SUPPLEMENTARY INFORMATION

Further Delineation of the KAT6B Molecular and Phenotypic Spectrum.

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Molecular analysis

Primers were designed using the software design program, Primer3 (version.0.4.0) (http://frodo.wi.mit.edu/primer3/) and synthesized externally (Invitrogen, Life Technologies).

Supplementary Table 1 Primers used for molecular analysis of *KAT6B*

Exon	F name	F Seq 5' 3' direction	R name	R Seq 5' 3' direction	Product size (bp)	Annealing Temp (°C)
3_1	MYST4exon3_1_Fwd	TGCTTAATACAGTCTGGAATACTCTG	MYST4exon3_1_Rev	TCGGCTCCTCAAGTCCTTC	493	57.5
3_2	MYST4exon3_2_Fwd	TTTCCTAAGTCAGCCAAGGG	MYST4exon3_2_Rev	CCACATAAAACTTAAGTGAAACCG	437	57.5
4	MYST4exon4_Fwd	CGTTATTTAGCAGTTTTTCTTTGTT	MYST4exon4_Rev	TGCTATTACGATATAACTGCCTTAAA	350	56
5 to 6	MYST4exon5to6_Fwd	CATTGGCTAGCCTCATCAGC	MYST4exon5to6_Rev	GAACCTGTATACCTCTGAGCTGTC	634	57.5
7	MYST4exon7_Fwd	TTGAGCAGCTTATTACAGCTACAAC	MYST4exon7_Rev	GGAATCTGAGAGAATAACAATCACC	301	57.5
8_1	MYST4exon8_1_Fwd	GAGAAACATCTAGGTGGTGGC	MYST4exon8_1_Rev	GGCACACTGCACTGGCTGG	668	57.5
8_2	MYST4exon8_2_Fwd	CCACACAAAAGCTAAAACCTCC	MYST4exon8_2_Rev	ACCTATGTGACAAATAGGAATTGA	638	57.5
9	MYST4exon9_Fwd	TCCCTATTTGCCCAGTATGC	MYST4exon9_Rev	TCACAAAGGATGTTGAAAGGG	337	57.5
10	MYST4exon10_Fwd	TTGATAGTCACTGGTGAAAGAACC	MYST4exon10_Rev	TCCCTAATTCCTGCTTTAGAATC	283	57.5
11	MYST4exon11_Fwd	AAATTGAACAATATTTAATCTTCCCC	MYST4exon11_Rev	TTTGTACCTTCTAAACACAAATCAGC	309	57.5
12	MYST4exon12_Fwd	GTTTAGGATTTGGAAATAAAATGAT	MYST4exon12_Rev	AGCCCTGTTCAAAGAATTGG	351	57.5
13	MYST4exon13_Fwd	TACAAGGACAGTGGCAGGTG	MYST4exon13_Rev	CTCAATTTCCTAGAGTACAATCACTG	378	57.5
14	MYST4exon14_Fwd	GCGAATGCACTTCTCTGTTG	MYST4exon14_Rev	TGAATGCACCTATCTCCAGG	433	57.5
15	MYST4exon15_Fwd	CATTAGTGCTAGCATATGTCCG	MYST4exon15_Rev	GACAACGATCTTAATGACTTTCTTAGC	330	57.5
16	MYST4exon16_Fwd	GAGACACTTTGCCATTGATCC	MYST4exon16_Rev	AATAGACAGAATGTCTGCAATGAC	509	57.5
17	MYST4exon17_Fwd	CATGTCTACTGCATATCGACTCAAC	MYST4exon17_Rev	CAGAGGCCTGCTCTTTGG	442	57.5
18_1	MYST4exon18_1_Fwd	GGAATGAATTCAAGTTGCCC	MYST4exon18_1_Rev	GTGATCATCCAAACGTGCAG	637	57.5
18_2	MYST4exon18_2_Fwd	GATGATCTCATCAAACCTGAGG	MYST4exon18_2_Rev	TCTGCACGGGTGTAGTTCTG	694	57.5

18_3	MYST4exon18_3_Fwd	CCGTTCAGTCTTTGACCCAG	MYST4exon18_3_Rev	AGCCTTGAGGAGACTTGACG	630	57.5
18_4	MYST4exon18_4_Fwd	CCTCCAGCAGTCTGACACAG	MYST4exon18_4_Rev	GCTGGCAATCTGGGTTTG	629	57.5
18_5	MYST4exon18_5_Fwd	CTCCAATGAATCTGCCGC	MYST4exon18_5_Rev	AATTGCTGGTTTTGAAATCG	656	57.5

Supplementary Table 2a Clinical Features of *KAT6B* positive patients

Individual	Sex	Birth weight (kg)	Typical face	Intellectual Disability/Dev	Feeding difficulties	Contractures	Long great toes	Long thumbs	Dental anomalies	Congenital heart defects	Genital anomalies	Thyroid abnormalities	Abnormal patella	Head circumference %	Hypotonia	Cleft palate	Corpus	Renal anomalies	Optic atrophy	Other
1	F	3.55	+	++	+	+	+	-	-	-	-	-	-	33	+	-	-	-	NK	
2	F	3.38	+	++	+	+	+	+	+	+	-	-	-	<3	+	-	-	NK	+	
3	F	2.67	+	++	+	+	-	-	+	+	-	-	-	NK	+	-	NK	NK	NK	
4	F	2.70	+	++	+	+	-	-	-	+	-	-	-	NK	+	-	NK	NK	NK	
5	M	3.56	+	+	+	+	+	+	-	-	+	-	-	5-10	+	+/-	+	-	-	Narrow EAM and lacrimal ducts, bifid uvula
6	M	2.92	+	++	NK	+	+	+	+	-	+	+	+	+	+	-	+	NK	NK	
7	M	3.05	+	++	+	+	+	+	+	+	+	+	+	25	+	-	-	-	-	
8	F	2.72	+	++	+	+	+	+	-	+	-	+	+	?	+	-	NK	NK	NK	
9	M	3.81	+	+	+	+	-	+	-	-	+	-	-	25	+	-	-	NK	-	>5 CAL , nystagmus, loose skin
10	F	2.98	+	++	+	-	+	+	+	-	-	-	-	50	+	+	+	-	-	Died 3 years bronchiolitis
11	F	3.4	+	++	+	-	-	+	+	+	-	+	+	50-98	+	+	-	-	-	Hypoventilation, tracheomalacia, hearing loss, small EAM
12	M	2.8	+	++	+	-	+	+	+	-	+	-	-	?	+	+	NK	NK	NK	
13	M	3.46	+/-	++	++	-	+	+/-	-	-	+	+	-	10	+	-	-	-	-	Laryngeal cleft
14	F	3.18	+	++	+	+	-	+	-	+	+	-	-	<3	+	-	-	-	-	
15	F	3.54	+/-	++	-	+	+	+	-	-	-	-	-	?	+	-	?	?	?	
16	F	2.80	+	++	+	+	+	+	+/-	+	-	+	-	<0.4	+	-	-	-	NK	
17	M	3040	+	++	NK	+	-	-	+	-	+	-	+	25	+	-	NK	+	NK	Now age 36. Hypermobile
18	M	?	+	++	+	-	+/-	+/-	NK	+	+	+	-	NK	++	-	-	+		Pachygyria, HL
19	M	2.33	++	+	++	+	+	+	+	+	+	-	+	75	++	-	?	?	?	Born 36w deceased
20	F	1.9	+/-	+	++	+/-	-	-	-	+	-	NK	-	NK	-	-	-	-	NK	Malrotation, SNHL
21	M	2.19	+	++	++	+	+	+	NA	I	+	+	-	2-9	-	-	-	-	-	Thyroid agenesis, ing. hernia
22	F	3.3	+	+	++	-	+	+	-	-	-	-	-	10	+	-	NK	+	-	Horner's

															+				
23	M	3.92	+/-	++	+	+/-	+/-	+/-	NK	+	+	-	-	<3	+	+	NK	NK	NK	
24	M	3.44	+	++	++	+	-	+/-	+	+	+	+	NK	25-50	+	+	-	-	-	Sensori-neural hearing loss
25	M	2.40	+	++	+	+/-	+	++	NK	+	+	+	-	25	+	+	-	-	+	Cerebellar abn.
26	F	2.89	+	+	+		+	-	-	-	-	+	-	<3	+	-	-	-	-	myopia
48	F	3.10	-	++	+	+	-	+	-	+	-	+	+	<3	+	+	+	+	NK	malrotation
49	M	2.58	-	++	+	+	-	+	+	+	+	+	+	-	+	-	NK	NK	NK	
50	M	NK	+/-	++	+	+	+	+	n/a	+	+	+	+	NK	+	-	+	+	NK	
51	F	n/a	n/a	n/a	n/a	n/a	++	+	-	n/a	-	-	n/a	?+	nk	n/a	-	+	+	n/a	Fetus 24/40. knee pterygia
53	M	2.81	+	++	NK	-	NK	NK	NK	+	+	+	-	<3	?	+	NK	+	NK	Pre-auricular pit
54	M	3.80	+	++	+	+	-	-	+	+	+	+	+	<3	+	-	+	NK	NK	
55	M	1.90	+	++	-	+	+	+	+	+	+	-	-	-	-	-	NK	NK	NK	
56	M	3.08	+	++	+	-	+	-	+	+	++	+	-	40	+	-	+	-	+	
58	M	50	+	++	+	NK	+	+	+	-	+	-	-	<3	+	-	NK	NK	NK	
59	F	75	+	++	+	NK	+	+	+	-	-	-	+	NK	+	-				Hypoplastic Lacrimal duct
60	F	24	+	++	+	NK	-	-	+	-	-	+	-	-	+	-				
61	M	0.4	+	++	+	NK	+	+	n/k	+	+	+	+	<3	+	+	-	-	NK	hypospadias
62	M	50	+	++	+	NK	+	+	+	+	+	-	+	-	+	-	-	-	+	Athetoid movements
63	F	50	+	++	+	NK	+	+	+	+	-	+	-	-	+	-	NK	-	NK	Mirror movements, absences
64	M	n/k	+	++	+	+	-	-	+	+	+	+	-	NK	+	+	NK	NK	NK	Died at 18m
65	F	9	+	++	+	NK	+	+	+	+	-	+	+	NK	+	+	-	+	NK	
66	F	50	+	++	+	NK	+	+	+	+	-	-	-	-	+	+	-	-	+	Hip dysplasia, seizures
67	F	9	+	++	+	NK	+	+	+	-	-	-	-	-	+	-	-	-	+	11 pairs ribs
68	F	50	+	++	+	NK	+	+	+	-	-	-	-	-	+	+	NK	-	NK	Syncopal episodes
69	M	n/k	+	+	+	NK	-	-	+	-	-	+	-	<3	+	-	-	-	NK	Abn. periventricular s-gnal
71	M	50	+/-	++	+	NK	-	-	-	-	+	-	-	-	+	-	-	-	NK	Mild phenotype

Supplementary Table 2b Clinical Features of *KAT6B* negative patients

Individual	Sex	Weight (%)	Typical face	Intellectual Disability/Developmental delay	Feeding difficulties	Contractures	Long great toes	Long thumbs	Dental anomalies	Congenital heart defects	Genital anomalies	Thyroid abnormalities	Abnormal patella	Microcephaly	Hypotonia	Cleft palate	Agenesis of the cornus callosum	Renal anomalies	Optic atrophy
27	M	2	+/-	+	+	-	-	+	-	+	+	-	+	+	-	-	-	-	-
28	F	NK	+/-	+	-	-	-	-	-	-	-	-	-	+	-	-	-	-	-
29	M	NK	-	NK	-	+	-	-	-	+	-	-	-	-	-	+	-	-	-
30	M	NK	+/-	+	+	-	-	-	-	+	-	-	-	+	-	-	-	-	-
31	F	NK	+/-	-	+	+	+	+	+	+	-	-	-	+	-	-	-	-	-
32	F	NK	+/-	-	-	-	-	-	-	+	-	-	-	-	-	-	-	-	-
33	F	NK	-	-	-	-	-	-	+	+	-	-	-	-	-	-	-	-	-
34	M	NK	+/-	+	+	+	-	-	-	-	-	-	-	+	-	-	-	-	-
35	F	NK	+/-	+	+	+	+	-	-	+	+	-	-	-	+	-	-	-	-
36	M	NK	+/-	+	+	+	-	-	+	+	+	+	-	NK	+	-	-	-	-
37	M	10	+/-	+	-	+	-	-	+	+	-	-	-	+	-	-	NK	-	NK
43	M	NK	+/-	+	+	-	-	-	NK	NK	-	-	-	NK	+/-	-	-	-	-

44	F	3	+	++	++	-	+/-	-	-	-	-	-	-	-	-	-	-	+	-
45	F	NK	+/-	++	+	-	-	-	-	-	-	-	-	NK	++	-	-	-	-
47	M	50	+/-	+	-	-	-	-	-	-	-	-	-	NK	++	-	-	-	-
48	F	<10	+	++	+	-	-	-	-	+	-	-	-	-	+	-	-	-	-
52	M	50	+/-	+	-	+	-	-	-	+	+	-	-	+	+	+	-	-	-
57	M	15	-	++	++	-	-	-	+	-	-	-	-	-	+	-	+	+	-
72	M	NK	+/-	+	+	NK	-	-	+	-	+	-	-	-	-	-	-	-	-
73	M	25	+/-	+	+	NK	-	-	-	-	NK	-	-	+	+	-	-	-	-
74	F	2	+/-	+	+	NK	-	+	-	-	-	-	-	+	-	-	-	-	-
75	?	2-9	+/-	+	-	NK	+	-	-	+	+	-	-	-	-	-	NK	-	NK
76	?	nk	-	+	+	NK	-	-	+	-	-	-	-	-	+	-	NK	NK	NK

Supplementary Table 3 Summary of published sequence variants in *KAT6B*

Changes that are highlighted in GREY have been seen in more than one patient
Reference sequence used NG_032048.1, covering KAT6B transcript NM_012330.3

Phenotype	Exon	Mutation	Type of mutation	Predicted protein change	confirmed to be de novo	Publication	Comment
SBBS	18	c.4405dup	indel	p.(Ser1469Phefs*18)	parents not tested	Clayton-Smith et al.	Case 3 in Clayton-

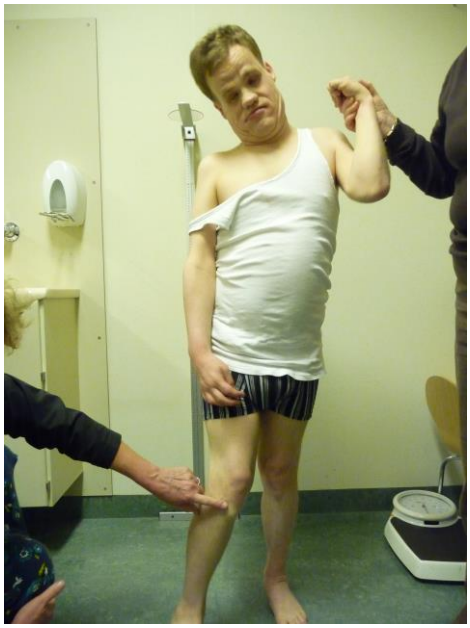
						2011	Smith et al 1994
SBBS	18	c.5370_5373dup	indel	p.(Ile1792Glnfs*12)	parents not tested	Clayton-Smith et al. 2011	
SBBS	15	c.3018del	indel	p.(Glu1007Argfs*5)	De novo	Clayton-Smith et al. 2011	Patient also has 1q21.1 microdup
SBBS	18	c.4069G>T	nonsense	p.(Glu1357*)	De novo	Clayton-Smith et al. 2011	Figure 3 in Day et al 2008
SBBS	18	c.4205_4206del	indel	p.(Ser1402Cysfs*5)	De novo	Clayton-Smith et al. 2011	
SBBS	18	c.5734A>T	nonsense	p.(Arg1912*)	parents not tested	Clayton-Smith et al. 2011	Case 2 in Clayton-Smith et al 1994
SBBS	18	c.5030del	indel	p.(Thr1677Metfs*38)	parents not tested	Clayton-Smith et al. 2011	
SBBS	18	c.5201_5210dup	indel	p.(Gln1737Hisfs*41)	parents not tested	Clayton-Smith et al. 2011	
SBBS	18	c.5201_5210dup	indel	p.(Gln1737Hisfs*41)	De novo	Clayton-Smith et al. 2011	
SBBS	18	c.4205_4206del	indel	p.(Ser1402Cysfs*5)	parents not tested	Clayton-Smith et al. 2011	Case 4 in Clayton-Smith et al 1994
SBBS	3	c.527dup	indel	p.(Tyr176*)	parents not tested	Clayton-Smith et al. 2011	Milder SBBYSS phenotype
SBBS	18	c.4911_4921del	indel	p.(Val1638Alafs*27)	De novo	Clayton-Smith et al. 2011	
SBBS	18	c.5389C>T	nonsense	p.(Arg1797*)	De novo	Clayton-Smith et al. 2011	
SBBYSS	8	c.1078G>A	missense	p.(Glu360Lys)	parents not tested	Clayton-Smith et al. 2011	non-pathogenic, seen in control
GPS	18	c.3892G>T	nonsense	p.(Gly1298*)	parents not tested	Campeau et al. 2012	Case 1 in Abdul-Rahman et al 2006
GPS	18	c.4360_4368delins	indel	p.(Glu1454Lysfs*8)	De novo	Campeau et al. 2012	Case 2 in Abdul-Rahman et al 2006
GPS	18	c.3802G>T	nonsense	p. (Gly1268*)	De novo	Campeau et al. 2012	Case in Lammer and Abrams 2002
GPS	18	c.3769_3772del	indel	p.(Lys1258Glyfs*13)	De novo	Campeau et	Case in

						al. 2012	Lifchez el al 2002
GPS	18	c.3788_3789del	indel	p.(Lys1263Argfs*7)	De novo	Campeau et al. 2012	
GPS	18	c.3769_3772del	indel	p.(Lys1258Glyfs*13)	De novo	Campeau et al. 2012	
GPS	18	c.3680_3695del	indel	p.(Asp1227Glufs*11)	De novo	Simpson et al. 2012	Case in Brugha et al 2011
GPS	18	c.3768_3771del	indel	p.(Lys1258Glyfs*13)	De novo	Simpson et al. 2012	
GPS	18	c.3768_3771del	indel	p.(Lys1258Glyfs*13)	De novo	Simpson et al. 2012	
GPS	18	c.3877A>T	nonsense	p.(Lys1293*)	De novo	Simpson et al. 2012	
GPS	18	c.3773dup	indel	p.(Trp1259Valfs*12)	De novo	Simpson et al. 2012	
SBBS	18	c.5064_5071del	indel	p.(Met1690Glufs*24)	De novo	Szakson et al. 2013	
SBBS	18	c.5389C>T	nonsense	p.(Arg1797*)	De novo	Szakson et al. 2013	Case in Szakszon et al 2011
SBBS	18	c.5623_5624dup	indel	p.(Gln1875Hisfs*5)	De novo	Yu et al. 2013	Initially suspected to have BPES
SBBS	17	c.3401_3402del	indel	p. (Gly1134Ala fs*2)	parents not tested	Patient 1 This study	
SBBS	18	c.4911_4921del	indel	p.(Val1638Alafs*27)	De novo	Patient 2 This study	
SBBS	18	c.5424C>A	nonsense	p. (Tyr1808*)	parents not tested	Patient 3 This study	
SBBS	18	c.5624_5625del	indel	p.(Ala1876Leufs*3)	parents not tested	Patient 4 This study	
SBBS	18	c.5442_5445del	indel	p.(Phe1815Alafs*8)	De novo	Patient 5 This study	
SBBS	18	c.5209 C>T	nonsense	p.(Gln1737*)	De novo	Patient 6 This study	
SBBS	18	c.4205_4206del	indel	p.(Ser1402Cysfs*5)	De novo	Patient 7 This study	
SBBS	18	c.5115_5116del	indel	p.(Tyr170611efs*9)	De novo	Patient 8 This study	
SBBS	18	c.5731C>T	nonsense	p.(Gln1911*)	parents not tested	Patient 9 This study	
SBBS	18	c.5302 C>T	nonsense	p. (Gln1768*)	parents not tested	Patient 10 This study	
SBBS	18	c.5389 C>T	nonsense	p.(Arg1797*)	parents not tested	Patient 11 This study	
SBBS	16	c.3147 G>A	missense*	p. Pro1049Pro	De novo	Patient 12 This study	
SBBS	16	c.3147 G>A	missense	p. Pro1049Pro	De novo	Patient 13 This study	
SBBS	16	c.3147 G>A	missense	p. Pro1049Pro	De novo	Patient 14 This study	
SBBS	18	c.4728_4729del	indel	p.(Arg1577Cysfs*21)	De novo	Patient 15 This study	
SBBS	18	c.4312del	indel	p.(Glu1438Lysfs*23)	parents not tested	Patient 16 This study	
SBBS	17	c.3366_3369del	indel	p.(Lys1124Glyfs*5)	De novo	Patient 17	

						This study	
SBBS	18	c.5389 C>T	nonsense	p.(Arg1797*)	parents not tested	Patient 18 This study	
SBBS	18	c.5389 C>T	nonsense	p.(Arg1797*)	parents not tested	Patient 19 This study	
SBBS	16	c.3064 G>T	nonsense	p.(Glu1022*)	parents not tested	Patient 20 This study	
SBBS	18	c.4205_4206del	indel	p.(Ser1402Cysfs*5)	parents not tested	Patient 21 This study	
SBBS	18	c.3040C>T	nonsense	p.(Gln1014*)	De novo	Patient 22 This study	
SBBS	18	c.4114G>T	nonsense	p.(Glu1372*)	De novo	Patient 23 This study	
SBBS	18	c.5131del	indel	p.(Ser1711Profs*4)	parents not tested	Patient 24 This study	
SBBS	18	c.5302C>T	nonsense	p.(Gln1768*)	De novo	Patient 25 This study	
SBBS	18	c.2959T>C	missense	p.(Trp987Arg)	parents not tested	Patient 26 This study	
SBBS	18	c.4320_4321del	indel	p.(Lys1441Glyfs*23)	De novo	Patient 53 This study	
SBBS	18	c.4205_4206del	indel	p.(Ser1402Cysfs*5)	De novo	Patient 54 This study	
SBBS	18	c.4074_4079del	indel	p.(Glu1367_Glu1368)	De novo	Patient 55 This study	
GPS	17	c.3598_3622dup25	indel	p.(Gly1208Glufs*19)	De novo	Patient 48 This study	
GPS	18	c.3962_3963del	indel	p.(Gln1321Argfs*20)	parents not tested	Patient 49 This study	
GPS	18	c.3769_3772del	nonsense	p.(Lys1258Glyfs*13)	Parents not tested	Patient 50 This study	
GPS	18	c.4110dupA	nonsense	p.(Glu1371Argfs*16)	De novo	Patient 51 This study	
GPS/SBBS	18	c.5302C>T	nonsense	p. (Gln1768*)	De novo	Patient 56 This study	

Supplementary Figure 1

Whole Body Habitus of Patient 17, demonstrating adult phenotype of SBBS



Supplementary Figure 2

Long, straight thumbs which are proximally placed, long hallux and overlapping of toes which are common features in Say Barber Biesecker syndrome.