Supplementary Table S1. *AKAP9* and Long QT/ Romano-Ward Syndrome				
	Curator 1	Curator 2	Curator 3	References
Genetic Evidence				
# Probands (0-4)	1	1	1	(Chen, Marquardt et al. 2007)
# Publications (0-3)	1	1	1	
# Variants (0-4)	1	1	1	
Other Statistical Evidence (0-1)				
Experimental Evidence				
Gene Function (0-2)	2	2	2	(Chen, Marquardt et al. 2007, Mullally, Goldenberg et al. 2013, Brion, Blanco-Verea et al. 2014, Shigemizu, Aiba et al. 2015)
Gene Disruption (0-2)				
Animal Model (0-2)				
Calculated score	5	5	5	
Curator Classification	Limited	Limited	Limited	

Notes:
Mullally *et al*. 2013 reported a patient with c.2113T>G p.L705V detected in *AKAP9* from a screen of Long QT genes, but this alteration was detected possibly in combination with alterations in other genes, so omit from consideration.
Brion *et al.* 2014 reported a patient with c.A9943G p.T3315A and LQTS within a mixed cohort with no specific patient phenotype, so do not count.
Shigemezu *et al.* 2015 reported 2 patients with c.5341T>A p.S1781T and c.2295T>A p.D765E, but asymptomatic and QTc is within normal range, so do not count.

Supplementary Table S2. *ATF6* and Achromatopsia				
	Curator 1	Curator 2	Curator 3	References
Genetic Evidence				
# Probands (0-4)	4	4	4	(Ansar, Santos-Cortez et al. 2015, Kohl, Zobor et al. 2015, Xu, Gelowani et al. 2015)
# Publications (0-3)	3	3	3	
# Variants (0-4)	3	3	3	
Other Statistical Evidence (0-1)		1	1	(Kohl, Zobor et al. 2015)
Experimental Evidence				
Gene Function (0-2)	1	1	1	(Ansar, Santos-Cortez et al. 2015)
Gene Disruption (0-2)				
Animal Model (0-2)	2	2	2	(Nakanishi, Shimazawa et al. 2013, Kohl, Zobor et al. 2015)
Calculated score	13	14	14	
Curator Classification	Strong	Strong	Strong	

Supplementary Table S3. *C1QB* and Immunodeficiency due to Early C1q Deficiency				
	Curator 1	Curator 2	Curator 3	References
Genetic Evidence				
# Probands (0-4)	3	3	3	(McAdam, Goundis et al. 1988, Petry, Hauptmann et al. 1997, Marquart, Schejbel et al. 2007, Roumenina, Sene et al. 2011, Higuchi, Shimizu et al. 2013, Troedson, Wong et al. 2013, van Schaarenburg, Daha et al. 2015)
# Publications (0-3)	3	3	3	
# Variants (0-4)	3	3	3	
Other Statistical Evidence (0-1)				
Experimental Evidence				
Gene Function (0-2)	2	2	2	(Higuchi, Shimizu et al. 2013)
Gene Disruption (0-2)			1	(van Schaarenburg, Daha et al. 2015)
Animal Model (0-2)				(Miura-Shimura, Nakamura et al. 2002)
Other				
Calculated score	11	11	12	
Curator Classification	Moderate	Moderate	Moderate	

Notes:
The precise location of the genetic alteration in the Miura-Shimura mouse is not clear, so do not count.

Supplementary Table S4. *CD3E* and Severe Combined Immunodeficiency				
	Curator 1	Curator 2	Curator 3	References
Genetic Evidence				
# Probands (0-4)	2	2	2	(Soudais, de Villartay et al. 1993, de Saint Basile, Geissmann et al. 2004, Fuehrer, Pannicke et al. 2014)
# Publications (0-3)	3	4	3	
# Variants (0-4)	3	3	4	
Other Statistical Evidence (0-1)				
Experimental Evidence				
Gene Function (0-2)	2	2	2	(DeJarnette, Sommers et al. 1998)
Gene Disruption (0-2)	2	1	1	(Renard, Ardouin et al. 1995, DeJarnette, Sommers et al. 1998)
Animal Model (0-2)	1	2	2	(DeJarnette, Sommers et al. 1998)
Calculated score	13	14	14	
Curator Classification	Strong	Strong	Strong	

Supplementary Table S5. *CHD1L* and Renal or Urinary Tract malformation (CAKUT)				
	Curator 1	Curator 2	Curator 3	References
Genetic Evidence				
# Probands (0-4)	2	2	1	(Brockschmidt, Chung et al. 2012, Hwang, Dworschak et al. 2014, Nicolaou, Pulit et al. 2016)
# Publications (0-3)	2	2	1	
# Variants (0-4)				
Other Statistical Evidence (0-1)				
Experimental Evidence				
Gene Function (0-2)	1	1		(Ahel, Horejsi et al. 2009)
Gene Disruption (0-2)				
Animal Model (0-2)				
Calculated score	5	5	4	
Curator Classification	Limited	Limited	Limited	

Notes:
Hwang et al. 2014 reported 5 families with CAKUT and five separate missense alterations in CDH1L; only 2 of the families had rare enough variants to be considered: c.998C>G p.P333R is predicted probably damaging/deleterious and not found in ExAC and c.1199A>G p.E400G is predicted probably damaging/deleterious and found in 2 hets in ExAC: VUS.
Brockschmidt et al. 2012: reported three patients with het alterations in CDH1L plus functional studies; transfected HEK293T cells with c-myc-tagged wild-type and mutant CHD1L constructs showed interaction between PARP1 and WT CHD1L protein but not mutant CHD1L protein. All three alterations are too common in controls (ExAC) to cause disease, omit entire paper from consideration.
Nicolaou et al. 2016 was a study representing the largest set of genes analyzed in CAKUT patients to date, the contribution of previously implicated genes to CAKUT risk was significantly smaller than expected, and the disease may be more complex than previously assumed. Only c.1291_1293delACA from Nicolaou (2015) is rare enough to cause disease and predicted pathogenic. The authors report 2 patients with c.1386-2A>G, but one also had a variant in UMOD, plus this variant is found in 109 hets in ExAC, so omit from consideration.

Supplementary Table S6. *COL2A1* and Spondyloepiphyseal Dysplasia, Stanescu Type				
	Curator 1	Curator 2	Curator 3	References
Genetic Evidence				
# Probands (0-4)	2	2	2	(Rukavina, Mortier et al. 2014, Jurgens, Sobreira et al. 2015, Hammarsjo, Nordgren et al. 2016)
# Publications (0-3)	3	3	3	
# Variants (0-4)	3	3	3	
Other Statistical Evidence (0-1)				
Experimental Evidence				
Gene Function (0-2)	2	1	1	(Jensen, Steplewski et al. 2011, Arita, Fertala et al. 2015)
Gene Disruption (0-2)		1	1	(Jensen, Steplewski et al. 2011)
Animal Model (0-2)	1	2	2	(Li, Prockop et al. 1995, Arita, Fertala et al. 2015)
Calculated score	11	12	12	
Curator Classification	Moderate	Moderate	Moderate	

Supplementary Table S7. *NGLY1* and Congenital Disorder of Deglycosylation				
	Curator 1	Curator 2	Curator 3	References
Genetic Evidence				
# Probands (0-4)	3	4	4	(Need, Shashi et al. 2012, Enns, Shashi et al. 2014, Caglayan, Comu et al. 2015, He, Grotzke et al. 2015, Heeley and Shinawi 2015, Karaca, Harel et al. 2015, Bosch, Boonstra et al. 2016)
# Publications (0-3)	4	3	4	
# Variants (0-4)	4	4	3	
Other Statistical Evidence (0-1)				
Experimental Evidence				
Gene Function (0-2)	1	1	1	(Zhou, Zhao et al. 2006, Enns, Shashi et al. 2014)
Gene Disruption (0-2)	2	2	1	(He, Grotzke et al. 2015, Huang, Harada et al. 2015)
Animal Model (0-2)	1	1		(Habibi-Babadi, Su et al. 2010)
Calculated score	15	15	13	
Curator Classification	Strong	Strong	Strong	

Supplementary Table S8. *NHP2* and Dyskeratosis Congenita				
	Curator 1	Curator 2	Curator 3	References
Genetic Evidence				
# Probands (0-4)	1	1	1	(Vulliamy, Beswick et al. 2008, Trahan, Martel et al. 2010)
# Publications (0-3)	1	1	1	
# Variants (0-4)	3	3	2	
Other Statistical Evidence (0-1)				
Experimental Evidence				
Gene Function (0-2)	2	2	2	(Pogacic, Dragon et al. 2000)
Gene Disruption (0-2)	1	2	2	(Vulliamy, Beswick et al. 2008)
Animal Model (0-2)				
Calculated score	8	9	8	
Curator Classification	Moderate	Moderate	Moderate	

Supplementary Table S9. *RAD51C* and Fanconi Anemia				
	Curator 1	Curator 2	Curator 3	References
Genetic Evidence				
# Probands (0-4)	1	1	1	(Vaz, Hanenberg et al. 2010, Somyajit, Subramanya et al. 2012)
# Publications (0-3)	1	1	1	
# Variants (0-4)	1	1	1	
Other Statistical Evidence (0-1)				
Experimental Evidence				
Gene Function (0-2)	2	2	2	(Somyajit, Subramanya et al. 2012)
Gene Disruption (0-2)	2	2	2	(Vaz, Hanenberg et al. 2010, Rodrigue, Coulombe et al. 2013)
Animal Model (0-2)	1	1	2	(Kuznetsov, Haines et al. 2009, Smeenk, de Groot et al. 2010)
Calculated score	8	8	9	
Curator Classification	Moderate	Moderate	Moderate	

Supplementary Table S10. *RPS10* and Diamond-Blackfan Anemia				
	Curator 1	Curator 2	Curator 3	References
Genetic Evidence				
# Probands (0-4)	3	3	3	(Doherty, Sheen et al. 2010, Yazaki, Kamei et al. 2012, Smetanina, Mersiyanova et al. 2015)
# Publications (0-3)	3	3	3	
# Variants (0-4)	4	4	4	
Other Statistical Evidence (0-1)				
Experimental Evidence				
Gene Function (0-2)	2	2	2	(Doherty, Sheen et al. 2010)
Gene Disruption (0-2)	1	1	1	(Doherty, Sheen et al. 2010)
Animal Model (0-2)				
Calculated score	13	13	13	
Curator Classification	Strong	Strong	Strong	

Supplementary Table S11. *RPS24* and Diamond-Blackfan Anemia				
	Curator 1	Curator 2	Curator 3	References
Genetic Evidence				
# Probands (0-4)	3	3	3	(Gazda, Grabowska et al. 2006, Badhai, Frojmark et al. 2009, Boria, Garelli et al. 2010, Quarello, Garelli et al. 2010, Ghemlas, Li et al. 2015, Smetanina, Mersiyanova et al. 2015)
# Publications (0-3)	3	3	3	
# Variants (0-4)	4	4	4	
Other Statistical Evidence (0-1)				
Experimental Evidence				
Gene Function (0-2)	2	2	2	(Choesmel, Fribourg et al. 2008)
Gene Disruption (0-2)	1	1	1	(Choesmel, Fribourg et al. 2008, Badhai, Frojmark et al. 2009)
Animal Model (0-2)				
Calculated score	13	13	13	
Curator Classification	Strong	Strong	Strong	

Supplementary Table S12. *SCN4B* and Long QT Syndrome				
	Curator 1	Curator 2	Curator 3	References
Genetic Evidence				
# Probands (0-4)	1	1	1	(Medeiros-Domingo, Kaku et al. 2007, Li, Wang et al. 2013)
# Publications (0-3)	3	2	2	
# Variants (0-4)			1	
Other Statistical Evidence (0-1)				
Experimental Evidence				
Gene Function (0-2)	1	2	1	(Olesen, Jespersen et al. 2011)
Gene Disruption (0-2)		2		(Medeiros-Domingo, Kaku et al. 2007)
Animal Model (0-2)				
Calculated score	5	7	5	
Curator Classification	Limited	Limited	Limited	

Notes:
Mullally (Mullally, Goldenberg et al. 2013) reported c.658G>A p.A220T in a patient with multiple mutations in long-QT associated genes, so omit from consideration.
Lieve (Lieve, Williams et al. 2013) reported an additional patient with c.658G>A p.A220T, but it's not clear if mutations in multiple genes were identified in this patient, so do not count.

Supplementary Table S13. *SOS2* and Noonan Syndrome				
	Curator 1	Curator 2	Curator 3	References
Genetic Evidence				
# Probands (0-4)	3	3	3	(Cordeddu, Yin et al. 2015, Yamamoto, Aguena et al. 2015)
# Publications (0-3)	2	2	2	
# Variants (0-4)	3	3	3	
Other Statistical Evidence (0-1)				
Experimental Evidence				
Gene Function (0-2)	2	2	2	(Yamamoto, Aguena et al. 2015)
Gene Disruption (0-2)	1		1	(Cordeddu, Yin et al. 2015)
Animal Model (0-2)				
Calculated score	11	10	11	
Curator Classification	Moderate	Moderate	Moderate	

Supplementary Table S14. *WRAP53* and Dyskeratosis Congenita				
	Curator 1	Curator 2	Curator 3	References
Genetic Evidence				
# Probands (0-4)	1	1	1	(Batista, Pech et al. 2011, Zhong, Savage et al. 2011)
# Publications (0-3)	1	1	1	
# Variants (0-4)	2	4	4	
Other Statistical Evidence (0-1)				
Experimental Evidence				
Gene Function (0-2)	2	2	2	(Venteicher, Abreu et al. 2009)
Gene Disruption (0-2)	1	1	1	(Batista, Pech et al. 2011)
Animal Model (0-2)				
Calculated score	7	9	9	
Curator Classification	Limited	Moderate	Moderate	

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