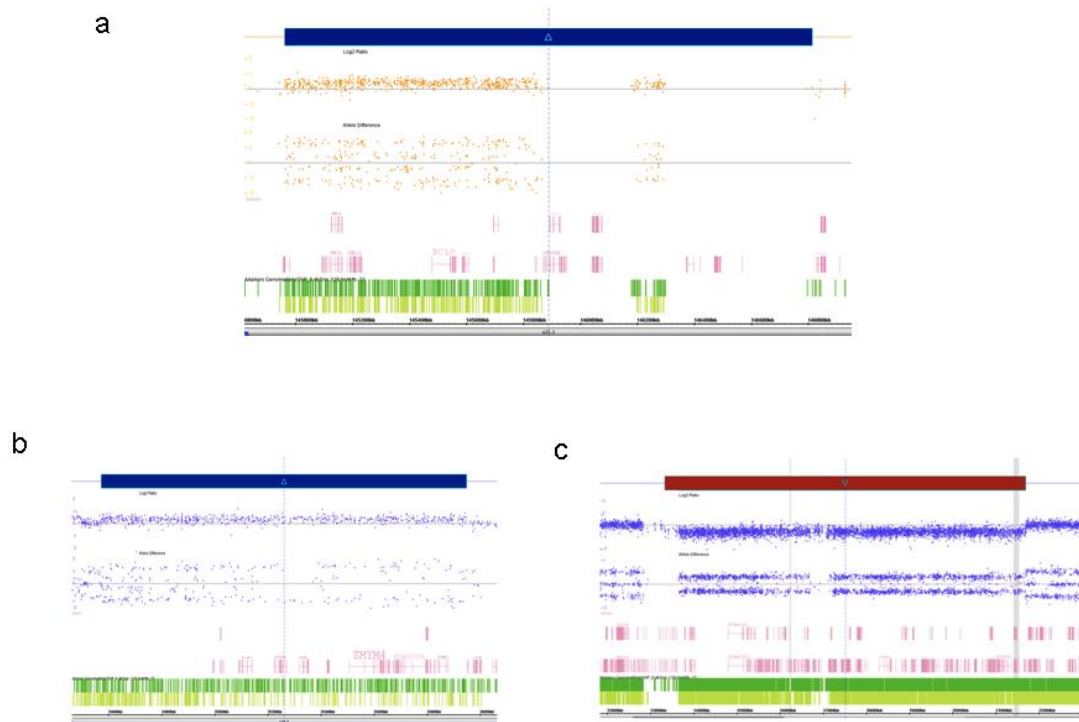


**The American Journal of Human Genetics, Volume 89  
Supplemental Data**

**Whole-Exome-Sequencing Identifies Mutations  
in Histone Acetyltransferase Gene *KAT6B* in Individuals  
with the Say-Barber-Biesecker Variant of Ohdo Syndrome**

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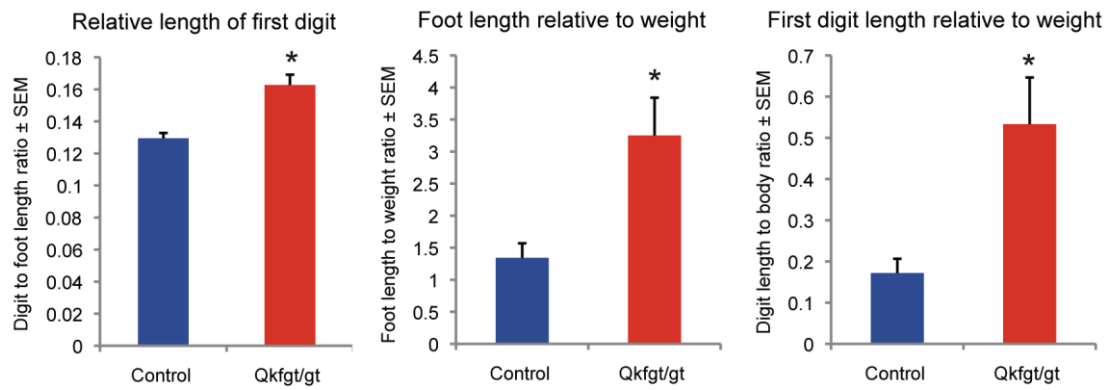
### Figure S1. Microarray Results of Individuals 3 and 17

(A) Individual 3 was shown to have a duplication of ~2Mb within the long arm of chromosome 1 at band q21.1, flanked by markers CN\_436418 and CN\_004940 (nucleotides chr1:144643813-146814404 based on Ensembl Build 36) This duplication was subsequently confirmed using BAC probe RP11-533N14 , with parental FISH studies showing a normal hybridisation pattern.

(B) Deletion of ~8.3Mb from band p36.12 to p36.21, flanked by markers CN\_450710 and CN\_483670 (nucleotides chr1: 13339380-21692325 based on Ensembl Build 36) identified in Individual 17

(C) Duplication of ~1.3Mb within band 1p34 in Individual 17, flanked by markers CN\_488937 and CN\_022530 (nucleotides chr1: 34572912-35949096)

Both changes were confirmed by fluorescent in-situ hybridisation (FISH) using the BAC probes RP11-174G17 and RP11-244H3 respectively.



**Figure S2. Demonstration of Disproportionately Long First Digits of Hind Limbs of  $Qkf^{gt/gt}$  Mouse Compared to Wild-Type**

Images were taken of hindfeet of mice and measurements were taken from these images. Data were presented as means  $\pm$  SEM of biological replicates, i.e. measurements taken from individual mice ( $n = 3-4$   $Qkf^{gt/gt}$  and 3-4 controls), and analysed by two-sided T-test using Stata Intercooled 10.0 software

**Table S1. Clinical Features of All Individuals Tested**

Individual	1	2	3	4	5	6	7	8	9
Sex	M	F	F	M	M	F	M	F	F
Typical face	+	+	+/-	+	+	+	+	+	+
Birth weight %	50	75	24	0.4	50	50	n/k	9	50
Hypotonia	+	+	+	+	+	+	+	+	+
Learning Disability	+	+	+	+	+	+	+	+	+
Feeding problems	+	+	+	+	+	+	+	+	+
Dental anomalies	+	+	+	n/k	+	+	+	+	+
Long thumbs	+	+	-	+	+	+	-	+	+
Long great toes	+	+	-	+	+	+	-	+	+
Hearing problem	-	+	+	+	+	-	+	+	+
Abnormal patella	-	+	-	+	+	-	-	+	-
Lacrimal duct abn.	-	+	+	+	+	+	+	+	+
Cleft palate	-	-	-	+	-	-	+	+	+
Thyroid abnormal	-	-	+	+	-	+	+	+	-
Heart Defect	-	-	-	+	+	+	+	+	+
Cryptorchidism	+	n/a	n/a	+	+	n/a	+	n/a	n/a
Hypospadias	+	n/a	n/a	+	-	n/a	+	n/a	n/a
Height %	50	n/k	25	25	50	9	n/k	n/k	2
Weight %	25	n/k	50	25	50	75	n/k	n/k	9
OFC%	<3	n/k	9	2	2	91	n/k	n/k	2-9

% = percentile

n/a = not applicable

n/k = not known

OFC= occipito-frontal circumference

<b>Individual</b>	<b>10</b>	<b>11</b>	<b>12</b>	<b>13</b>	<b>14</b>	<b>15</b>	<b>16</b>	<b>17</b>	<b>18</b>	<b>19</b>
<b>Sex</b>	<b>F</b>	<b>M</b>	<b>F</b>	<b>F</b>	<b>M</b>	<b>M</b>	<b>M</b>	<b>F</b>	<b>M</b>	<b>M</b>
<b>Typical face</b>	<b>+</b>	<b>+/-</b>	<b>+</b>	<b>+</b>	<b>+/-</b>	<b>+/-</b>	<b>+/-</b>	<b>+/-</b>	<b>+/-</b>	<b>+/-</b>
<b>Birth weight %</b>	<b>9</b>	<b>50</b>	<b>50</b>	<b>n/k</b>	<b>95</b>	<b>75</b>	<b>25</b>	<b>&lt;10</b>	<b>50</b>	<b>75</b>
<b>Hypotonia</b>	<b>+</b>	<b>+</b>	<b>+</b>	<b>+</b>	<b>+</b>	<b>-</b>	<b>+</b>	<b>-</b>	<b>-</b>	<b>+</b>
<b>Learning Disability</b>	<b>+</b>	<b>+</b>	<b>+</b>	<b>+/-</b>	<b>+</b>	<b>+</b>	<b>+</b>	<b>+</b>	<b>+</b>	<b>+</b>
<b>Feeding problems</b>	<b>+</b>	<b>+</b>	<b>+</b>	<b>+</b>	<b>+</b>	<b>+</b>	<b>+</b>	<b>+</b>	<b>-</b>	<b>+</b>
<b>Dental anomalies</b>	<b>+</b>	<b>-</b>	<b>+</b>	<b>+</b>	<b>+</b>	<b>+</b>	<b>-</b>	<b>-</b>	<b>-</b>	<b>+</b>
<b>Long thumbs</b>	<b>+</b>	<b>-</b>	<b>+</b>	<b>-</b>	<b>-</b>	<b>-</b>	<b>-</b>	<b>+</b>	<b>-</b>	<b>-</b>
<b>Long great toes</b>	<b>+</b>	<b>-</b>	<b>+</b>	<b>-</b>	<b>-</b>	<b>-</b>	<b>-</b>	<b>-</b>	<b>+</b>	<b>-</b>
<b>Hearing problem</b>	<b>-</b>	<b>-</b>	<b>-</b>	<b>-</b>	<b>-</b>	<b>+</b>	<b>-</b>	<b>-</b>	<b>-</b>	<b>+</b>
<b>Patellar abnormality</b>	<b>-</b>	<b>-</b>	<b>-</b>	<b>-</b>	<b>-</b>	<b>-</b>	<b>-</b>	<b>-</b>	<b>-</b>	<b>-</b>
<b>Lacrimal duct abn.</b>	<b>-</b>	<b>-</b>	<b>-</b>	<b>-</b>	<b>-</b>	<b>-</b>	<b>-</b>	<b>-</b>	<b>-</b>	<b>-</b>
<b>Cleft palate</b>	<b>-</b>	<b>-</b>	<b>+</b>	<b>-</b>	<b>+</b>	<b>-</b>	<b>-</b>	<b>-</b>	<b>-</b>	<b>-</b>
<b>Thyroid abnormality</b>	<b>-</b>	<b>-</b>	<b>-</b>	<b>+</b>	<b>-</b>	<b>-</b>	<b>-</b>	<b>-</b>	<b>-</b>	<b>-</b>
<b>Heart Defect</b>	<b>-</b>	<b>-</b>	<b>-</b>	<b>-</b>	<b>-</b>	<b>-</b>	<b>-</b>	<b>-</b>	<b>+</b>	<b>-</b>
<b>Cryptorchidism</b>	<b>n/a</b>	<b>+</b>	<b>n/a</b>	<b>n/a</b>	<b>+</b>	<b>+</b>	<b>n/k</b>	<b>n/a</b>	<b>+</b>	<b>-</b>
<b>Hypospadias</b>	<b>n/a</b>	<b>-</b>	<b>n/a</b>	<b>n/a</b>	<b>-</b>	<b>+</b>	<b>-</b>	<b>n/a</b>	<b>-</b>	<b>-</b>
<b>Height %</b>	<b>2-9</b>	<b>25-50</b>	<b>50</b>	<b>25</b>	<b>50</b>	<b>25</b>	<b>25</b>	<b>9</b>	<b>n/k</b>	<b>&lt;25</b>
<b>Weight %</b>	<b>50</b>	<b>98</b>	<b>75</b>	<b>25</b>	<b>50</b>	<b>n/k</b>	<b>25</b>	<b>2</b>	<b>2-9</b>	<b>n/k</b>
<b>OFC%</b>	<b>50-75</b>	<b>25-50</b>	<b>&lt;50</b>	<b>25</b>	<b>91</b>	<b>75</b>	<b>&lt;0.4</b>	<b>&lt;0.4</b>	<b>2</b>	<b>&lt;9</b>

**Table S2. Primer Sequences for *KAT6B***

Exon	Forward	Reverse	Product bp
3_1	TGCTTAATACAGTCTGGAATACTCTG	TCGGCTCCTCAAGTCCTTC	493
3_2	TTTCCTAAGTCAGCCAAGGG	CCACATAAAACTTAAGTGAAACCG	437
4	CTGCACTACAACCTGGCG	CCCACTGAATTACCAATGCTG	595
5 and 6	CATTGGCTAGCCTCATCAGC	GAACCTGTATACCTCTGAGCTGTC	634
7	TTGAGCAGCTTATTACAGCTACAAC	GGAATCTGAGAGAATAACAATCACC	301
8_1	GAGAAACATCTAGGTGGTGGC	GGCACACTGCACTGGCTGG	668
8_2	CCACACAAAAGCTAAAACCTCC	ACCTATGTGACAAATAGGAATTGA	638
9	TCCCTATTTGCCCAGTATGC	TCACAAAGGATGTTGAAAGGG	337
10	TTGATAGTCACTGGTGAAAGAACC	TCCCTAATTCCTGCTTTAGAATC	283
11	AAATTGAACAATATTTAATCTTCCCC	TTTGTACCTTCTAAACACAAATCAGC	309
12	GTTTAGGATTTGGAAATAAAATGAT	AGCCCTGTTCAAAGAATTGG	351
13	TACAAGGACAGTGGCAGGTG	CTCAATTCCTAGAGTACAATCACTG	378
14	GCGAATGCACTTCTCTGTTG	TGAATGCACCTATCTCCAGG	433
15	CATTAGTGCTAGCATATGTCCG	GACAACGATCTTAATGACTTTCTTAGC	330
16	GAGACACTTTGCCATTGATCC	AATAGACAGAATGTCTGCAATGAC	509
17	CATGTCTACTGCATATCGACTCAAC	CAGAGGCCTGCTCTTTGG	442
18_1	GGAATGAATTCAAGTTGCC	GTGATCATCCAAACGTGCAG	637
18_2	GATGATCTCATCAAACCTGAGG	TCTGCACGGGTGTAGTTCTG	694
18_3	CCGTTCAAGTCTTTGACCCAG	AGCCTTGAGGAGACTTGACG	630
18_4	CCTCCAGCAGTCTGACACAG	GCTGGCAATCTGGGTTTG	629
18_5	CTCCAATGAATCTGCCGC	AATTGCTGGTTTTGAAATCG	656
18	TGACGAGCAGGTAACAGTGG	GTTGTGGGATGGCTCTTCAT	415