Supplementary Table 1. Clinical features of the current and other patients with Weiss-Kruszka syndrome

No. [PMID]	Sex	Age	Variant	ACMG classification	Inheritance	Height at last exam (percentile)	DD	ASD	Craniofacial features						Ears/hearing	Feeding issues	CHD	Limb anomalies	Craniosynostosis /Metopic ridging	Brain abnormalities
									Ptosis	Downslanted palpebral fissures	Cupid's Bow	Arched eyebrows	Epicanthal folds	Short upturned nose						
P1 [31361404]	M	1 yr 4 mo	NM_021224.6:exon3:c.2590C>T (p.Arg864*)	LP	Maternal#	NA	Motor/speech	_	+	_	+	+	+	_	Low-set	+	NA	Fifth finger clinodactyly	_	_
P2 [31361404]	M	10 yr	NM_021224.6:exon3:c.2542del (p.Cys848Valfs*66)	P	*De novo*	Normal	Motor/speech	+	+	+	+	+	+	+	_	+	NA	NA	_	_
P3 [31361404]	M	6 yr	NM_021224.6:exon3:c.831_834del (p.Arg277Serfs*26)	P	*De novo*	NA	Motor/speech	_	_	_	_	_	_	+	Inner ear malformation	+	Bicuspid aortic valve; VSD	NA	_	_
P4 [31361404]	M	2 yr 7 mo	NM_021224.6:exon6:c.6214_6215del(p.His2072Tyrfs*8)	P	*De novo*	-2.33 SD	Speech	_	+	_	+	_	+	_	Small/low-set	+	NA	NA	+	NA
P5 [31361404]	F	14 yr	NM_021224.6:exon3:c.763C>T (p.Arg255*)	LP	Paternal#	NA	IEP/special education	_	+	_	_	+	+	_	Hearing loss	_	NA	NA	+	NA
P6 [31361404]	F	7 mo	NM_021224.6:exon11:c.7057-2A>G	VUS	*De novo*	<1st percentile	Early intervention for DD	_	+	+	+	+	+	+	Horizontal crus helix	+	VSD	Prominent creases on hands and feet	_	+
P7 [31361405]	M	13 yr	NM_021224.6:exon9:c.6794dup (p.Tyr2265*)	P	*De novo*	NA	Cognitive impairment	+	_	_	+	+	_	_	Prominent ears/ear pits/ hearing loss	_	NA	NA	+	NA
P8 [31361406]	M	2 yr	NM_021224.6:exon3:c.882dup (p.Ser295Glnfs*64)	P	*De novo*	NA	Speech	_	+	+	_	_	_	_	_	_	NA	NA	_	+
P9 [31361404]	M	15 yr	NM_021224.6:exon3:c.4165C>T (p.Gln1389*)	P	*De novo*	75th percentile	Global	_	+	+	+	+	_	_	Low-set	+	NA	NA	_	
P10 [31361404]	M	8 yr	NM_021224.6:exon3:c.1234_1235insAA (p.Ser412*)	LP	Unknown	3rd percentile	Motor/Speech; IEP	_	+	_	_	_	NA	_	Mildly cupped ears	+	NA	NA	_	_
P11 [31361404]	F	2 yr 5 mo	NM_021224.6:exon6:c.6214_6215del(p.His2072Tyrfs*8)	P	*De novo*	NA	_	_	+	_	_	_	NA	_	NA	+	NA	NA	_	
P12 [31361404]	M	9 mo	NM_021224.6:exon3:c.2049dup (p.Pro684Serfs*14)	P	*De novo*	NA	Motor	_	+	+	+	+	+	+	_	_	NA	NA	_	_
P13 [31361404]	M	8 yr 7 mo	NM_021224.6:exon8:c.6631del (p.Arg2211Glyfs*59)	P	*De novo*	5 yr 25–50%ile	_	_	+	+	+	_	_	_	_	_	NA	Fifth finger clinodactyly	_	
P14 [31361404]	F	8 yr	NM_021224.6:exon3:c.2695G>T (p.Glu899*)	LP	Mother negative Father unknown	NA	Cognitive impairment	_	_	_	_	+	_	+	_	+	NA	NA	_	_
P15 [28513610]	F	2 yr	NM_021224.6:exon3:c.3787C>T (p.Arg1263*)	P	Paternal&	75th percentile	_	_	+	+	+	+	_	+	_	NA	NA	NA	+	+
P16 28513610]	F	4 yr	NM_021224.6:exon3:c.3787C>T (p.Arg1263*)	P	Paternal&	50th percentile	_	_	+	+	+	_	+	_	_	NA	NA	NA	+	_
P17 [28513610]	M	34 yr	NM_021224.6:exon3:c.3787C>T (p.Arg1263*)	P	Maternal*	10th percentile	_	_	+	_	_	_	_	_	_	NA	NA	NA	+	
P18 [2,3]	M	2 yr	NM_021224.6:exon3:c.2979_2980delinsA (p.Val994Trpfs*147)	P	*De novo*	27th percentile	Speech	+	+	+	_	+	+	+	Left overfolded ear	+	NA	Single palmar crease; fifth finger clinodactyly	+	_
P19 [31361404]	M	1 yr 7 mo	NM_021224.6:exon3:c.4263del (p.Glu1422Serfs*6)	P	*De novo*	34th percentile	Motor/speech	+	+	_	_	_	+	+	Low-set	_	D-TGA	NA	+	Ventriculomegaly
P20 [2,3]	F	5 yr	Chr9:g.(108940763-110561397)del(hg19)	P	*De novo*	97th percentile	_	_	+	+	+	+	+	+	NA	NA	NA	NA	_	+
P21 [2,3]	F	15 yr	Chr9:g.(108464368-110362345)del(hg19)	P	*De novo*	NA	Motor/Intellectual	+	_	+	+	_	_	_	NA	NA	VSD	NA	_	
P22 [28513610]	M	9 yr	NM_021224.6:exon3:c.5145del (p.Tyr1716Thrfs*28)	P	*De novo*	25th percentile	Motor/speech	+	+	_	_	_	_	_	_	NA	NA	NA	_	_
P23 [4]	F	5 yr	t(2;9) (P24;q32) involving *ZNF462*, *ASXL2*	P	*De novo*	5th percentile	Intellectual disability	+	+	+	+	+	_	+	Low-set /hearing loss	+	VSD: left ventricular hypertrophy	Single palmar crease; hypoplastic fingernails	_	+
P24 [5]	M	24 yr	t(9;13) (q31.2;q22.1) involving *ZNF462*, *KLF12*	P	*De novo*	3rd percentile	Intellectual disability	+	+	+	_	_	+	_	Low-set	+	NA	Small hands and feet; proximally placed thumbs	+	+
P25 [35198003]	F	3 yr 4 mo	NM_021224.6:exon3:c.3306dup (p.Gln1103Thrfs*10)	P	*De novo*	4th percentile	Global	+	+	_	_	_	+	_	NA	+	Ventricular hypertrophy	_	_	NA
P26 [35198003]	M	16 yr 9 mo	NM_021224.6:exon3:c.4185del (p.Met1396*)	LP	Mother negative Father unknown	<3rd percentile	Global	_	+	+	+	+	+	+	_	_	_	_	+	+
P27 [8]	M	17 yr 7 mo	Chr9:g(108331353-110707332)del(hg19)	P	*De novo*	NA	Mild DD	+	+	_	_	_	_	+	Low-set/Hearing loss	NA	Muscular VSD	_	_	+
P28 [35198003]	M	8 mo	NM_021224.6:exon8:c.6431C>A (p.Ser2144*)	P	Paternal§	NA	DD	_	+	_	_	_	_	_	Hearing loss	_	PDA	_	_	_
P29 [35198003]	M	31 yr	NM_021224.6:exon8:c.6431C>A (p.Ser2144*)	P	*De novo*	NA	_	_	+	_	_	_	_	_	Hearing loss	_	_	_	_	NA
P30 [10]	F	4 yr	Chr9:g.(108399883-113591075)del(hg19)	P	*De novo*	0-1 SD	Motor	NA	+	+	+	+	_	_	Ears/ hearing	_	_	_	_	_
P31	M	9 yr 9 mo	NM_021224.6:exon10:c.6833-2A>T (Splicing)	LP	*De novo*	<3rd percentile	Global	_	+	_	_	+	+	_	Low-set	+	NA	Single palmar crease	_	+
Overall							24/31	10/31	27/31	15/31	15/31	15/31	14/31	12/31	17/31	14/31	8/31	7/31	10/31	9/31

M: male, F: female, P: pathogenic, LP: likely pathogenic, VUS: variant of uncertain significance, DD: developmental delay, ASD: autism spectrum disorder, CHD: congenital heart disease, NA: data not available, IEP: individualized education program, VSD: ventricular septal defect, PDA: patent ductus arteriosus, D-TGA: D-transposition of the great arteries, #: phenotypic information of his mother/father was not described in the literature, &: the father had a pointed forehead and mild left ptosis, *: the mother had bilateral ptosis, $X this patient was the child of patient 29