

Table 1. Cardiac phenotypes associated with mutations in PcG or TrxG genes

	Complex	Enzymatic activity	Subunits (core & accessory) ^c	Cardiac phenotypes in mutant or transgenic mice	Additional information	References	
PcG ^a	PRC2	methyltransferase specific for H3K27 (Czermin <i>et al.</i> 2002, Muller <i>et al.</i> 2002)	Ezh1/2	<i>Ezh2^{NK}</i> : hypoplasia in complex myocardium, excessive trabeculation, spetal defects, dilation in RV. <i>Ezh2^{SHF}</i> : cardiac hypertrophy and fibrosis in RV during adulthood	Ezh2 methylates GATA4 and inhibits GATA4 activity	Delgado-Olguín <i>et al.</i> 2012, He <i>et al.</i> 2012a, He <i>et al.</i> 2012b, Jung <i>et al.</i> 2005, Kim <i>et al.</i> 2004, Lee <i>et al.</i> 2000, Mysliwiec <i>et al.</i> 2011, Nakajima <i>et al.</i> 2011, Shirato <i>et al.</i> 2009, Takeuchi <i>et al.</i> 1999, Toyoda <i>et al.</i> 2003	
			Eed	<i>Eed^{TnT}</i> : hypoplasia in complex myocardium, excessive trabeculation			
			Suz12	n/d			
			Jmj	<i>Jmj^{-/-}</i> : excessive trabeculation, DORV, VSD, ventricular wall noncompaction			
			Phc1/2/3	<i>Phc1^{-/-}</i> : looping defect, VSD, pulmonary stenosis, aortic stenosis, tetralogy of Fallot <i>β MHC-Pch1</i> : reduced contractility, sarcomere disorganization, cardiomyocyte apoptosis			n/a
	Cbx2/4/8	n/d					
	Bmi1	n/d					
	Ring1	n/d					
	Rnf2	n/d					
	PR-DUB	deubiquitinase specific for H2AK119 (Scheuermann <i>et al.</i> 2010)	Bap1	n/d	n/a	n/a	
Asxl1/2			n/d				
TrxG ^b	BAF/BRM	chromatin remodeling (Kwon <i>et al.</i> 1994, Phelan <i>et al.</i> 2000, Wang <i>et al.</i> 1996)	Brm/Brg1	<i>Tie2Cre;Brg1^{F/F}</i> : sparse trabeculation	<i>Brg1</i> genetically interacts with <i>Nkx2.5</i> , <i>Tbx5</i> , <i>GATA4</i> , <i>Tbx20</i> ;	Hang <i>et al.</i> 2010, Licket <i>et al.</i> 2004, Stankunas <i>et al.</i> 2008, Takeuchi and Bruneau 2009, Takeuchi <i>et al.</i> 2011	
				<i>Sm22a-cre;Brg1^{ff}</i> : thin compact myocardium and failure to form IVS, both due to proliferation defect; early differentiation of cardiomyocyte			
				<i>Brg1</i> inactivation in adult heart (<i>Tnnt2-rtTA;Tre-cre;Brg1^{ff}</i>): reduced hypertrophic response to TAC			
			BAF60a/b/c	<i>siBaf60c</i> : trabeculation defect, impaired development of SHF	BAF60c together with GATA4 induce cardiac fate in non-cardiogenic mesoderm; BAF60c enables GATA4 binding to target promoters; BAF60c potentiates GATA4, <i>Nkx2.5</i> , <i>Tbx5</i> activity in reporter assays by recruiting BAF		
				BAF250			n/d
				BAF170			n/d
	MLL	methyltransferase specific for H3K4 (Milne <i>et al.</i> 2002)	MLL1/2/3	Wdr5	n/d	Ash2l interacts with <i>Tbx1</i> in a cancer cell line and promotes <i>Tbx1</i> activity in luciferase assays	Stoller <i>et al.</i> 2010
				Ash2l	n/d		

^aDrosophila contains a fourth PcG complex, PhoRC, which is thought to recruit other PcG complexes to special regulatory elements known as Polycomb Response Elements (PRE). However, the mechanism of PcG recruitment in mammals appears to be distinct from that in Drosophila, and a mammalian complex that is functionally equivalent to PhoRC has not been identified.

^bA "supercomplex" containing components of BRM/BAF, MLL and several non-TrxG complexes has also been identified in mammalian cells (Nakamura et al. 2002).

^cSubunits that are essential for the complex's biochemical properties are considered core subunits. In some cases, a core component can be encoded by multiple homologs that are expressed in a cell type-specific manner. Subunits that are not essential for the complex's activity but are part of the complex under specific circumstances are considered accessory subunits.

n/d: not determined

n/a: not available