SUPPLIMENTARY TABLE S1. Reported cases of genetically diagnosed WS and related diseases (TS, PCWH/PCH, and YDHS)

Affected gene	Phenotype subtype	No. of families[a]	No. of individuals	Supplimentary references
AB genes	Total	262[b]	737	
	WS1	155[c]	484	(Arias, *et al.*, 1997; Baldwin, *et al.*, 1993; Baldwin, *et al.*, 1994; Baldwin, *et al.*, 1995; Carey , *et al.*, 1998; Chen, *et al.*, 2010; Dronziewicka and Hass, 2014; Eigelshoven, *et al.*, 2009; Horan, *et al.*, 2013; Hul, *et al.*, 1995; Hul, *et al.*, 1998; Hieh, *et al.*, 1993; Jalilian, *et al.*, 2015a; Jalilian, *et al.*, 2015b; Jang, *et al.*, 2015; Kashima, *et al.*, 2011; Kim, *et al.*, 2015; Kozawa, *et al.*, 2009; Kojat, *et al.*, 2007; Lalwani, *et al.*, 1996; Li, *et al.*, 2015; Marasuaga, *et al.*, 2013; Merchant, *et al.*, 2001; Morell, *et al.*, 1992; Morell, *et al.*, 1993; Morell, *et al.*, 1997; Mousry, *et al.*, 2015; Nakamura, *et al.*, 2009; Pandya, *et al.*, 1996; Qin, *et al.*, 2006; Shin, *et al.*, 2004; Sieljma, *et al.*, 1997; Sezirova, *et al.*, 2000; Tasdemir, *et al.*, 2015; Tassabehji, *et al.*, 1992; Tassabehji, *et al.*, 1993; Tassabehji, *et al.*, 1994a; Tassabehji, *et al.*, 1995; Wang, *et al.*, 2010; Wildharth, *et al.*, 1996; Wildharth, *et al.*, 2013; Wollnik, *et al.*, 2003; Wu, *et al.*, 1993; Wu, *et al.*, 2016; Xiao, *et al.*, 2016; Yang, *et al.*, 2007; Yang, *et al.*, 2013b; Yoshida, *et al.*, 2016; Zieogava, *et al.*, 1995)
	WS2	67[c]	160[d]	(Bondurand, *et al.*, 2007; Bonnet, *et al.*, 2011; Chaoui, *et al.*, 2011; Chen, *et al.*, 2010; Chen, *et al.*, 2014; Chen, *et al.*, 2015; Chen, *et al.*, 2016; Chiang, *et al.*, 2009; Elsaidi-Berges, *et al.*, 2013; Haddad, *et al.*, 2011; Izumi, *et al.*, 2015; Jang, *et al.*, 2011; Kim *et al.*, 2015; Lalwani, *et al.*, 1998; Lautenschlager, *et al.*, 1996; Lupe, *et al.*, 2012; Ma, *et al.*, 2016; Morena, *et al.*, 2008; Nishokuni, *et al.*, 1996; Okamura, *et al.*, 2013; Pingault, *et al.*, 2002; Pingault, *et al.*, 2014; Sanchez-Martin, *et al.*, 2002; Schwarzbraun, *et al.*, 2007; Shi, *et al.*, 2016; Shigemura, *et al.*, 2010; Stanton, *et al.*, 2012; Tassabehji, *et al.*, 1994b; Tassabehji, *et al.*, 1995; Vilais, *et a.*l., 2011; Wang, *et al.*, 1998; Wenchi, *et al.*, 2015; Wildharth, *et al.*, 2013; Wu, *et al.*, 2016; Yao, *et al.*, 2011; Yang, *et al.*, 2013a; Yang, *et al.*, 2013b; Enzo Sera, *et al.*, 2015)
	WS3	11	17	(Fujimoto, *et al.*, 1998; Hieh, *et al.*, 1993; Mousry, *et al.*, 2015; Nya, *et al.*, 1998; Pastorio, *et al.*, 1993; Sommer and Bartholomew, 2003; Tassabehji, *et al.*, 1995; Tekin, *et al.*, 2001; Wollnik, *et al.*, 2003; Zieogava, *et al.*, 1995)
	WS4	18	86	(Arias, *et al.*, 1995; Boardman, *et al.*, 2001; Bondurand, *et al.*, 2007; Bondurand, *et al.*, 2012; Chaoui, *et al.*, 2011; Dodraj, *et al.*, 2015; Edery , *et al.*, 1996; Elsaidi-Berges, *et al.*, 2013; Fernandez, *et al.*, 2014; Haddad, *et al.*, 2011; Hofstra, *et al.*, 1996; Inoue, *et al.*, 1999; Inoue, *et al.*, 2002; Jang, *et al.*, 2011; Jang, *et al.*, 2013; Kapoor, *et al.*, 2012; Loupe, *et al.*, 2014; Morin, *et al.*, 2008; Pingault, *et al.*, 1998; Pingault, *et al.*, 2001; Pingault, *et al.*, 2002; Pingault, *et al.*, 2014; Sanchez-Mejias, *et al.*, 2010; Sanghokar, *et al.*, 2005; Sham, *et al.*, 2001; Shimozaki, *et al.*, 2007; Southard-Smith, *et al.*, 1999; Syrris, *et al.*, 1999; Scotpe, *et al.*, 2000; Tuka, *et al.*, 2003; Tuoratie, *et al.*, 2000; Verbeij, *et al.*, 2006; Vinuela, *et al.*, 2009; Wang, *et al.*, 2014)
	Atypical WS	1	1	(Jelena, *et al.*, 2014)
	TS	4	20	(Arnold, *et al.*, 1998; Schwarzbraun, *et al.*, 2007; Shigemura, *et al.*, 2010; Smith, *et al.*, 2000)
	PCWH/PCWH	20	23	(Bondurand, *et al.*, 2007; Chaoui, *et al.*, 2011; Elsaidi-Berges, *et al.*, 2013; Inoue, *et al.*, 2004; Okimoto, *et al.*, 2012; Parthey , *et al.*, 2012; Pingault, *et al.*, 2000; Uzuzikee, *et al.*, 2011)
	YDHS	1	1	(Bondurand, *et al.*, 1999)
PAX3	Total	110	429	
	WS1	98[e]	402[d]	(Arias, *et al.*, 1997; Baldwin, *et al.*, 1992; Baldwin, *et al.*, 1994; Baldwin, *et al.*, 1995; Chen, *et al.*, 2010; Dronziewicka and Hass, 2014; Eigelshoven, *et al.*, 2009; Horan, *et al.*, 2013; Hul, *et al.*, 1995; Hul, *et al.*, 1998; Hieh, *et al.*, 1993; Jalilian, *et al.*, 2015a; Jalilian, *et al.*, 2015b; Jang, *et al.*, 2015; Kashima, *et al.*, 2011; Kim, *et al.*, 2015; Kozawa, *et al.*, 2009; Kojat, *et al.*, 2007; Lalwani, *et al.*, 1996; Li, *et al.*, 2015; Marasuaga, *et al.*, 2013; Merchant, *et al.*, 2001; Morell, *et al.*, 1992; Morell, *et al.*, 1993; Mousry, *et al.*, 2015; Nakamura, *et al.*, 2009; Pandya, *et al.*, 1996; Qin, *et al.*, 2006; Shin, *et al.*, 2004; Sieljma, *et al.*, 1997; Sezirova, *et al.*, 2000; Tasdemir, *et al.*, 2015; Tassabehji, *et al.*, 1992; Tassabehji, *et al.*, 1993; Tassabehji, *et al.*, 1994b; Tassabehji, *et al.*, 1995; Wang, *et al.*, 2010; Wildharth, *et al.*, 1996; Wildharth, *et al.*, 2013; Wollnik, *et al.*, 2003; Wu, *et al.*, 1993; Wu, *et al.*, 2016; Xiao, *et al.*, 2016; Yang, *et al.*, 2007; Yang, *et al.*, 2013b; Yoshida, *et al.*,
	WS2	2	10	(Chen, *et al.*, 2010; Wang, *et al.*, 1998)
	WS3	10	16	(Hieh, *et al.*, 1993; Mousry, *et al.*, 2015; Nya, *et al.*, 1998; Pastorio, *et al.*, 1993; Sommer and Bartholomew, 2003; Tassabehji, *et al.*, 1995; Tekin, *et al.*, 2001; Wollnik, *et al.*, 2003; Zieogava, *et al.*, 1995)
SOX10	Total	85[f]	168[d]	
	WS2	22	31[c]	(Actmess, *et al.*, 2014; Bondurand, *et al.*, 2007; Chen, *et al.*, 2010; Chen, *et al.*, 2014; Chen, *et al.*, 2015; Elsaidi-Berges, *et al.*, 2013; Izumi, *et al.*, 2015; Jang, *et al.*, 2011; Ma, *et al.*, 2016; Okamura, *et al.*, 2013; Pingault, *et al.*, 2002; Pingault, *et al.*, 2014; Stanton, *et al.*, 2012; Wenchi, *et al.*, 2015)
	WS4	40	44	(Bondurand, *et al.*, 1999; Bondurand, *et al.*, 2007; Bondurand, *et al.*, 2012; Chaoui, *et al.*, 2011; Elsaidi-Berges, *et al.*, 2013; Fernandez, *et al.*, 2014; Inoue, *et al.*, 1999; Inoue, *et al.*, 2002; Jang, *et al.*, 2011; Jang, *et al.*, 2013; Morin, *et al.*, 2008; Pingault, *et al.*, 1998; Pingault, *et al.*, 2002; Pingault, *et al.*, 2014; Sanchez-Mejias, *et al.*, 2010; Sham, *et al.*, 2001; Shimozaki, *et al.*, 2007; Southard-Smith, *et al.*, 1999; Scotpe, *et al.*, 2000; Tuka, *et al.*, 2003; Tuoratie, *et al.*, 2000; Verbeij, *et al.*, 2006; Vinuela, *et al.*, 2009; Wang, *et al.*, 2014)
	Atypical WS	1	1	(Jelena, *et al.*, 2014)
	PCW or PCWH	20	23	(Bondurand, *et al.*, 2007; Chaoui, *et al.*, 2011; Elsaidi-Berges, *et al.*, 2013; Inoue, *et al.*, 2004; Okimoto, *et al.*, 2012; Parthey , *et al.*, 2012; Pingault, *et al.*, 2000; Uzuzikee, *et al.*, 2011)
	YDHS	1	1	(Bondurand, *et al.*, 1999)
MITF	Total	42	156	
	WS2	5	19	(Yang, *et al.*, 2013b)
	WS2	37	135	(Arnold, *et al.*, 1998; Bonnet, *et al.*, 2011; Chen, *et al.*, 2010; Chen, *et al.*, 2016; Chiang, *et al.*, 2009; Haddad, *et al.*, 2011; Kim, *et al.*, 2015; Lalwani, *et al.*, 1998; Lautenschlager, *et al.*, 1996; Lupe, *et al.*, 2012; Morena, *et al.*, 2008; Nishokuni, *et al.*, 1996; Schwarzbraun, *et al.*, 2007; Shi, *et al.*, 2016; Shigemura, *et al.*, 2010; Smith, *et al.*, 2000; Tassabehji, *et al.*, 1994b; Tassabehji, *et al.*, 1995; Wildharth, *et al.*, 2013; Wu, *et al.*, 2016; Yao, *et al.*, 2011; Yang, *et al.*, 2013a; Yang, *et al.*, 2013b)
	TS	4	20	(Arnold, *et al.*, 1998; Schwarzbraun, *et al.*, 2007; Shigemura, *et al.*, 2010; Smith, *et al.*, 2000)
EDNRB	Total	12[g]	29[d]	
	WS4	12	29	(Arias, *et al.*, 1995; Boardman, *et al.*, 2001; Dodraj, *et al.*, 2015; Haddad, *et al.*, 2011; Loupe, *et al.*, 2014; Pingault, *et al.*, 2002; Sanghokar, *et al.*, 2005; Syrris, *et al.*, 1999; Tuyroz, *et al.*, 2009; Verbeij, *et al.*, 2002)
EDN3	Total	8	13	
	WS1	1	1	(Wu, *et al.*, 2016)
	WS4	7	12	(Edery, *et al.*, 1996; Hofstra, *et al.*, 1996; Kapoor, *et al.*, 2012; Pingault, *et al.*, 2001; Pingault, *et al.*, 2002; Shamseldin, *et al.*, 2010; Vinuela, *et al.*, 2009)
KIT	Total	2	7	
	WS2	1	6	(Inoue, *et al.*, 2002)
	WS3	1	1	(Fujimoto, *et al.*, 1998)
SNAI2	Total	2	2	
	WS2	2	2	(Sanchez-Martin, *et al.*, 2002)
KITLG	Total	2[h]	2[i]	
	WS2	2[h]	2[i]	(Enzo Sera, *et al.*, 2015)

WS, Waardenburg syndrome; TS, Tietz syndrome; PCWH/PCH, peripheral demyelinating neuropathy , central dysmyelination, Waardenburg syndrome, and Hirschsprung disease; YDHS, Yemenite deaf-blind hypopigmentation syndrome

[a]Families and individuals with established genetic diagnosis and reported in English literatures. Reports without clinical description of the patients were excluded from the study.

[b]One or more families manifested multiple subtypes.

[c]Two cases could not be definitely differentiated from WS4.

[d]Includes the present case.

[e]One case could not be definitely differentiated from WS3 by the description

[f]Two subtypes ocurred in one family.

[g]Two cases showed intestinal obstruction but no aganglionosis.

[h]A cosegring PAX3 mutation was found in this case.

[i]In one family, three members has no description of intestinal symptoms, thus there is a possibility that they may had WS2 instead of WS4.

[j]Includes the present case.