KAT6A Syndrome: Genotype-phenotype correlation in 76 patients with pathogenic *KAT6A* variants

Supplementary Materials

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Table 1

Patient number	genomic DNA (GRCh37/hg19)	coding DNA (NM_006766.3)	protein change (NP_006757.2)	Exon	Inheritance
1	chr8:g.41790230_41790233del	c.5505_5508del	p.N1836Lfs*15	17	De Novo
2	chr8:g.41791525C>A	c.4213G>T	p.E1405*	17	De Novo
3	chr8:g.41791694_41791695del	c.4043_4044del	p.Q1348Rfs*7	17	Maternal mosaic
4	chr8:g.41812905delC	c.1507delG	p.D503Ifs*42	9	De Novo
5	chr8:g.41801291G>A	c.2203C>T	p.R735*	13	De Novo
6	chr8:g.41792223delC	c.3515delG	p.G1172Dfs*4	17	De Novo
7	chr8:g.41906298_41906301del	c.195_198del	p.Asn65Lysfs*15	2	De Novo
8	chr8:g.41839377G>A	c.805C>T	p.R269*	4	De Novo
9	chr8:g.41795071G>A	c.3055C>T	p.R1019*	16	De Novo
10	chr8:g.41792357_41794780del	c.3345_3346insAGTCAGATGA	p.A1116Sfs*11	16	De Novo
11	chr8:g.41791669G>A	c.4069C>T	p.Q1357*	17	De Novo
12	chr8:g.41832264_41834580del	c.1308_1309insCGCAA	p.Y437Rfs*43	7	De Novo
13	chr8:g.41791481_41791484del	c.4254_4257del	p.E1419Wfs*12	17	De Novo
14	chr8:g.41906450del	c.46del	p.A16Pfs*5	2	De Novo
15	chr8:g.41791093C>T	c.4645G>A	p.G1549S	17	De Novo
16	chr8:g.41791066T>G	c.4672A>C	p.S1558R	17	De Novo
17	chr8:g.41832342T>A	c.1364-2A>T	acceptor splice site for exon 8	8	unknown
18	chr8:g.41792353G>A	c.3385C>T	p.R1129*	17	De Novo
19	chr8:g.41834753G>T	c.1136C>A	p.R1129*	7	Father deceased, no maternal inheritance
20	chr8:g.41791696G>A	c.4042C>T	p.Q1348*	17	De Novo
21	chr8:g.41795056G>A	c.3070C>T	p.R1024*	16	De Novo
22	chr8:g.41792353G>A	c.3385C>T	p.R1129*	17	De Novo
23	chr8:g.41838363delC	c.907+1delG	Splice site	intron_between_exon_5_and_6	De Novo
24	chr8:g.41834605_41834606insA	c.1283_1284insT	p.E429Gfs*7	7	De Novo
25	chr8:g.41791713del	c.4025del	p.K1342Rfs*11	17	De Novo
26	chr8:g.41795071G>A	c.3055C>T	p.R1019*	16	De Novo
27	chr8:g.41791376dupC	c.4362dupG	p.T1455Dfs*9	17	De Novo
28	chr8:g.41834777G>T	c.1112C>A	p.S371Y	7	maternally inherited
29	chr8:g.41791357G>A	c.4381C>T	p.Q1461*	17	unknown
30	chr8:g.41791085A>C	c.4653T>G	p.S1551R	17	De Novo
31	chr8:g.41792353G>A	c.3385C>T	p.R1129*	17	De Novo
32	chr8:g.41794839_41794840insG	c.3286_3287insC	p.Cys1096Serfs*6	16	De Novo
33	chr8:g.41791464_41791465del	c.4273_4274del	p.VAL1425Thrfs*13	17	unknown
34	chr8:g.41791506_41791510del	c.4228_4232del	p.K1410Gfs*7	17	Not tested
35	chr8:g.41794944A>C	c.3182T>G	p.L1061*	16	De Novo
36	chr8:g.41795056G>A	c.3070C>T	p.R1024*	16	De Novo
37	chr8:g.41798488dupG	c.2911dupC	p.Arg971Profs*5	15	De Novo
38	chr8:g.41836254G>A	c.949C>T	p.Arg317*	6	De Novo
39	chr8:g.41805352dupA	c.1819dupT	p.Y607Lfs*16	11	De Novo
40	chr8:g.41794840dupA	c.3286dupT	p.C1096Lfs*6	16	De Novo
41	chr8:g.41798716G>A	c.2683C>T	p.P895S	15	De Novo
42	chr8:g.41792373A>C	c.3365T>G	p.Leu1122*	17	De Novo
43	chr8:g.41792077C>A	c.3661G>T	p.E1221*	17	De Novo
44	chr8:g.41792304del	c.3434del	p.Pro1145Leufs*2	17	De Novo
45	chr8:g.41795056G>A	c.3070C>T	p.Arg1024*	16	De Novo
46	chr8:g.41792353G>A	c.3385C>T	p.Arg1129*	17	De Novo
47	chr8:g.41806840_41806841del	c.1639_1640del	p.Met547Glufs*3	10	De Novo
48	chr8:g.41804203C>T	c.1903-1G>A	Splice site	Intron 11, Exon 12	De Novo
49	chr8:g.41906495T>C	c.1A>G	p.M1V	Loss of Start Codon	paternally inherited
50	chr8:g.41906495T>C	c.1A>G	p.M1V	Loss of Start Codon	paternally inherited
51	chr8:g.41834793G>A	c.1096C>T	p.Arg366*	7	De Novo
52	chr8:g.41792353G>A	c.3385C>T	p.R1129*	17	De Novo

Previously Reported Cases					
53	chr8:g.41791858_41791859insG	c.3879_3880insC	p.E1294Rfs*19	17	De Novo
54	chr8:g.41795009_41795010delAGinsTT	c.3116_3117delCTinsAA	p.S1039*	16	De Novo
55	chr8:g.41795009_41795010delAGinsTT	c.3116_3117delCTinsAA	p.S1039*	16	De Novo
56	chr8:g.41791445_41791446insG	c.4292_4293insC	p.L1431Ffs*8	17	De Novo
57	chr8:g.41791630C>A	c.4108G>T	p.E1370*	17	De Novo
58	chr8:g.41791906_41791907insCT	c.3831_3832insAG	p.R1278Sfs*17	17	De Novo
59	arr 8p11.21 (41,786,230-42,022,328) exons 1–18			deletion of gene	De Novo
60	chr8:g.41792353G>A	c.3385C>T	p.R1129*	17	De Novo
61	chr8:g.41792353G>A	c.3385C>T	p.R1129*	17	De Novo
62	chr8:g.41795056G>A	c.3070C>T	p.R1024*	16	De Novo
63	chr8:g.41792353G>A	c.3385C>T	p.R1129*	17	De Novo
64	chr8:g.41792275delC	c.3464delG	p.G1155Afs*21	17	De Novo
65	chr8:g.41794839delC	c.3287delG	p.C1096Ffs*27	16	De Novo
66	chr8:g.41794899delT	c.3230delA	p.N1077Mfs*46	16	De Novo
67	chr8:g.41795056G>A	c.3070C>T	p.R1024*	16	De Novo
68	chr8:g.41804177T>C	c.1928A>G	p.N643S	12	De Novo
69	chr8:g.41937569_41937570delCT	c.3040-1_3040delGA	splice site	[16]	De Novo
70	chr8:g.41791886_41791887dupCC	c.3851_3852dupGG	p.Q1285Gfs*10	17	De Novo
71	chr8:g.41791049A>T	c.4689T>A	p.Y1563*	17	De Novo
72	chr8:g.41906261G>A	c.235C>T	p.R79*	2	De Novo
73	chr8:g.41792081_41792083	c.3655_3657	p.L1219Tfs*75	17	De Novo
74	chr8:g.41795056G>A	c.3070C>T	p.R1024*	16	germline mosaic
75	chr8:g.41795056G>A	c.3070C>T	p.R1024*	16	germline mosaic
76	chr8:g.41795056G>A	c.3070C>T	p.R1024*	16	germline mosaic

Supplemental Table 1: **Reported genetic Mutations in *KAT6A* in our cohort of 76 individuals.** If known, inheritance of mutation was provided.

Sex	F	F	F	M	M	M	F	M	F	F	M	F	F	M	M	M	M	M	F	M	F	M	F	M	F	M
Age (years)	16	8	18	9	9	5	21	23	15	7	11	9	4	5	13	23	14	7	30	10	7	9	8	5	9	9
Mutation type	fs	n	fs	fs	n	fs	fs	n	n	fs	n	fs	fs	fs	m	m	S	n	n	n	n	n	s	n	fs	n
Exon (for splice site cases in=intron)	17	17	17	9	13	17	2	4	16	16	17	7	8	2	17	2	In7	17	7	17	16	17	In5	7	17	16
Collection route C=clinician P=patient/family L=literature	c	c	c	c	c	c	c	c	c	c	c	c	c	c	c	c	c	c	c	c	c	c	p	P	p	c
SGA	-	-	-	-	-	-	-	?	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-
Microcephaly	-	?	+	-	-	?	-	-	-	-	+	-	-	-	?	-	-	+	-	-	+	-	-	-	-	+
Presence/Degree of ID*	2	2	3	1	1	3	1	1	2	3	3	1	2	1	3	2	2	2/3	1	?	3	3	2	+	+	3
Neonatal Hypotonia	+	+	-	-	+	+	-	-	+	+	?	-	-	+	+	-	+	+	-	+	+	+	+	-	+	+
Seizures	-	-	-	-	-	-	-	-	-	-	-	+	-	-	+	-	+	-	-	-	-	-	-	-	-	-
Speech delay	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+
Strabismus	+	+	-	-	-	+	-	-	-	+	+	+	-	+	?	+	?	+	+	-	-	-	+	+	+	-
Ptosis	-	-	-	-	+	-	-	-	?	-	-	-	-	-	-	-	-	-	-	-	+	-	-	+	-	-
Visual Defect	?	+	+	-	+	+	-	-	+	?	+	+	-	+	?	-	?	+	+	-	+	+	+	+	+	+
Broad nasal tip	+	+	+	+	+	+	+	+	+	+	+	+	+	-	+	+	+	+	+	?	+	+	-	-	?	+
Thin upper lip	+	+	+	+	+	+	-	-	+	+	+	+	+	-	?	+	+	-	+	?	?	+	+	-	?	+
Feeding difficulties	+	+	+	+	+	+	-	-	+	+	+	-	+	+	+	+	?	+	+	+	+	-	+	+	+	-
Reflux	+	+	-	+	-	+	-	-	+	?	-	-	+	-	-	+	?	-	+	?	-	-	+	+	+	-
Constipation	?	+	-	+	-	?	-	-	+	+	-	-	+	-	+	+	?	+	-	?	-	+	+	-	-	+
Congenital heart defect	-	-	+	-	+	+	-	-	-	+	-	+	+	-	-	-	-	+	+	+	-	+	+	+	+	+
Frequent Infection	+	+	+	-	-	-	+	-	+	+	+	-	+	-	-	-	+	+	-	+	-	+	?	+	-	+
Behavioural problems	?	+	+	-	-	-	-	+	-	+	+	-	+	-	+	+	+	-	-	?	-	+	-	-	-	+
Sleep disturbance	-	+	+	+	-	+	-	-	-	?	+	-	+	-	+	+	-	-	-	+	-	+	?	+	+	-

Feature	Patient Number																										
Patient number	27	28	29	30	31	32	33	34	35	36	37	38	39	40	41	42	43	44	45	46	47	48	49	50	51	52
Sex	M	F	F	M	F	M	M	F	M	F	F	M	F	M	M	M	M	F	F	F	F	F	M	M	M	M
Age (years)	3	4	1	2	5	2	7	8	9	1	22	21	2	12	6	3	1	5	18	5	7	19	8	20	4	3
Mutation type	fs	m	n	m	n	fs	fs	fs	n	n	fs	n	Fs	fs	m	n	N	fs	n	n	fs	s	fs[1]	fs[1]	n	n
Exon (for splice site cases in=intron)	17	7	17	17	17	16	17	17	16	16	15	6	11	16	15	17	17	17	16	17	10	In11	1	1	7	17
Collection route C=clinician P=patient/family	p	p	p	p	p	p	p	p	p	p	c	c	P	p	p	c	P	c	c	c	c	c	C	C	C	C
SGA	-	-	+	?	-	-	+	+	-	+	-	+	+	-	-	+	-	-	-	-	-	-	?	?	-	-
Microcephaly (A=acquired)	-	-	?	+	+	-	-	-	-	-	-	-	-	?	-	+	+	+A	-	+A	-	?	-	-	-	+
Presence/Degree of ID*	+	?	+	?	?	+	?	1	+	?	1	1	1	+	+	2	?	3	3	3	1	2	2	3	2	3
Neonatal Hypotonia	+	+	+	+	+	-	+	+	-	+	-	+	+	-	+	+	+	+	+	-	+	+	-	-	+	+
Seizures	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	?	+	-	-	-	-	-
Speech delay	+	+	?	?	+	?	+	+	+	?	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+
Strabismus	+	-	+	-	-	-	+	+	+	-	-	+	?	-	-	-	-	+	+	+	+	?	-	+	+	+
Ptosis	+	-	-	-	?	-	-	-	-	-	-	-	-	-	-	-	-	-	-	?	-	+	-	-	-	-
Visual Defect	+	-	-	+	+	-	+	-	+	-	+	-	-	-	?	+	+	+	-	?	+	+	-	-	+	+
Broad nasal tip	+	-	-	-	+	-	-	-	+	-	+	+	+	?	?	+	+	+	+	+	+	+	+	+	+	+
Thin upper lip	+	-	-	-	+	+	-	-	+	-	-	-	?	?	?	+	+	+	-	-	+	+	-	-	-	-
Feeding difficulties	+	+	+	+	+	+	+	-	-	+	-	+	+	+	-	+	+	+	+	+	-	+	-	-	+	+
Reflux	+	+	+	+	+	+	+	-	-	+	-	+	+	+	-	+	+	+	+	+	-	-	-	-	-	-
Constipation	+	-	-	+	+	+	+	+	+	-	-	-	-	+	-	+	-	+	?	?	+	+	-	+	-	-
Congenital heart defect	+	-	-	-	+	-	+	+	-	+	-	-	-	-	-	+	+	+	-	+	-	-	-	-	-	+
Frequent Infection	+	+	?	-	+	-	+	-	-	-	-	+	+	-	-	+	+	-	?	-	-	-	-	-	-	+
Behavioural problems	?	?	?	?	?	?	?	?	?	?	+	+	-	?	+	-	?	-	-	-	-	-	-	-	?	?
Sleep disturbance	+	-	?	-	-	-	+	-	-	+	-	-	-	-	?	+	+	-	?	?	-	-	-	-	-	-

Feature	Patient Number																							
Patient number	53	54	55	56	57	58	59	60	61	62	63	64	65	66	67	68	69	70	71	72	73	74	75	76
Sex	F	F	F	M	F	M	M	M	F	M	F	F	M	F	F	F	M	F	F	M	F	M	M	M
Age (years)	5	12	D	3	8	4	19	5	5	7	6	3	10	12	6	7	30	5	10	3	3	14	11	1
Mutation type	fs	n	n	fs	n	fs	del	n	n	n	n	fs	fs	fs	n	m	S	n	n	Fs	fs	n	n	n
Exon (for splice site cases in=intron)	17	16	16	17	17	17	1-17	17	17	16	17	17	17	17	16	12	16	17	2	17	17	16	16	16
Collection route C=clinician P=patient/family L=literature[2]	L T1	L T2	L T3	L T4	L T5	L T6	L T7	L A1	L A2	L A3	L A4	L M1	L M2	L M3	L M4	L M5	L M6	L GV1	L ZS1	L MU1	L E1	L S1	L S2	L S3
SGA	-	-	-	-	-	-	-	-	-	+	-	?	?	?	?	?	?	-	-	-	+	+	-	-
Microcephaly	+	-	-	-	-	+	+	+	+	+	+	+	-	-	-	-	-	-	-	+	+	-	-	-
Presence/Degree of ID*	+	+	+	+	+	+	1	+	+	+	+	+	+	+	+	+	+	+	1	+	+	+	+	+
Neonatal Hypotonia	+	+	+	+	+	+	+	+	-	+	+	+	+	-	+	+	+	+	+	+	+	+	+	+
Seizures	-	-	+	-	-	+	-	-	-	-	-	-	-	-	-	-	+	-	?	-	-	-	-	-
Speech delay	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	?
Strabismus	-	+	+	-	+	-	+	+	+	+	-	-	-	-	-	-	-	+	-	+	+	+	+	?
Ptosis	-	+	+	-	-	-	-	-	-	+	+	-	-	-	+	-	-	-	+	+	-	-	-	-
Visual Defect	-	-	-	-	+	+	+	?	?	+	+	?	?	?	?	?	?	+	?	+	+	+	-	?
Broad nasal tip	+	+	+	+	+	+	+	+	+	?	+	+	+	?	+	+	+	+	+	+	+	?	?	?
Thin upper lip	+	+	+	+	+	+	+	+	+	?	+	+	+	?	?	+	+	+	-	-	+	?	?	?
Feeding difficulties	+	+	+	+	-	+	-	+	+	+	-	?	+	+	?	-	?	+	+	+	+	+	+	+
Reflux	?	?	?	?	-	?	-	+	+	+	-	+	+	+	+	-	+	+	+	+	+	?	?	?
Constipation	?	?	?	?	?	?	?	?	?	?	?	-	+	+	-	-	-	?	?	?	+	?	?	?
Congenital heart defect	+	+	+	+	+	-	-	+	+	-	+	+	+	-	+	-	-	-	-	+	+	?	?	+
Frequent Infection	?	?	?	?	?	?	?	+	+	-	+	?	+	?	?	?	?	-	?	?	+	?	+	?
Behavioural problems	?	?	?	?	?	?	?	?	+	?	?	?	?	?	?	?	+	+	?	?	-	?	?	?
Sleep disturbance	?	?	?	?	?	?	?	+	?	?	+	?	?	?	?	?	?	-	?	?	+	?	?	?

Supplemental Table 2: Complete Summary Table individual level data of phenotypic features. [1]=In cases 49 and 50 the start codon ATG is changed to GTG, the nearest ATG's (both before and after) are out of frame so this would be predicted to result in a frameshift , [2]The literature codes refer to the first initial of the first author and the given patient number in the paper reporting each individual. **The references for each paper are also indicated at the end of this file.** T=[1], A = [2], M=[3] GV=[4] , MU=[5], E =[6] ,ZS =[7], S=[8]

Supplemental Table 3: List of additional features found in patients

Patient Number	*Further details and features of note*
1	Strabismus required surgery, bilateral 5th finger clinodactyly, possible absent lacrimal duct on right, persistent thrombocytopenia since birth. Used signs as communication aid.
2	Sensory issues (doesn't like loud noises, likes and dislikes certain textures) Hand flapping and chews when anxious. Found to be aspirating feeds at 1 year, thickeners helped. Delayed visual maturation, myopia, occasional strabismus. Initial MRI scan showed the corpus collosum was mildly thinned, delayed myelination was noted but has since resolved. Used mackaton signs as communication aid. B cell immunodeficiency and hypogammaglobulinaemia, cyclical neutropenia (low IgG, low mannan-binding, IgA low, CD3 and CD4 low, poor response to pneumococcal vaccine) on subcutaneous immunoglobulins. IV Immunoglobulin therapy 3 weekly for Common Variable Immune Disease (CVID). Chronic fatigue. Severe ear infections and permanent perforated ear drums. Frequent respiratory tract infections and reactive airway disease. On Melatonin for sleep dysfunction.
3	Sociable, happy as young child; anxious+++ and aggressive as teenager; chews e.g. blankets, straps; hand flaps when excited. Episodes of "inappropriate laughter" and hand-flapping. Significant speech delay, only uses a few words at 17yrs. Intermittent strabismus, glasses for refractory correction. Bilateral 5th finger camptodactyly. ASD did not require surgery. Hypothyroidism. On Melatonin for sleep dysfunction.
4	Persistent swallowing difficulties well beyond infancy. He vomits up to an hour and a half after eating. Right inguinal hernia repaired aged 9 weeks. Significant articulation difficulties, receptive language assessed as within normal range.
5	PDA present at 7 years. Hypermetropia. Bilateral cervical ribs. Used signs as communication aid but now has good verbal vocabulary. Bilateral conductive hearing loss, grommets and hearing aids.
6	Uses a PECS board as communication aid. Poor oral-oesophageal co-ordination and a gut motility disorder. Surgery as infant for duodenal web and malrotation. Mild pulmonary stenosis. Bilateral undescended testes requiring surgery. Cortical visual impairment.
7	Mostly extensive herpes simplex infections, involving the face and eyes (keratitis) requiring valaciclovir prophylaxis
8	Features of autism and attention deficit disorder.
9	Supernumerary teeth. Right sided conductive hearing loss (has frequent ear infections). Registered partially sighted due to photophobia. Significant speech delay, mainly uses signs.
10	Autistic features. Unable to walk at 6 years. No meaningful speech. Oculomotor apraxia. Plagiocephally. Kyphosis. ASD and PDA requiring surgery at 2 years. Conductive hearing loss, grommets inserted.
11	Oromotor dysfunction, uses Makaton and own signs, Age 11 less than 10 words, understanding said to be good. Left amblyopia. Mild plagiocephaly, bitemporal narrowing, prominent columella. Testosterone undetectable. Bilateral cryptorchidism with bilateral orchidopexy Poorly formed scrotum. Unilateral 5th finger clinodactyly. Latent horizontal nystagmus. Hypermetropia. Given booster immunisations as low immune response mounted to HiB and pneumococcal vaccine. Igs normal levels. Minimal tears. Drooling is a significant problem. Doesn't like sunlight. Protects his eyes, and walks with head down.

12	Had several complex partial seizures seizure between the ages of two to three. An EEG performed at age 2 was normal. Single word speech started at 2years. VSD which did not require surgery. Strabismus, treated with patching from age three, she has residual amblyopia. Myopia. 5th finger clinodactyly.
13	Left pre-auricular pit. Lower incisors very delayed to appear and pointy. Supernumerary nipple, raised ridge down centre of tongue. Mild LPA stenosis, small PFO, small PDA.
14	Sagittal Craniosynostosis. Horse shoe kidney. Amblyopia.
15	High palate. Small teeth.
16	Autistic features. Inappropriate laughter. Delayed myelination noted on MRI brain. Right convergent squint when tired. T shaped incisor teeth. Supernumerary nipples. Significant problems with reflux continued well after infancy, regurgitation of food up to an hour after eating. Brachydactaly and tapering fingers. Prominent costal cartilages on the left side of the chest.
17	Autistic features (borderline diagnosis of autism) Possibility of ADHD considered. Behaviour described as challenging and unpredictable. Hysterical uncontrolled laughing at times when younger. Absences with abnormal EEG. Oromotor dyspraxia, articulation difficulties with dysphonic voice. Habitual clearing of throat. Slightly up slanting palpebral fissures. Right preauricular pit. Sub-mucosal cleft palate. Recurrent ear infections.
18	MRI showed delayed myelination and thin corpus collosum. Bilateral esotropia with increasing myopia. Small posteriorly rotated ears with extra folding. Retromicrognathia. High arched thin palate. Small peg shaped teeth. Bi temporal narrowing. Primitive oral motor co-ordination. Difficulty chewing. Episodes of choking. ASD and PDA not causing significant haemodynamic compromise. Bilateral cryptorchidism requiring surgery, hypospadias. Chronic serous otitis media and cough prior to bilateral myringotomy with tympanostomy tube placement. Mild to moderate hearing loss, combination of conductive and sensory neural. Bilateral esotropia, myopia, astigmatism. Progressive scoliosis and torticollis to the left. He had multiple contractures and was diagnosed with arthrogryposis multiplex congenita. Most prominent involvement was the hands and feet, but the hips knees and elbows were also contracted. Hands were initially held in flexion with fisting present bilaterally. They can, though, be taken to the neutral position. His hands have ulnar deviation bilaterally. Long fingers and mild camptodactyly.
19	4 words at 4 years, now speaks in full sentences. Had a fundoplication (severe GERD/reflux) and G/J tube placement for 1 year. ASD required surgical repair, Mild mitral and tricuspid regurgitation. Accessory pancreas removed. Reactive airway disease, requires oxygen at night. Iron deficiency anaemia, on supplements. Hashimoto's thyroiditis, diabetes, unclear whether type one or type two due to unusual clinical features, clinical team considering whether intermittent elevated BS should be treated with once a week insulin.
20	Diagnosed clinically with a mitochondrial disorder. Although his muscle biopsy electron transport chain analysis was not diagnostic, his multi-system disorder with persistent elevations in pyruvate and abnormal mitochondrial on muscle electron microscopy placed him in a category of a definite disorder of mitochondrial metabolism (based on the modified Walker criteria). MRI brain shows mild, non-specific findings of delayed myelination with periventricular white matter changes. Absent speech at 4 years. Feeding difficulties and growth failure lead to gastrostomy. Large atrial septal defect. Cardiomyopathy. Hypogammaglobulinaemia. Central

	obstructive sleep apnoea.
21	Blepharophimosis, low set and posteriorly rotated ears, hypermetropia.
22	Acquired microcephaly, required 3x neurosurgery (3rd ventriculostomy at 11 months, Chiari decompression at 23 months, tethered cord repair at 3 years, 3 months) Note- regression was noted prior to surgeries which is not generally seen in KAT6A syndrome. MRI brain showed variant venous anatomy including anomalous venous sinus that traverses the falx cerebri; overall pattern suggests a much greater than typical fraction of the venous return is via anterior pathways. End gaze nystagmus, blue tinted sclera, palpebral fissure R 27mm L 27mm; significant lower lid lagophthalmos; upper lid skin was thin with eyelid vasculature showing; dry eye syndrome, myopia with astigmatism. Small teeth, short teeth with enamel dysplasia. Mandibular midline displaced to right. Mild pectus carinatum and narrow hypotonic shoulder girdle, low set wide spaced nipples, small accessory nipples medial and inferior to primary nipples. Small bowel obstruction with necrosis of 5cm resection of ileum. PFO resolved, ASD. Mild to moderate hearing loss in right ear; etiology unknown. Small fingers and toes with tapering appearance, persistent fetal pads, bilateral simian crease, hypoplastic nails, contractures of 3.4.5 fingers bilaterally, pes planus. Skeletal abnormalities include kyphosis, mild lateral curve of spine, small sacral pit; beaten copper skull, thick skull, slightly think clavicles, cervical spine and rib anomalies, slightly advanced bone age
23	No speech at 3.5 years. Low set ears with thick helices. High palate. Incisor notching. Unilateral talipes equinovarus. Hypermetropia. Congenital hip dysplasia.
24	Fundoplication, Gastro-jejunal tube placement for 1 year. ASD requiring surgical repair. Mild mitral and tricuspid regurgitation. Supraventricular tachycardia at 8 weeks, on propranolol. Infection of site after muscle biopsy, for 8 weeks post biopsy.
25	Some behavioural difficulties, difficultly maintaining focus, sensory issues. Frequent gagging and vomiting in early years. Immature chewing and possibly a slow swallow reflex. ASD required repair. Central obstructive sleep apnoea. Several allergies.
26	Some behavioural difficulties, difficultly maintaining focus, sensory issues. First word "mama" at 7 years, can sight read but only says 4 words, communicates with adaptive technology. Left preauricular ear pit, thick helices. Repaired sagittal craniosynostosis, micrognathia, prominent sagittal suture ridge and flat occiput. PDA required closure at 12 mo, hypoplastic left pulmonary vein leading to severe pulmonary hypertension with chronic lung disease. Moderate hydronephrosis. Lymphopenia in first year of life. Recurrent infections.
27	Low set rotated ears. Small jaw. Widely spaced nipples. Asymmetry of facial features. ASD, VSD, PDA, surgically corrected. Nephrocalcinosis. Chronic otitis media requiring grommets, Severe RSV associated pneumonia requiring hospitalisation for 29 days.
28	Low set ears. Small teeth.
29	Low set ears. Small jaw. Wide set nipples. Dysphagia. Blocked tear ducts.
30	NICU for 2 weeks, traceobroncomalaceia. Microcephaly. Plagiocephaly. Frontal bossing. Dysphagia. Moderate to mild sensory neural hearing loss.
31	Slight thinning of the corpus callosum. Dysphagia. Chronic otitis media requiring grommets.
32	Astigmatism.
33	MRI showed Hydrocephalus Slit Ventricles Stiff Ventricles, Hyperintense signals in the brain, Chiari Malformation. Low set ears and ear tags. Wide spaced nipples. Craniosynostosis. Dysphagia. Eosinophilic esophagitis. Gastrostomy and gastrostomy-

	jejunostomy required at times. ASD. Cardiomyopathy and an enlarged ascending aorta, requiring surgery. Blocked tear ducts. Atlanto-Axial Instability/Dislocation C1 bone was not developed fully (bipartite and splayed). Interstitial Lung Disease, chronic Respiratory Failure, Restrictive Lung Disease, Tracheomalacia. Central obstructive sleep apnoea. Bone marrow failures after RSV infection requiring platelet and red blood cell transfusions. Persistent oxygen requirement following RSV infection.
34	MRI showed delayed myelination. Low set ears with ear tags. Dysphagia. PFO which resolved.
35	Autism. Malabsorption. Iron deficiency. Amblyopia and astigmatism. Inguinal hernia repair.
36	Cleft Palete and small jaw. VSD. Pulmonary hypertension. Gastrostomy and GI dysmotility. Central obstructive sleep apnoea.
37	Vocal cord paresis secondary to abnormal tracheal rings. Sporadic anger outbursts. Small deep-set eyes, small palpebral fissures, epicanthus fold as a child. Mild Hypertelorism. Myopia. Tracheomalacia. Bronchial hyper reactivity.
38	Autism. Didn't speak until 6 years of age, nasal speech. Synophris. Small palpebral fissures and deep-set eyes. Crowding of the teeth. Broad forehead and prominent chin. Gastrostomy required. Growth retardation in younger years. Small hands. Recurrent otitis media.
39	Dysphagia. Gagging/retching/vomiting.
40	Abnormal teeth.
41	Sensory processing disorder. Lack of myelination noted on initial MRI which has since resolved. Hydronephrosis. Myopia and Astigmatism. Blocked tear ducts. C1 bone was not fully developed (bipartite and splayed).
42	MRI findings: Borderline type 1 chiari malformation, bilateral trigone choroid plexus cyst. No specific features on MRI to explain DD. Thin arched eyebrows. Low set, posteriorly rotated ears. Surgical intervention for bowl malrotation, umbilical hernia, drooling, mild iron deficient anaemia, cow's milk protein intolerance. ASD requiring surgical closure. Unilateral orchidoplexy surgery to correct malrotation. Bi-lateral inguinal hernia repair. Recurrent otitis media requiring grommets. Unilateral overlapping 2nd toe. Broad big toes. Left single palmer crease. Dysplastic nails. Eczema.
43	Microcephaly. Required gastrostomy. Dysphagia. ASD and PDA. Undescended testes requiring double orchidopexy. Craniosynostosis. Required intubation for two viral respiratory tract infections. Tracheomalacia. Obstructive sleep apnoea.
44	Acquired microcephaly. Four limb dystonia. Low set ears. Required NG feeding for first 5mths of life. ASD. Astigmatism. Intermittent esotropia. Coxa vara deformity of the hips. Reduced sweeting.
45	Non-verbal into adulthood. Surgically corrected strabismus. Low set, posteriorly rotated ears. High palate. Severe reflux. Multiple episodes of aspiration pneumonia. Chronic lung disease. Oxygen dependent 9 weeks from birth.
46	Staring episodes, no grand mal seizures. Low lying cerebellar tonsils (query Arnold-Chiari type I malformation). Severely delayed speech. Crumpled ears with pre-auricular pits. Dilated foetal bowel loops, Recurrent bilious vomiting, Severe GER, Diagnosed with gastroesophageal nerve impairment. GI Procedures: Gastro-jujenostomy, Nissen fundoplication. Anal stenosis. Acute bowel obstruction that required 3 laparotomies. She had GI failure and parents decided to transfer her to Hospice care where she passed away. Patent ductus arteriosus (PDA) and large

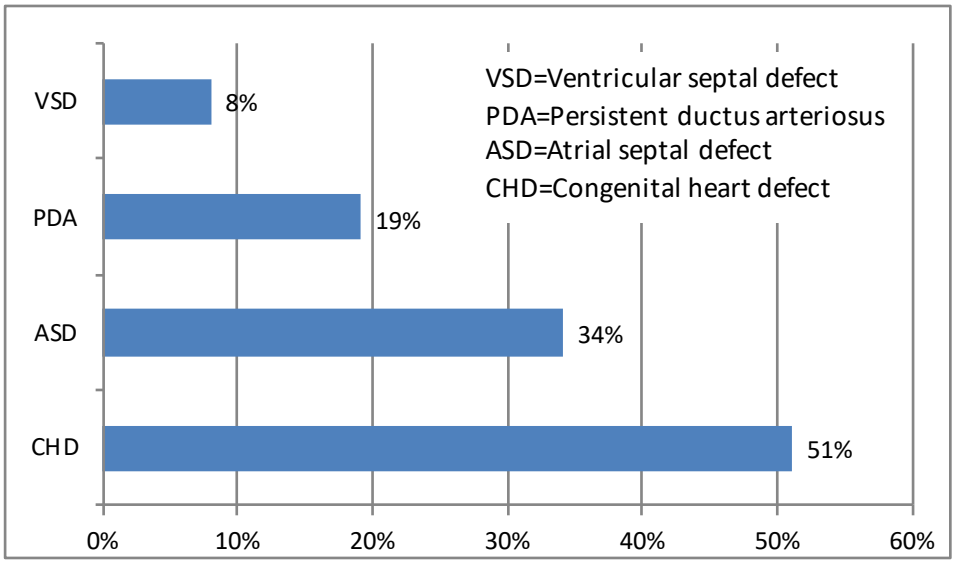
	secondum atrial septal defect (ASD). Bilateral fetal hydronephrosis.
47	Absence seizures and eye myoclonia, classified as Jeavons epilepsy. Verbal dysplaxia, understanding much better that speech, articulation difficulties. One extra tooth noted, large incisors. Brachydactaly, mild clinodactyly. Some striped of hyperpigmentation.
48	Synophrys. Ptosis. Coarse face. Prominent chin. Short broad neck. Tapering fingers. Hypermetropia. Amblyopia in the left eye. Talipes of the left foot.
49	Hypertelorism. Pre-auricular ear pit. Flat nasal bridge. Talipes.
50	A few words at 10years, mainly signs. Ears have over folded helices. Prominent central upper incisors. Curved toes. Tapering fingers.
51	Hyperactive, reduced attention span. MRI-Bilateral periventricular hyperintense signals in T2, notably in trigonum. Severe speech and language delay, spoke only few words at age of 2 years and 10 months, high pitched voice. Hypertelorism, strabismus, small palpebral fissures, periorbital fullness. Open appearance, full lips, tended upper vermilion. High arched palate. Large upper incisors. Microretrogenia, clinodactyly digit V both hands. Tube feeding in neonatal period, at age of 2 years and 10 months only pureed food. Severe failure to thrive in first 2 years of life. Hyperopia. Biochemical abnormalities; Elevated lactate and glycine in CSF, elevated glutamine, arginine and ornithine in blood, elevated GAG in urine.
52	Slightly increased nuchal translucency. Postnatal apnea-bradycardia syndrome. Delayed myelination seen on MRI. Ataxic movement pattern. Head control at 1 year, rolls and crawls, still not able to sit unsupported at the age of 37 months. Vocalises, no words at 3 years. Secondary microcephaly, brachycephaly. Discrete protrusio bulbi, swollen eyelids, intermittent esotropia. High-arched palate. Prominent forehead, increased intermamillar distance. ASD (surgical procedure planned). Mitral valve prolapse (surgical procedure planned), enlarged right ventricle. Prominent renal pelvis. Shawl scrotum. Bilateral inguinal hernias requiring surgery. Myopia (-6 dioptres), astigmatism. Frequent upper respiratory tract infections. Hyper-responsive bronchial system, bronchial asthma, impaired mucociliary clearance, bronchomalacia.
T1	Craniosynostosis. Large, low set, posteriorly rotated ears. Patent ductus arteriosus. Atrial septal defect. Clinodactyly. Widely spaced nipples/
T2	Craniosynostosis. Large, low set posteriorly rotated ears. Patent ductus arteriosus. Atrial septal defect. Pre-auricular pits. Required gastrostomy.
T3	Sudden death at 10 years. Recurrent craniosynostosis. Large, low set posteriorly rotated ears. Patent ductus arteriosus. Patent foranum ovale. Required gastrostomy. Seizures with EEG changes.
T4	Low set posteriorly rotated ears. Nasolacrimal stenosis. Patent ductus arteriosus, atrial septal defect, ventricular septal defect. Hypospadias. Pre-auricular pits. Widely spaces nipples. Peg shaped teeth. Bilateral grade 2 brain haemorrhages at 7 days which fully resolved. Large anterior fontanel.
T5	Atrial septal defect. Patent ducus arteriosus. Suspected cortical visual impairment. Epicanthal folds. Clinodactyly. Left single palmer crease.
T6	Plagiocephally. Suspected cortical visual impairment. Bilateral single palmar creases. Low set posteriorly rotated ears. Required gastrostomy. Transient infantile spasms which responded to ACTH.
T7	Posteriorly rotated ears. Hypermetropia. Crowded abnormally shaped teeth.
A1	Sleep apnoea. Epicanthal fold. Atrial septal defect. Broncho- or laryngomalacia. Chronic lung disease. Dystonia. Required gastrostomy. Recurrent aspiration pneumonias. Brachydactyly.

A2	Cleft palate. Large upper canines. Ventricular septal defect. Intestinal malrotation. Hydronephrosis. Left hand flexed PIP. Bronchomalacia. Recurrent urinary tract, ear and respiratory infections. Autism.
A3	Bifid tip to the nose. Small peg shaped lower teeth. Epicanthic folds. Optic nerve atrophy. Cryptorchidism. Brachydactyly.
A4	Myopia. Atrial septal defect. Dystonia. Chronic ear infections requiring grommets.
M1	ASD, mild mitral valve prolapse. Failure to thrive. Coarse facial features. Small dysplastic ears with a right pre-auricular sinus. Supernumerary left nipple.
M2	Right sided cryptorchidism requiring orchiopexy. Recurrent otitis media with eardrum perforation. Chronic cough. Large posteriorly rotated ears. Narrow palate. Mitral valve prolapse with regurgitation.
M3	Two early episodes of febrile illnesses associated with poor feeding and developmental regression. Hypotelorism.
M4	Patent foranem ovale and Patent Ductus Arteriosus. Laryngomalacia. Micrognathia.
M5	Stereotyped behaviours. Flattened midface. Recessed hairline. Hypertelorism and downward slanting palpebral fissures. Protruding tongue. Widely spaced teeth. Small ears with thickened helices.
M6	Absence of olfactory bulb. Autism. Obsessive compulsive behaviours. Epilepsy (complex partial seizures with secondary generalisation)
GV1	Severe intermittent isolated congenital neutropenia without recurrent infections. Polyhydramnios. Opposition, withdrawal, and anxiety disorder. Prominent forehead, low-set ears, narrow mouth with an upper labial frenulum, and microretrognathia. Brittle nails. Dental abnormalities (bifid incisors). Anisometropia and astigmatism. MRI showed benign enlargement of the pericerebral areas.
ZS1	Multiple pituitary abnormalities including growth hormone and adrenal insufficiency, and central hypothyroidism. Small anterior pituitary lobe, absent pituitary stalk and ectopic posterior pituitary lobe. Note the presence of two paternally inherited rare variants in two genes that are associated with pituitary pathology. BMP4 (p.R269_L272del) and GLI3 (p.Glu1294Argfs*19). Small mouth with dental crowding. Gastric tube feeding.
MU1	
E1	Multiple allergies. Small for gestational age. Dysphagia. ASD and PDA. Cortical visual impairment, nystagmus, anisocoria and myopia. Blocked tear duct.
S1	Persistent pulmonary hypertension at birth. Scaphocephaly. Micrognathia. Low set ears. Dental Malocclusions. Myopic astigmatism. Cryptorchidism. Inguinal hernia.
S2	Scaphocephaly. Micrognathia. Low set ears. Dental Malocclusions. Mild hearing loss. Repeated hospital admission in infancy with pulmonary infections.
S3	Scaphocephaly. Micrognathia. Low set ears. Laryngomalacia. Atrial septal defect.

Supplemental Table 4: Cardiac lesions found in patients.

Patient 3	ASD not requiring surgery
Patient 5	PDA at 7 years of age
Patient 6	mild pulmonary stenosis
Patient 10	ASD and PDA and underwent surgery at 2 years of age
Patient 12	VSD not requiring surgery
Patient 13	mild left pulmonary artery stenosis
Patient 18	ASD and PDA not requiring surgery
Patient 19	ASD which required surgery and mild tricuspid and mitral regurgitation
Patient 20	large ASD, Pulmonary valve stenosis, Cardiomyopathy
Patient 22	PFO resolved
Patient 23	PDA
Patient 24	ASD requiring surgical repair
Patient 25	ASD requiring surgery
Patient 26	PDA required closure at 12 mths, hypoplastic left pulmonary vein leading to severe pulmonary hypertension with chronic lung disease
Patient 27	ASD, VSD, PDA required surgery
Patient 31	ASD surgically repaired, VSD not requiring surgery
Patient 33	ASD
Patient 34	PFO resolved
Patient 36	VSD, no treatment
Patient 42	ASD requiring surgery
Patient 43	ASD, PDA, Mitral Regurgitation
Patient 44	ASD
Patient 46	PDA requiring ligation and large ASD requiring surgery
Patient 52	ASD, surgical procedure planned
E1	ASD requiring surgery
M1	ASD, mitral valve prolapse
M2	mitral valve prolapse
M4	PFO and PDA
A1	large ASD, repaired at 5yrs
A2	VSD
A4	ASD
T1	PDA, ASD/surgery
T2	PDA, ASD/Catheter
T3	PDA, PFO/spontaneous closure
T4	PDA, ASD, VSD/Surgery
T5	ASD sec, PDA/Catheter
MU1	ASD
S3	ASD

CHD = 38, ASD = 25, VSD = 6, PDA = 14. 36 without known cardiac abnormality



No cardiac issues

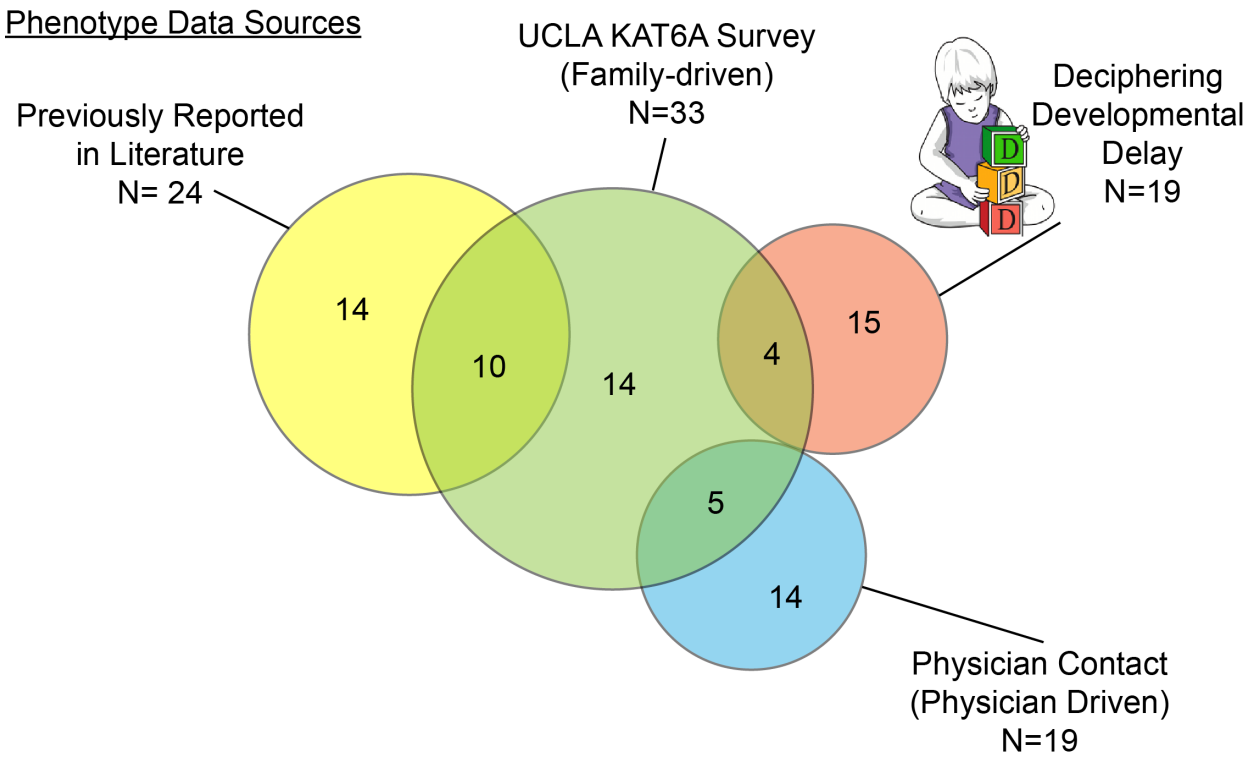
Patients 1,2,4,7,8,9,11,14,15,16,17,21,28,29,30,32(heart murmur, no treatment ?ECHO),35,37,38, 39, 40, 41, 45,47,48,49, 50,51 GV1,M3,M5,M6,A3, T6, T7 ZS1

Supplemental Table 5: ID numbers for patients in our co-hort that are registered on decipher (this includes many patients recruited through the DDD study).

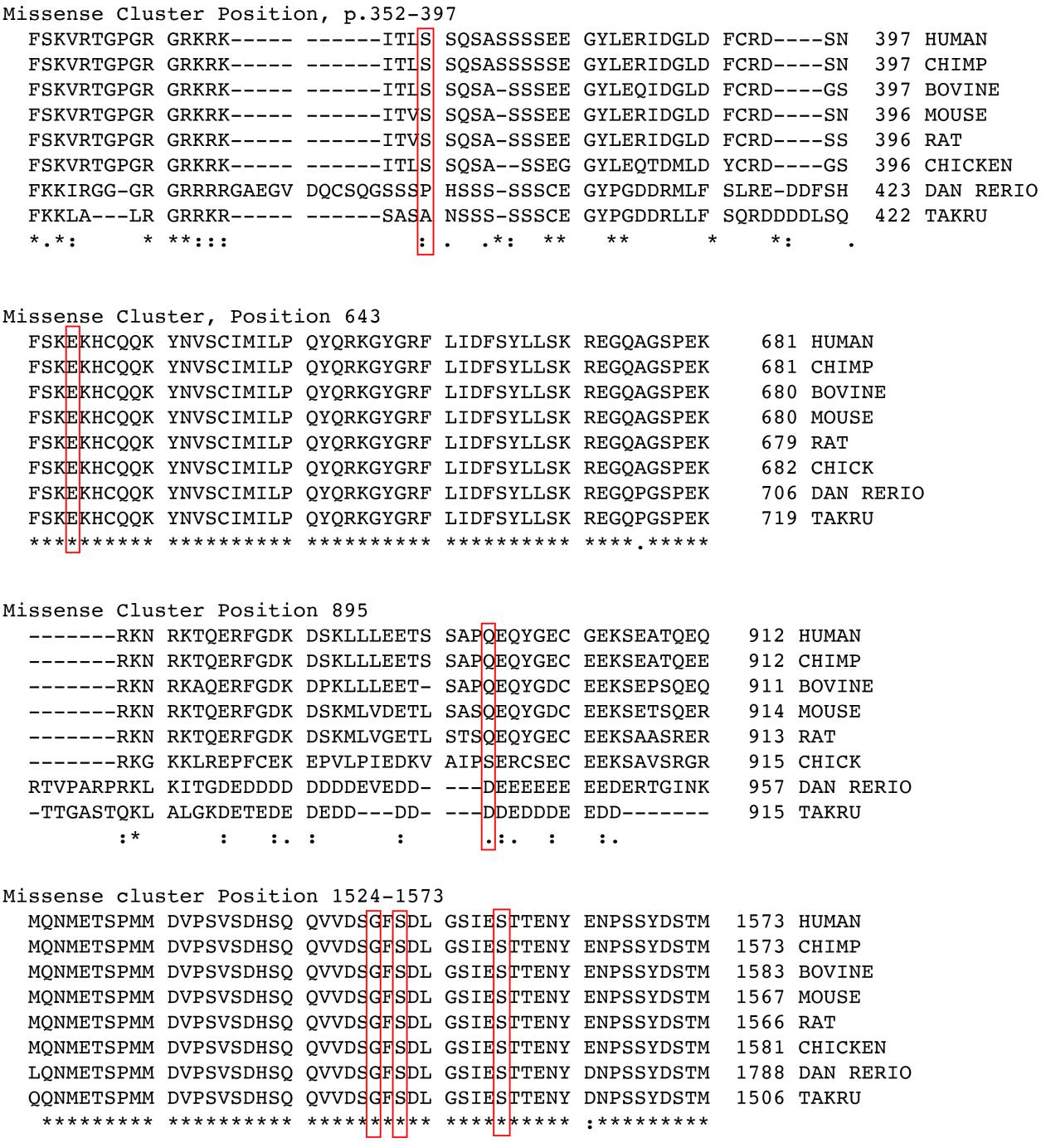
Patient Number	Decipher ID
1	259417
2	261878
3	264529
4	273922
5	276224
6	279139
7	332285
8	336836
9	279703
10	273855
11	285727
15	259354
16	281174
17	290996
45	291405
46	293926

Supplementary table 6 – Comparison between the prevalence of features for the previously reported and newly reported patients

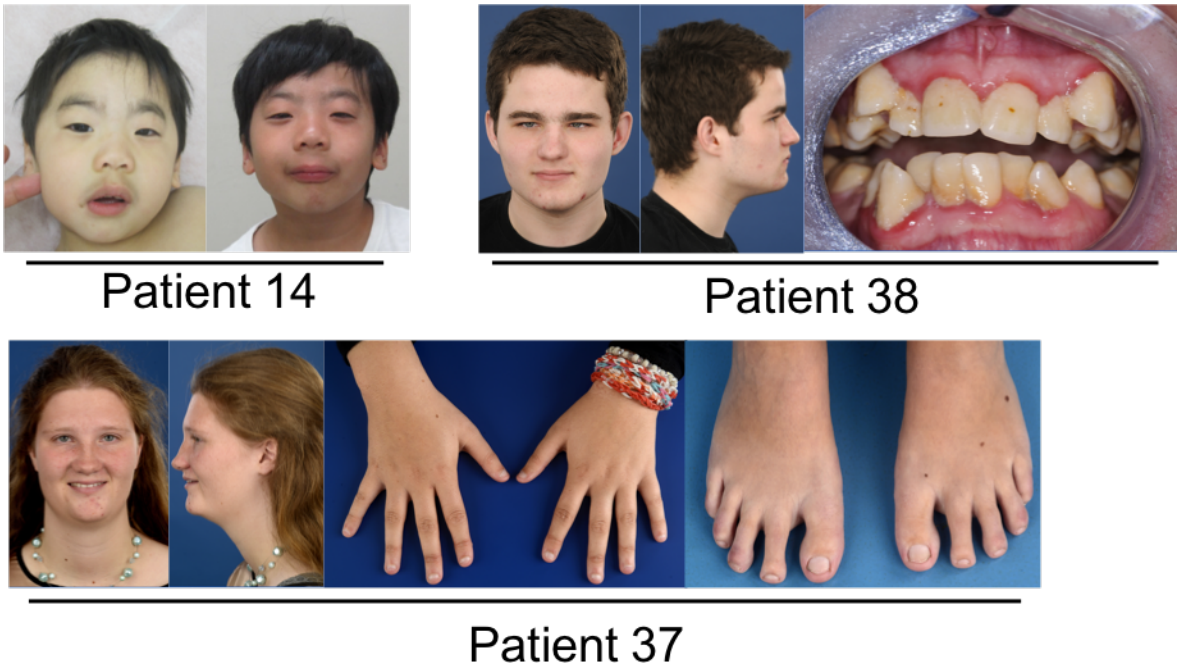
Feature	Previously reported cases (24)	Newly reported cases (52)	Total cases (76)
Sex	F=13 M=11	F=24 M=28	F=37 M=39
Mutation type	Fs=8 n=13 m=1 s=1 del=1	Fs=21 n=23 m=5 s=3	Fs=29 n=36 m=6 s=4 del=1
SGA	17% (3/18)	15% (7/48)	15% (10/66)
Microcephaly	42% (10/24)	26% (12/46)	31% (22/70)
Presence of ID	100% (24/24)	100% (45/45)	100% (69/69)
Neonatal Hypotonia	92% (22/24)	69% (35/51)	76% (57/75)
Seizures	13% (3/23)	8% (4/51)	9% (7/74)
Speech delay	100% (23/23)	100% (48/48)	100% (71/71)
Strabismus	52% (12/23)	54% (26/48)	54% (38/71)
Ptosis	29% (7/24)	10% (5/49)	16% (12/73)
Visual Defect	64% (9/14)	63% (29/46)	63% (38/60)
Broad nasal tip	100% (19/19)	80% (38/48)	85% (57/67)
Thin upper lip	89% (16/18)	58% (26/45)	67% (42/63)
Feeding difficulties	81% (17/21)	76% (39/51)	78% (56/72)
Reflux	75% (12/16)	55% (27/49)	60% (38/60)
Constipation	43% (3/7)	52% (24/46)	51% (27/53)
Congenital heart defect	64% (14/22)	46% (24/52)	51% (38/74)
Frequent Infection	Reported in 6	47% (23/49)	47% (23/49)*
Behavioural problems	Reported in 3	39% (14/36)	39% (14/36)*
Sleep disturbance	Reported in 3	37% (17/46)	37% (17/46)*



Supplemental Figure 1: Data collection sources. Multiple parallel data collection methods allowed for multiple sources of phenotypic data per individual. Several patient were identified through duplicate methods, however we were able to collect sufficient information to identify these duplicates and merge their records.



Supplemental Figure 2: Amino acid conservation across multiple species at locations with missense mutations in KAT6A, outlined by the red boxes, show that these residues tend to be highly conserved and are indicated by an asterisk. All *KAT6A* missense mutations affect highly conserved residues. A colon indicates conservation between groups of strongly similar properties, and a period indicates conservation between groups of weakly similar properties.



Supplemental Figure 3: We included three additional photos of patients with KAT6A syndrome here.

Supplemental Methods for cDNA Sequencing for confirmation of functional effect of splice changes in Patient 17.

Agarose gel electrophoresis of *KAT6A* amplicons were generated for both the Patient 17 with the *KAT6A* heterozygous variant c.1364-2A>T and an unaffected familial control displayed bands equivalent to the expected size (377bp) of the WT transcript (NM_006766.4) in both instances. Sequencing of total PCR for amplicons generated for the unaffected familial control confirmed this band to be homologous to the WT *KAT6A* sequence. A secondary band was visible in amplicons generated for the affected c.[1364-2A>T];[=] patient, sequencing of total PCR displayed this to represent a transcript with a deletion of the first 8bp of exon 8. Due to the high homology displayed between the WT and mutant transcript it is likely that the upward location of the 8bp shorter mutant transcript is due to heteroduplex formation between the WT and mutant transcript.

Supplmental Methods for RNA extraction.

Nucleic acid extraction

Peripheral blood was supplied in PAXgene (Preanalytix-Qiagen) RNA stabilisation tubes. RNA was extracted using an RNeasy mini kit (Qiagen) and an automated protocol on the Qiacube ® (Qiagen) as according to the manufacturer's instructions. RNA was stored at -80 °C.

cDNA synthesis

cDNA was synthesised using a High Capacity cDNA RT kit (Thermo Fisher Scientific) in a total volume of 50 µL, with the addition of n of 1.0 U/µl RNA inhibitor, as according to the manufacturers instruction's. cDNA was stored at -20 °C.

PCR and sequencing

All PCR was conducted using Phusion® DNA polymerase as according to the manufacturer's instructions; annealing temp 58°C, 15 s extension (*KAT6A* fwd 5' GATGGCTTGGACTTCTGCAG 3', *KAT6A* rev 5' TCAATGACAGAGGGACAGCG 3'. Primers were designed using Primer3 (http://primer3.ut.ee/) and SNP checked using SNPcheck3 (NGRL Manchester). Total PCR was sequenced using the BigDye® Terminator kit v3.1 (Applied Biosystems) and analysed using an ABI 3730 DNA sequencer (Applied

Biosystems). Sequence analysis was conducted using Mutation Surveyor® (SoftGenetics) and the Sequence Scanner Software 2.0 (Applied Biosystems). PCR amplicons were run on a 3% agarose gel (50:50 Nusieve (Lonza): Agarose MP (Roche)) and 1x TBE buffer at 90v for 120 minutes. Amplicons were sized against 5µl of 50bp GeneRuler ladder (Thermo Fisher Scientific).

REFERENCES CITED

1. Tham, E., et al., *Dominant mutations in KAT6A cause intellectual disability with recognizable syndromic features.* Am J Hum Genet, 2015. **96**(3): p. 507-13.
2. Arboleda, V.A., et al., *De novo nonsense mutations in KAT6A, a lysine acetyl-transferase gene, cause a syndrome including microcephaly and global developmental delay.* Am J Hum Genet, 2015. **96**(3): p. 498-506.
3. Millan, F., et al., *Whole exome sequencing reveals de novo pathogenic variants in KAT6A as a cause of a neurodevelopmental disorder.* Am J Med Genet A, 2016. **170**(7): p. 1791-8.
4. Gauthier-Vasserot, A., et al., *Application of whole-exome sequencing to unravel the molecular basis of undiagnosed syndromic congenital neutropenia with intellectual disability.* Am J Med Genet A, 2017. **173**(1): p. 62-71.
5. Murray, C.R., et al., *Novel Causative Variants in DYRK1A, KARS, and KAT6A Associated with Intellectual Disability and Additional Phenotypic Features.* J Pediatr Genet, 2017. **6**(2): p. 77-83.
6. Elenius, V., et al., *Food allergy in a child with de novo KAT6A mutation.* Clin Transl Allergy, 2017. **7**: p. 19.
7. Zwaveling-Soonawala, N., et al., *Variants in KAT6A and pituitary anomalies.* Am J Med Genet A, 2017. **173**(9): p. 2562-2565.
8. Chisei Satoh, R.M., Akira Kinoshita, Hiroyuki Mishima, Michiko Doi, Mutsuko Miyazaki, Masafumi Fukuda, Haruo Takahashi, Tatsuro Kondoh & Koh-ichiro Yoshiura, *Three brothers with a nonsense mutation in KAT6A caused by parental germline mosaicism.* Human Genome Variation, (4): p. 17045.