 2009]
		Prostate cancer	Increased expression						[Heeboll et al., 2008]
		Cervical intraepithelial neoplasia	Increased expression						[Shadeo et al., 2008]
BAF170	*SMARCC2*	Gastric cancer	Mutation						[Kim et al., 2013]
		Colorectal carcinoma	Mutation						[Kim et al., 2013]

Am J Med Genet C Semin Med Genet. Author manuscript; available in PMC 2015 September 01.

Subunit	Gene	Cancer Types	Alteration	Germline	*Missense*	*Truncating*	*Loss*	*Gene fusion*	Reference(s)
BAF60A	*SMARCD1*	Breast cancer	Mutation		■	■			[Stephens et al., 2012]
		Gastric cancer	Mutation		■	■			[Wang et al., 2014a]
BAF60B	*SMARCD2*	Lung adenocarcinoma	Mutation		■				[Imielinski et al., 2012]
		Colon cancer	Mutation		■				[Seshagiri et al., 2012]
BAF60C	*SMARCD3*	Neuroblastoma	Increased expression						[Takita et al., 2004]
BAF57	*SMARCE1*	Multiple spinal meningiomas	Mutation and deletion	Yes		■	■		[Smith et al., 2013]
BAF53A	*ACTL6A*	Colorectal carcinoma	Mutation		■	■			[Cancer Genome Atlas Network, 2012]
		Lung adenocarcinoma	Mutation		■				[Imielinski et al., 2012]
BAF53B	*ACTL6B*	Urothelial cancer	Decreased expression						[Ibragimova et al., 2014]
		Hepatocellular carcinoma	Decreased expression						[Revill et al., 2013]
Beta-actin	*ACTB*	Pericytoma with t(7;12)	Translocation					■	[Bridge et al., 2012; Dahlen et al., 2004b; Dahlen et al., 2004a]
		Diffuse large B-cell lymphoma	Mutation		■				[Lohr et al., 2012]
BAF complex subunits									
BAF45B	*DPF1*	Esophageal adenocarcinoma	Mutation		■				[Dulak et al., 2013]
		Lung adenocarcinoma	Mutation		■	■			[Imielinski et al., 2012]
		Colon cancer	Mutation		■				[Seshagiri et al., 2012]
BAF45C	*DPF3*	Esophageal adenocarcinoma	Mutation		■				[Dulak et al., 2013]
		Lung adenocarcinoma	Mutation		■	■			[Imielinski et al., 2012]
		Colorectal cancer	Mutation		■				[Cancer Genome Atlas Network, 2012]
BAF45D	*DPF2*	Esophageal adenocarcinoma	Mutation		■	■			[Dulak et al., 2013]

Subunit	Gene	Cancer Types	Alteration	Germline	*Missense*	*Truncating*	*Loss*	*Gene fusion*	Reference(s)
		Lung adenocarcinoma	Mutation						[Imielinski et al., 2012]
		Colorectal cancer	Mutation						[Cancer Genome Atlas Network, 2012]
BAF250A	*ARID1A*	Ovarian clear cell carcinoma	Mutation and deletion						[Wiegand et al., 2010; Jones et al., 2010]
		Endometrioid ovarian carcinoma	Mutation						[Wiegand et al., 2010]
		Endometrial carcinoma	Mutation						[Liang et al., 2012; Guan et al., 2011; Le Gallo et al., 2012]
		Cervical carcinoma	Decreased expression						[Katagiri et al., 2012; Cho et al., 2013]
		Breast cancer	Mutation and deletion						[Jones et al., 2012b; Stephens et al., 2012; Cornen et al., 2012; Mamo et al., 2012]
		Pancreatic ductal adenocarcinoma	Mutation and deletion						[Biankin et al., 2012; Birnbaum et al., 2011]
		Pancreatic carcinoma with acinar differentiation	Mutation						[Jiao et al., 2014]
		Hepatocellular carcinoma	Mutation and deletion						[Huang et al., 2012; Fujimoto et al., 2012a; Guichard et al., 2012]
		Intrahepatic cholangiocarcinomas	Mutation						[Ross et al., 2014; Jiao et al., 2013]
		Gastric adenocarcinoma	Mutation and deletion						[Zang et al., 2012; Wang et al., 2011; Wang et al., 2014a]
		Esophageal adenocarcinoma	Mutation						[Dulak et al., 2013]
		Oesophagogastric junctional adenocarcinoma	Mutation						[Chong et al., 2013]
		Colorectal carcinoma	Mutation						[Cancer Genome Atlas Network,

Subunit	Gene	Cancer Types	Alteration	Germline	*Missense*	*Truncating*	*Loss*	*Gene fusion*	Reference(s)
									2012; Cajuso et al., 2014 Cajuso et al., 2014
		Renal clear cell carcinoma	Mutation						[Varela et al., 2011]
		Transitional cell carcinoma of the bladder	Mutation						[Gui et al., 2011]
		Urothelial bladder carcinoma	Mutation						[Balbas-Martinez et al., 2013]
		Medulloblastoma	Mutation						[Jones et al., 2012b; Parsons et al., 2011]
		Neuroblastoma	Mutation						[Sausen et al., 2013; Pugh et al., 2013]
		Lung adenocarcinoma	Mutation and deletion						[Imielinski et al., 2012; Seo et al., 2012]
		Pulmonary carcinoids	Mutation						[Fernandez-Cuesta et al., 2014]
		Adenoid cystic carcinoma	Mutation and deletion						[Ho et al., 2013]
		Prostate cancer	Mutation						[Jones et al., 2012b; Barbieri et al., 2012]
		Burkitt lymphoma	Mutation						[Love et al., 2012; Giulino-Roth et al., 2012]
		Diffuse large B-cell lymphoma	Mutation						[Zhang et al., 2013]
		Follicular lymphoma	Mutation						[Li et al., 2014]
		Melanoma	Mutation						[Hodis et al., 2012]
BAF250B	*ARID1B*	Hepatocellular carcinoma	Mutation and deletion						[Fujimoto et al., 2012a]
		Colorectal carcinoma	Mutation						[Cajuso et al., 2014]
		Breast cancer	Mutation						[Stephens et al., 2012]
		Prostate cancer	Mutation						[Barbieri et al., 2012]

Subunit	Gene	Cancer Types	Alteration	Germline	*Missense*	*Truncating*	*Loss*	*Gene fusion*	Reference(s)
		Neuroblastoma	Deletion				■		[Sausen et al., 2013; Pugh et al., 2013]
		Melanoma	Mutation		■	■			[Hodis et al., 2012]
BCL7A	*BCL7A*	Pilocytic astrocytoma	Deletion				■		[Potter et al., 2008]
		Mycosis fungoides (primary cutaneous T cell lymphoma subtype)	Deletion				■		[Carbone et al., 2008]
		Multiple myeloma	Decreased expression						[Ramos-Medina et al., 2013]
		Cutaneous T cell lymphoma	Decreased expression						[van Doorn et al., 2005]
BCL7B	*BCL7B*	Pilocytic astrocytoma	Deletion				■		[Potter et al., 2008]
		Gastric cancer	Mutation		■				[Wang et al., 2014a]
BCL7C	*BCL7C*	Gastric cancer	Mutation		■	■			[Wang et al., 2014a]
BCL11A	*BCL11A*	Non-small cell lung cancer	Increased expression						[Jiang et al., 2013]
		Lung squamous cell carcinoma	Copy number amplification						[Boelens et al., 2009; Jiang et al., 2013]
		Chronic lymphocytic leukemia	Translocation and copy number gain					■	[Pfeifer et al., 2007; Ferreira et al., 2008; Yin et al., 2009]
		Acute lymphoblastic leukemia	Increased expression						[Agueli et al., 2010]
		Acute myeloid leukemia	Translocation					■	[Trubia et al., 2006]
		Low-grade B cell lymphoma	Copy number gain						[Ferreira et al., 2008]
		Mediastinal B cell lymphoma	Copy number gain/amplification						[Weniger et al., 2006]
		Diffuse large B-cell lymphoma	Copy number gain/amplification						[Fukuhara et al., 2006; Bea et al., 2004]
		Marginal zone B cell lymphoma	Copy number gain/amplification						[Flossbach et al., 2013]
		Gray zone lymphoma	Copy number gain/amplification						[Eberle et al., 2011]

Am J Med Genet C Semin Med Genet. Author manuscript; available in PMC 2015 September 01.

Subunit	Gene	Cancer Types	Alteration	Germline	*Missense*	*Truncating*	*Loss*	*Gene fusion*	Reference(s)
		Classical Hodgkin lymphoma	Copy number gain/amplification						[Martin-Subero et al., 2002]
BCL11B	*BCL11B*	Acute myeloid leukemia	Focal amplification/ translocation						[Abbas et al., 2014; Bezrookove et al., 2004]
		T cell acute lymphoblastic leukemia	Deletion, mutation, and gene fusions						[Przybylski et al., 2005; De Keersmaecker et al., 2010; Gutierrez et al., 2011; Kraszewska et al., 2013; Van Vlierberghe et al., 2013]
		Mycosis fungoides (primary cutaneous T cell lymphoma subtype)	Increased expression						[Gu et al., 2013]
		Adult T cell leukemia/lymphoma	Decreased expression and translocation						[Kurosawa et al., 2013; Fujimoto et al., 2012b]
		Head and neck squamous cell carcinoma	Increased expression						[Ganguli-Indra et al., 2009]
BRD9	*BRD9*	Gastric cancer	Mutation						[Wang et al., 2014a]
SS18L1	*CREST*	Synovial sarcoma	Translocation						[Storlazzi et al., 2003]
SS18	*SYT*	Synovial sarcoma	Translocation						[Crew et al., 1995; Fligman et al., 1995; Panagopoulos et al., 2001]
PBAF complex subunits									
BAF45A	*PHF10*	Colon cancer	Mutation						[Seshagiri et al., 2012]
		Hepatocellular carcinoma	Mutation						[Kan et al., 2013]
BAF180	*PBRM1*	Renal clear cell carcinoma	Biallelic inactivation	Yes					[Varela et al., 2011; Pena-Llopis et al., 2012; Duns et al., 2012; Guo et al., 2011]
		Intrahepatic cholangiocarcinomas	Mutation						[Jiao et al., 2013]
		Gallbladder carcinoma	Mutation						[Jiao et al., 2013]

Subunit	Gene	Cancer Types	Alteration	Germline	*Missense*	*Truncating*	*Loss*	*Gene fusion*	Reference(s)
		Breast cancer	Mutation and LOH						[Xia et al., 2008]
		Esophageal adenocarcinoma	Mutation						[Dulak et al., 2013]
BAF200	*ARID2*	Hepatocellular carcinoma	Mutation and deletion						[Fujimoto et al., 2012a; Guichard et al., 2012; Li et al., 2011]
		Pancreatic ductal adenocarcinoma	Mutation and deletion						[Biankin et al., 2012]
		Non-small cell lung cancer	Biallelic inactivation						[Manceau et al., 2013]
		Colorectal carcinoma	Mutation						[Cajuso et al., 2014]
		Esophageal adenocarcinoma	Mutation						[Dulak et al., 2013]
		Oral squamous cell carcinoma (gingivo-buccal)	Mutation						[India Project Team of the International Cancer Genome Consortium, 2013]
		Breast cancer	Mutation						[Stephens et al., 2012]
		Melanoma	Mutation						[Hodis et al., 2012]
BRD7	*BRD7*	Epithelial ovarian carcinoma	Decreased expression						[Park et al., 2014]
		Colorectal carcinoma	Decreased expression						[Wu et al., 2013]
		Nasopharyngeal carcinoma	Decreased expression						[Liu et al., 2008]

*Data from cell lines and xenografts or single case reports were not included in the table.

Light gray shading; heterozygous alteration, Solid black shading; heterozygous or homozygous alteration