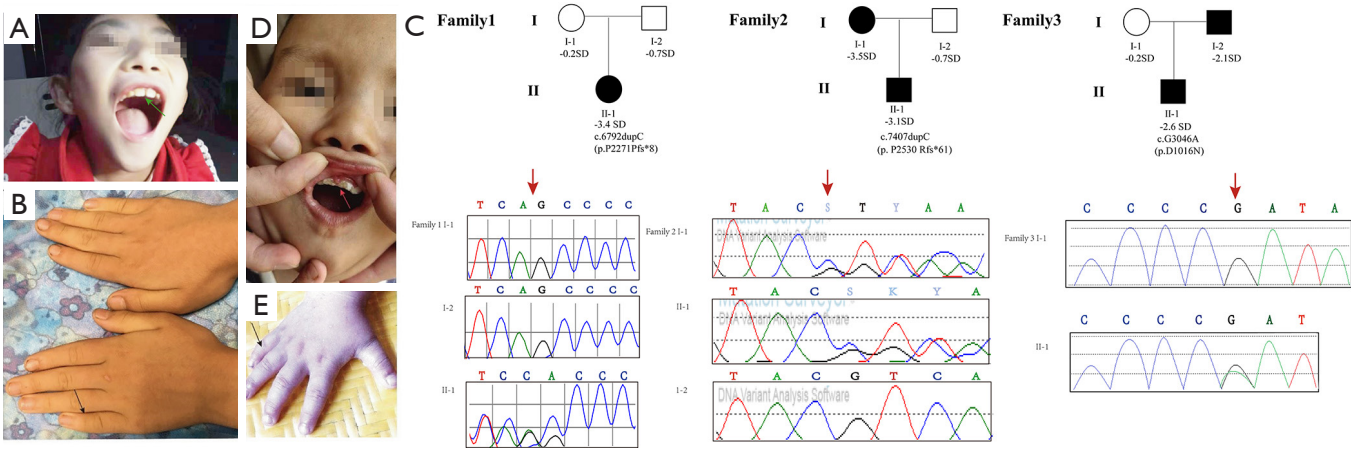

Figure S1 Pictures and molecular analysis of *ANKRD11* gene by Sanger sequencing. (A,B) Pictures of patient 1: (A) the green arrow showed the macrodontia; (B) the black arrow showed brachydactyly. (C) Molecular analysis of *ANKRD11* gene showed a heterozygous frameshift mutation c.6792dupC p.P2271pfs*8 for patient 1, c.7407dupC (p.P2530Rfs*61) for patient 2 and a missense mutation c.C3046A p.D2016N for patient 3. The testing of patient 1's parent showed that c.6792dupC p.P2271pfs*8 was *de novo*. The mutation [c.7407dupC (p.P2530Rfs*61)] of patient 2 was inherited from his mother. While the DNA of patient 3's father was not available, his mother did not carry the c.C3046A p.D1016N mutation. (D) The primary teeth of patient 2 at age 2 years. The arrows indicate the enamel hypoplasia and wide upper central incisors. (E) The right hand of patient 2. The arrow indicates mild fifth finger clinodactyly.

Table S1 Frequent *ANKRD11* mutations

Genetic mutation	Amino acid change	Number of patients	Number of families
c.1903_1907delAAACA	p.Lys635Glnfs*26	21	16
c.1381_1384delGAAA	p.Glu461Glnfs*48	4	3
c.3224_3227delCTTT	p.Glu1075Glyfs*242	2	2
c.3045delC	p.Asp1016Ilefs*302	2	2
c.1801C>T	p.Arg601*	2	2
Deletions between c.2395 and c.2412			
c.2395_2398delAAAG	p.Glu800Asnfs*62	3	2
c.2398_2401delGAAA	p.Glu800Asnfs*62	2	1
c.2408_2412delAAAAA	p.Lys803Argfs*5	4	4

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Table S2 Position of mutation in *ANKRD11* and short stature in patients with KBG syndrome

Intellectual disability	*ANKRD11* deletion (n=33)	Before RD1 (n=16)	No AD and RD2 (n=92)	No RD2 or D-box (n=19)	Missense (n=4)	P value for independence	P value for trend
None, n [%]	12 [36]	7 [44]	43 [47]	10 [53]	2 [50]	0.2	0.4
Short, n [%]	10 [30]	5 [31]	22 [24]	2 [11]	0	0.4	0.1
Severe, n [%]	3 [9]	3 [19]	9 [10]	2 [11]	0	0.8	0.7
Unknown, n [%]	8 [6]	1 [6]	15 [8]	5 [26]	2 [50]	0.2	0.7

P values were determined using the Fisher's exact test. Internal notes: Deletion of *ANKRD11* = 16q24.3 mutation causing deletion of all ANKRD codon or the start codon; Before RD1 = "X_ANK", "X_ANK-RD1", "X_RD1"; No AD and RD2 = "X_RD1-AD", "X_AD"; No RD2 or D-box = "X_AD-RD2", "X_RD2", "S_Dbox2", "S_Dbox1/2", "S_Dbox3"; Missense = "S_before ANK", "S_RD1-AD", "S_AD". RD1, repression domain-1; AD, activation domain; RD2, repression domain-2.

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