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Supplemental Data

Mutations in *KAT6B*, Encoding a Histone

Acetyltransferase, Cause Genitopatellar Syndrome

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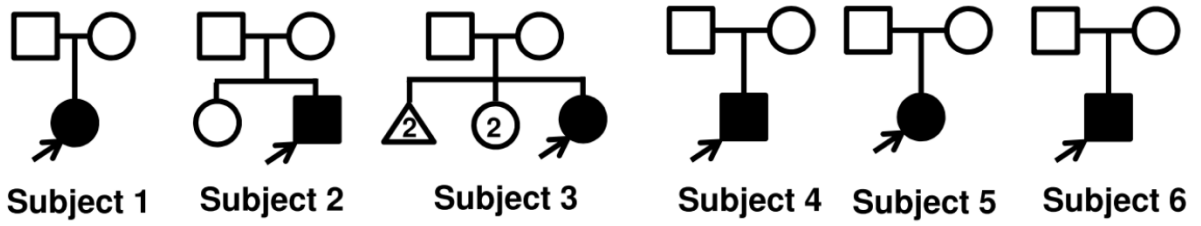


Figure S1. Pedigrees

Pedigrees of the families included in this study.

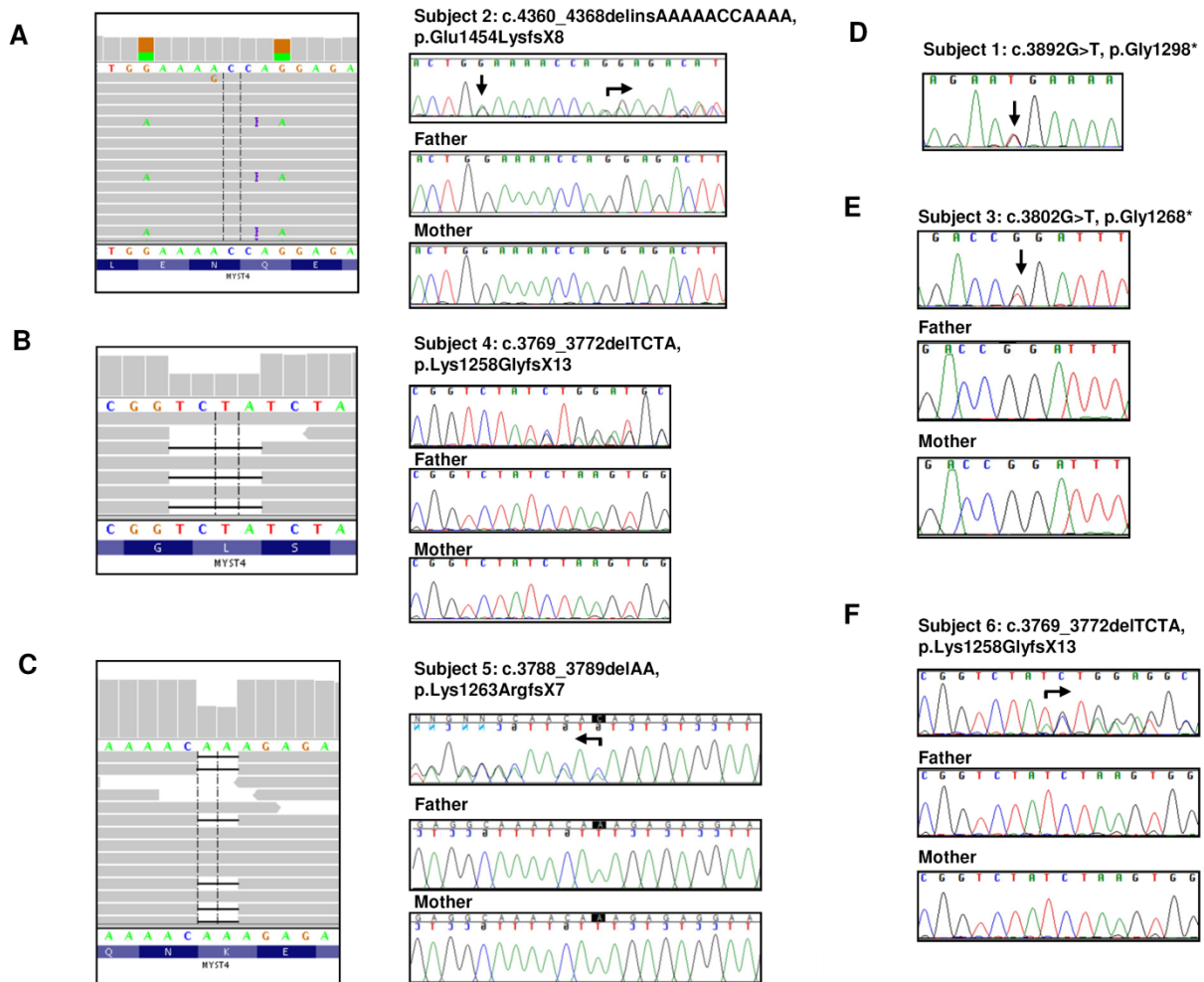


Figure S2. Confirmation of Mutations

KAT6B mutations by exome sequencing in subjects 2 (A), 4 (B), and 5 (C). Confirmations by Sanger sequencing for these three and subjects 1 (D), 3 (E) and 6 (F). Sanger sequencing confirmed all mutations detected by exome sequencing, and in all cases where parental samples were available, the parents did not carry the mutation.

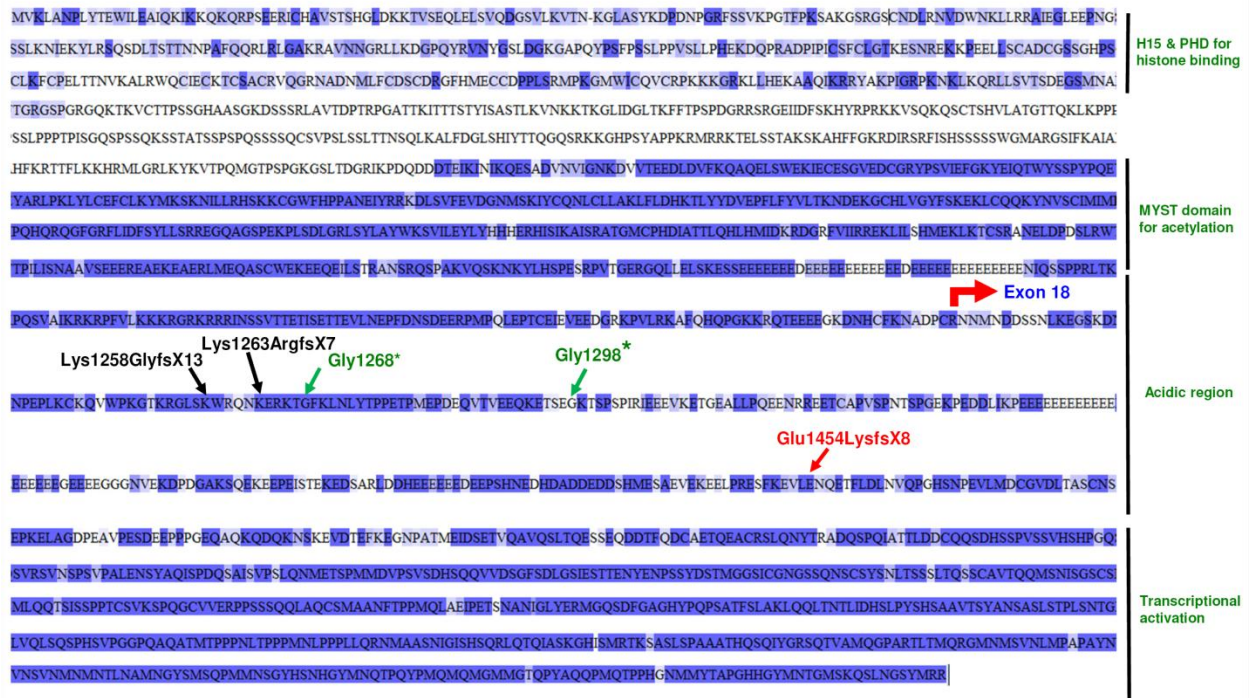


Figure S3. KAT6B Sequence Conservation

The amino acid sequence of human KAT6B (Uniprot Q8WYB5) is shown along with conservation information. The homologues from *Mus musculus* (Q8BRB7), *Bos taurus* (F1MF5), *Macaca fascicularis* (Q8WML3) and *Canis familiaris* (F1PWC5) were aligned to the human protein using JalView. Perfectly conserved residues are shown in dark blue. The mouse isoform in Uniprot does not contain amino acids corresponding to human amino acids 482-668, and the *Bos taurus* and *Macaca fascicularis* isoforms do not contain amino acids corresponding to human amino acids 373-668, which explains the large region between the PHD and MYST domains without conservation. The mutation locations are illustrated with arrows as in Figure 3.

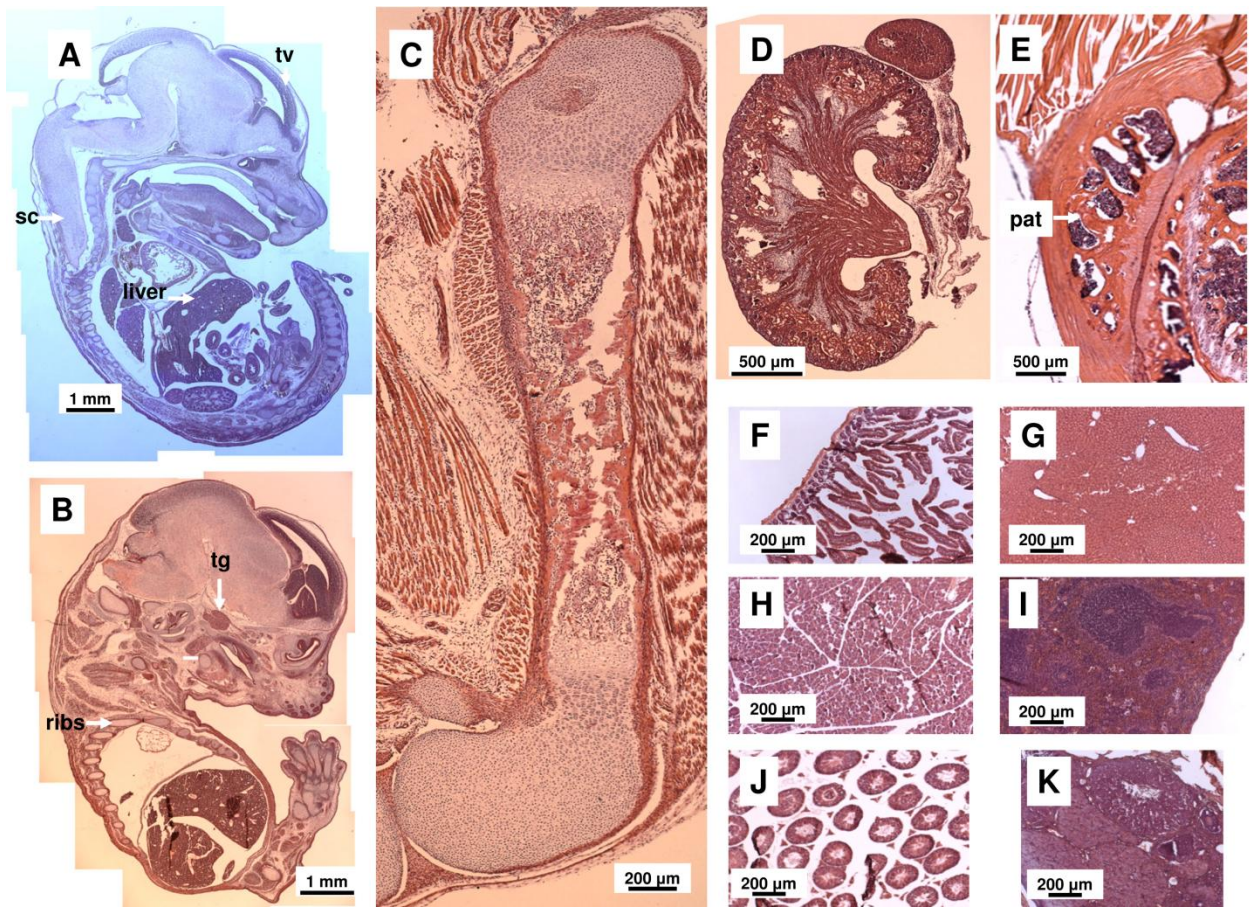


Figure S4. Tissues Corresponding to Figure 5

Haematoxylin and eosin staining of tissues corresponding to the tissues shown in Fig. 5. Tissues shown are: (A and B) whole embryos at E15.5, (C) femur at P1, and (D) kidney at P1. The other tissues are at 8 weeks of age: (E) patella, (F) duodenum, (G) liver, (H) pancreas, (I) spleen, (J) testis, and (K) ovary.

Table S1. Primers Used in This Study

Purpose of primer	Primer name	Sequence
Genomic DNA sequencing	hMYST4ex3_1F	AGTTCTGTAAATACAAAGAGAACCTC
	hMYST4ex3_1R	TCGGCTCCTCAAGTCCTTC
	hMYST4ex3_2F	TTTCCTAAGTCAGCCAAGGG
	hMYST4ex3_2R	CCACATAAACTTAAGTGAAACCG
	hMYST4ex4F	ctgcactacaacctggcg
	hMYST4ex4R	AGCATGAAGCTATGATAATGGG
	hMYST4ex5-6F	CATTGGCTAGCCTCATCAGC
	hMYST4ex5-6R	GAACCTGTATACCTCTGAGCTGTC
	hMYST4ex7F	TACAGGCATCCTTGAGCAGC
	hMYST4ex7R	AAACCCCTAAAGGAATCTGAGAG
	hMYST4ex8_1F	TGCCTTTCCAACAGAGAAAC
	hMYST4ex8_1R	ACTGCACTGGCTGGAAGAAC
	hMYST4ex8_2F	TCCACCTTCTTCACTTCCAC
	hMYST4ex8_2R	TTCAGGGTTGGGGAGAAAG
	hMYST4ex9F	TCCCTATTTGCCAGTATGC
	hMYST4ex9R	AAGGGATTCTCTATAAAATCAACACAG
	hMYST4ex10F	GGTGAAGAACC AAAATGTAAGTG
	hMYST4ex10R	AAAATCATCCCTAATTCCTGC
	hMYST4ex11F	AAATTGAACAATATTTAATCTTCCCC
	hMYST4ex11R	TCAGCACATAGTCAACAATTAGG
	hMYST4ex12F	GAAGAAACAATTTTATATGGTGCAG
	hMYST4ex12R	CCTGTTCAAAGAATTGGGCTC
	hMYST4ex13F	TACAAGGACAGTGGCAGGTG
	hMYST4ex13R	CTGTAACGGCCATCAGCTTC
	hMYST4ex14F	GGCTCCTGTTCTAAGCTCTGG
	hMYST4ex14R	ATCTCCAGGGACCCTTTCTG
hMYST4ex15F	CATTAGTGCTAGCATATGTCCG	

	hMYST4ex15R	AAGCTTTTGACAACGATCTTAATG
	hMYST4ex16F	GAGGCTCTGGCTGTGTAACCTG
	hMYST4ex16R	AATAGACAGAATGTCTGCAATGAC
	hMYST4ex17F	TGTCTACTGCATATCGACTCAACC
	hMYST4ex17R	CTCTTTGGCACCCAGTCAG
	hMYST4ex18_1F	TCAAGTTGCCCATGGTTATG
	hMYST4ex18_1R	TTGGCTTTTAGCACCATCTG
	hMYST4ex18_2F	TCATCAAACCTgaggaagagg
	hMYST4ex18_2R	CCTTGCTGTTCTTTTGGTCC
	hMYST4ex18_3F	TTGTAACAGTGAGCCCAAGG
	hMYST4ex18_3R	CCACTGTCTACGACTTGCTGTG
	hMYST4ex18_4F	TCAAAGTGCCATCTCAGTGC
	hMYST4ex18_4R	CCCCAAAATCACTCTGACCC
	hMYST4ex18_5F	CATGGCTGCTAACTTCACCC
	hMYST4ex18_5R	AGCGCCCATAGATTTGTGAC
	hMYST4ex18_6F	ACAGCCAAAGACTGCAAACC
	hMYST4ex18_6R	TTTGTGCAGATCAATCCAATAG
cDNA amplification and sequencing	hMYST4 cDNA seq F	GACGAAACCACAGTCAGTTGCCATAA
	hMYST4 cDNA seq R	CACCTGGTGATGTGTTTGGACTTA
RT-PCR	hMYST RT primer F	AATCCAATAGTGATGCCCCG
	hMYST RT primer R	TCACGGCTACATGAACACAG
	hMYST probe	5'FAM/ACTGCCAC/ZEN/GTTGTCTACCTTCTC/3'IBFQ
	hGAPDH RT primer F	CTTCAACAGCGACACCCA
	hGAPDH RT primer R	ATGAGGTCCACCACCCT
	hGAPDH probe	5'FAM/CATTGCCCT/ZEN/CAACGACCACTTTGT/3'IBFQ