

# Molecular genetic variation of animals and plants under domestication

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## Supplementary Information Appendix

**Table S1.** Molecular characterization of Mendel's pea genes

**Table S2.** Examples of structural variants associated with phenotypic traits in domestic animals

**Table S3.** Examples of evolution of alleles in domestic animals

### Literature cited Supplementary Information Appendix

1. M. Bhattacharyya, A. M. Smith, T. H. N. Ellis, C. Hedley, C. Martin, The wrinkled-seed character of pea described by Mendel is caused by a transposon-like insertion in a gene encoding starch-branching enzyme. *Cell* **60**, 115–122 (1990).
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**Table S1.** Molecular characterization of Mendel's pea genes

Gene	Trait	Phenotype	Gene Function	Mutation	Ref.
<i>R</i>	Seed shape	Round vs. Wrinkled	Starch branching enzyme 1	0.8-kb TE insertion	(1)
<i>LE</i>	Stem length	Tall vs. Dwarf	Giberellic acid 3- oxidase 1	G-to-A missense substitution	(2,3)
<i>I</i>	Cotyledon color	Yellow vs. Green	Stay-green gene	6-bp insertion	(4,5)
<i>A</i>	Seed/flower color	Purple vs. White	bHLH transcription factor	G-to-A splice donor substitution	(6)

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**Table S2.** Examples of structural variants associated with phenotypic traits in domestic animals

Species	Trait	Mutation	Gene(s)
Cattle	Color sidedness	492 kb translocation	<i>KIT</i> (7)
Chicken	Pea-comb	copy number expansion	<i>SOX5</i> (8)
	Rose-comb	7.4 Mb inversion	<i>MNR2</i> (9)
	Dark brown color	8.3 kb deletion	<i>SOX10</i> (10)
	Naked neck	~ 70 kb translocation	<i>BMP12</i> (11)
	Fibromelanosis	complex	<i>EDN3</i> (12)
	Blue eggshell	retroviral insertion	<i>SLCO1B3</i> (13)
	Henny feathering	retroviral insertion	<i>CYP19A1</i> (14,15)
	Feathered leg	17.7 kb deletion	<i>PITX1</i> (16, 17)
	Crest	195 bp duplication	<i>HOXC10</i> (18)
Dog	Hair ridge	133 kb duplication	<i>FGF3, FGF4, FGF18, ORAOV1</i> (19)
	Chondrodysplasia	retrogene insertion	<i>FGF4</i> (20)
	Wrinkles <sup>d</sup>	16.1 kb duplication <sup>d</sup>	<i>HAS2</i> (21)
	Amylase activity	~ 8 kb duplication	<i>AMY2B</i> (22)
	Polled <sup>e</sup>	11.7 kb deletion	<i>PISRT1, FOXL2</i> (23)
Goat	Coat color	copy number variation	<i>EDNRA</i> (24)
	Coat color	copy number variation	<i>ASIP</i> (25)
	Coat color	copy number variation	<i>KIT</i> (25)
	Greying with age <sup>f</sup>	4.6 kb duplication	<i>STX17, NR4A3</i> (26)
Horse	Tobiano white spotting	~ 40 Mb inversion	<i>KIT</i> (27)
	Leopard spotting, stationary night blindness	retroviral insertion	<i>TRPM1</i> (28)
	Non-dun color	1.6 kb deletion	<i>TBX3</i> (29)
	Dominant white color	several duplications	<i>KIT</i> (30)
Pigeon	Feathered leg	44 kb deletion	<i>PITX1</i> (31)
	Recessive red plumage	7.5 kb deletion	<i>SOX10</i> (31)
	Recessive red plumage	2.5 kb deletion	<i>SOX10</i> (31)
Rabbit	Dwarf	12.1 kb deletion	<i>HMGA2</i> (32)

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**Table S3.** Examples of evolution of alleles in domestic animals

Species	Phenotype	Gene	Allele	Mutation	Ref. <sup>d</sup>
Pig	Patch	<i>KIT</i>	<i>Patch</i>	a: 450-kb duplication	(34)
	Belt		<i>Belt</i>	b: three duplications <sup>a</sup>	(30,35)
	Dominant white		<i>Dominant white</i>	a + b + c: splice mutation	(30,36)
Pig	Dominant black	<i>MC1R</i>	<i>E<sup>D</sup></i>	a: missense mutation	(37)
	Black spotting		<i>e<sup>p</sup></i>	a + b: 2 bp insertion	(37)
Chicken	Dominant white	<i>PMEL</i>	<i>Dominant white</i>	a: 9 bp insertion	(38)
	Smoky		<i>Smoky</i>	a: + b: 12-bp deletion	(38)
Chicken	Rose-comb + low sperm motility	<i>MNR2</i> + <i>CCDC108</i>	<i>Rose1</i>	a: 7.2-Mb inversion	(9)
	Rose-comb		<i>Rose2<sup>b</sup></i>	b: non-homologous recombination	(9)
Chicken	Plumage color	<i>MC1R</i>	<i>Multiple</i>	Various combinations of missense mutations	(39)
Cattle	Color sidedness	<i>KIT</i>	<i>C<sub>S29</sub></i>	a: 492-kb translocation	(9)
	Color sidedness		<i>C<sub>S6</sub><sup>c</sup></i>	b: 575-kb translocation	(9)

<sup>a</sup>Not all of the three duplications have to be functionally important, but all three are exclusively found in domestic pigs showing white spotting.

<sup>b</sup>Originating from a recombination event between *Rose1* and a wild-type chromosome.

<sup>c</sup>Originating from a recombination event between *C<sub>S29</sub>* and a wild-type chromosome.

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