SUPPORTING INFORMATION

Supplementary Table 1

a. Primers used for amplification of BC004690:

Nucleotides 5118-5529

F1: 5’<TCATTCGTCTGGCCGAGATGG>3’

R1: 5’<GCTGCTGTCATTTCCGACCGAAG>3’

Nucleotides 4242-6368

F2: 5’<CAATTGGCATGTGAAAATAGAAGAA>3’

R2: 5’<AAAGACTGAGAATATTTCTCCTTTGAA>3’

b. Primers used for generating probe for Southern Blot

Nucleotides 4805 to 5072

F3: 5’<AAACTAAAAGAAAAAGAATCTGC>3’
R3: 5’<CTCTCTGGCCTTCTCCAGAA>3’

SUPPLEMENTARY TABLE 2

Analyzer Results Blood and Urine on *rd16*

BXD-14/Ty Male DOB 6/15/05 , Specimen drawn on 9/21/05

Serum

Test	Result	Normal Range
ALT (SGPT)	22	8-84 IU/L
Total Protein	5.4	6.9 - 7.1 g/dl
Albumin	3.4	3.8 - 4.4 g/dl
Calcium	9.5	10.4 - 12.0 mg/dl
Phosporous	7.0	11.9 - 13.1 mg/dl
Glucose	212	109 - 233 mg/dl
BUN	26	18 - 26 mg/dl
Cholesterol	86	85 - 129 mg/dl
T4	5.2	3.8 - 7.0 ug/dl
Magnesium	2.6	2.4 - 3.9 mg/dl*
Sodium	149.0	152.5 - 178.9 mmol/L
Potassium	6.18	5.06 - 7.50 mmol/L
Chloride	118	114.6 - 136.6 mmol/L
CO2	17.6	5.1 - 11.1 mmol/L

Urine

Test	Result	Normal Range
Sodium	143.1	135 - 249 mmol/L**
Potassium	202.96	70 - 260 mmol/L**
Chloride	163.3	112 - 184 mmol/L**
Creatinine	64.94	57 - 67 mg/dl*
Micralbumin	0.10	<1.9 mg/dl***

Normal Range established at TJL, C57BL/6J Male 8 Wks. Of age

* Normal Range from The Clinical Chemistry of Laboratory Animals, 2nd Edition, Loeb. Generic strain & age

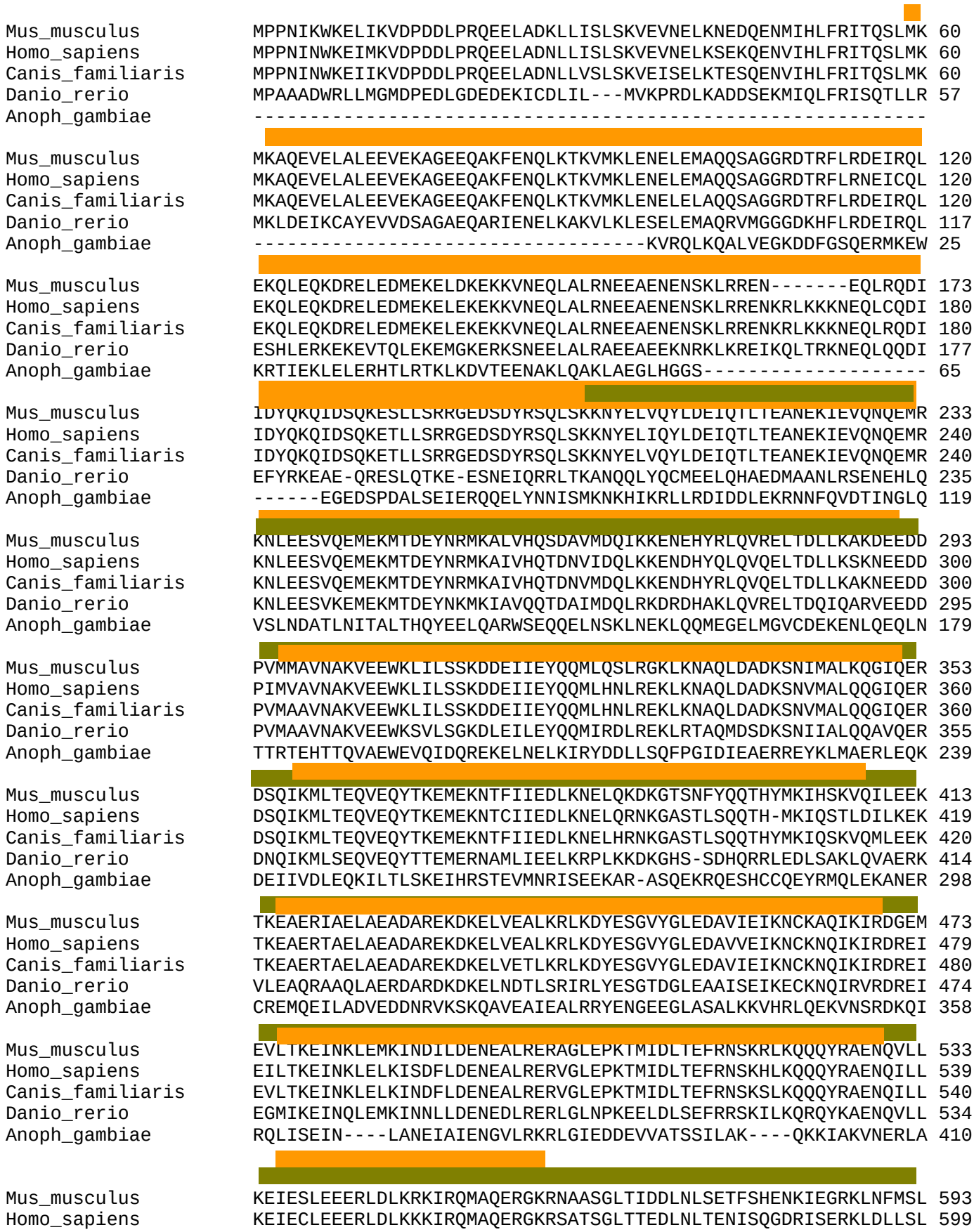
** Rat Normal range, formal ranges for mouse not yet established for urine.

*** Human ranges, formal mouse ranges not yet established.

Supplementary Figure Legends:

Supplementary Figure 1: Evolutionary conservation of CEP290. CLUSTAL analysis of protein sequences from different species was performed using the CLUSTALW alignment program. The CEP290 protein is conserved in evolution, with the region that is deleted in *rd16*, showing high degree of identity (blue) among mammalian species (Alignment scores between 83% and 89%). Major putative domains and motifs are represented with colored bars. The deletion removes majority of the myosin-tail homology domain and KID domains I and II.

Supplementary Figure 2: Immunogold labeling of CEP290 in WT mouse retina. The signal is concentrated in the connecting cilium (CC) (see inset); although some labeling is detected in the inner (IS) and outer segments (OS) as well. Quantitative analysis of the label revealed a four times higher concentration of CEP290 in the connecting cilium than that in the IS and OS of mouse retina (48). R: rootlet.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Canis_familiaris MPPNINWKEIIKVDPDDLPRQEELADNLLVSLSKVEISELKTESQENVIHLFRITQSLMK 60
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Anoph_gambiae --
Mus_musculus MKAQEVELALEEVEKAGEEQAKFENQLKTKVMKLENELEMAQQSAGGRDTRFLRDEIRQL 120
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Danio_rerio MKLDEIKCAYEVVDSAGAEQARIENELKAKVLKLESELEMAQRVMGGGDKHFLRDEIRQL 117
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Homo_sapiens KNLEESVQEMEKMTDEYNRMKAIVHQTDNVIDQLKKENDHYQLQVQELTDLLKSKNEEDD 300
Canis_familiaris KNLEESVQEMEKMTDEYNRMKAIVHQTDNVMDQLKKENDHYRLQVQELTDLLKAKNEEDD 300
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Homo_sapiens PIMVAVNAKVEEWKLILSSKDDEIIEYQQMLHNLREKLKNAQLDADKSNVMALQQGIQER 360
Canis_familiaris PVMAAVNAKVEEWKLILSSKDDEIIEYQQMLHNLREKLKNAQLDADKSNVMALQQGIQER 360
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Anoph_gambiae CREMQEILADVEDDNRVKSKQAVEAIEALRRYENGEEGLASALKKVHRLQEKVNSRDKQI 358
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Homo_sapiens EILTKEINKLELKISDFLDENEALRERVGLEPKTMIDLTEFRNSKHLKQQQYRAENQILL 539
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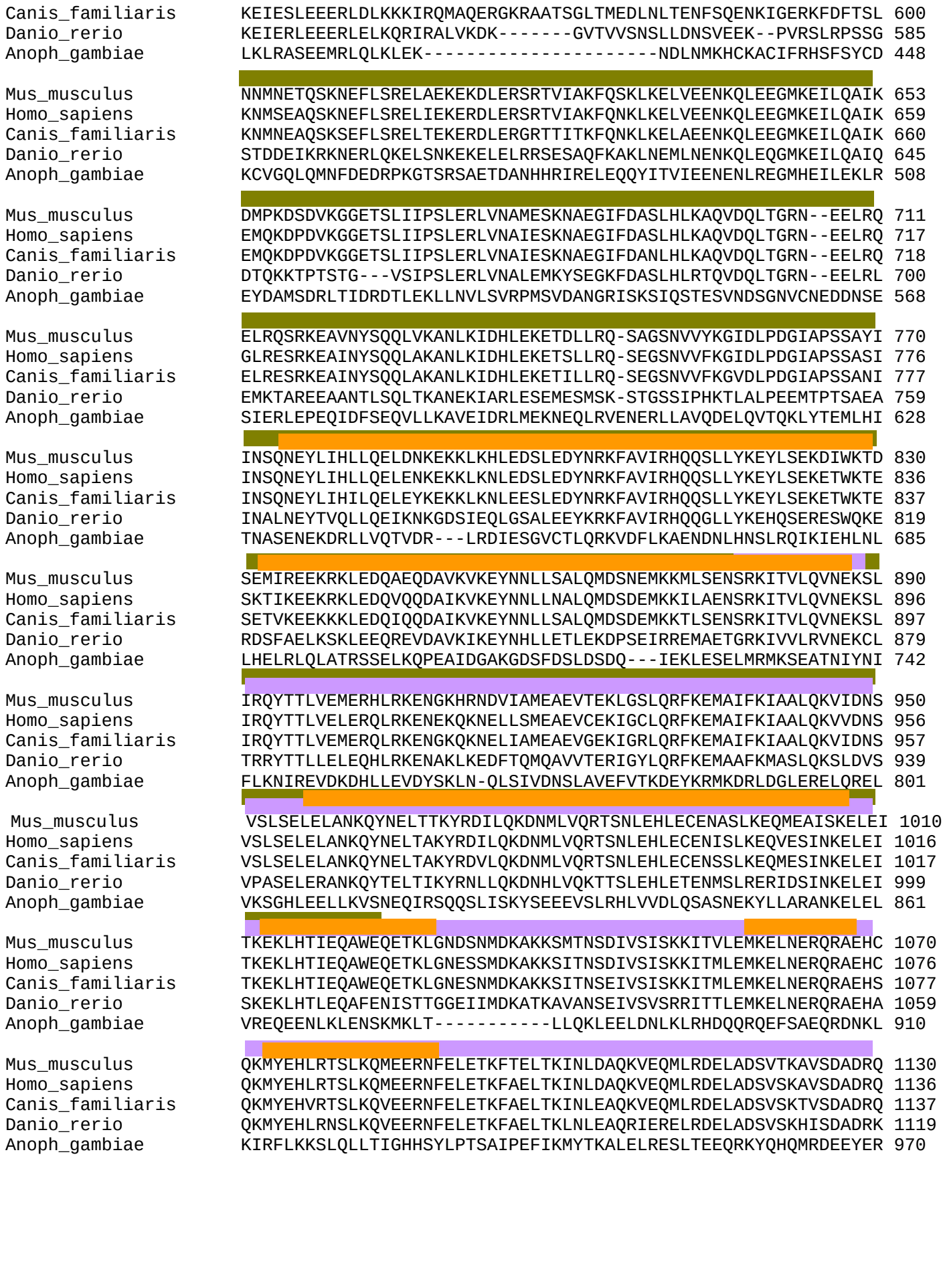
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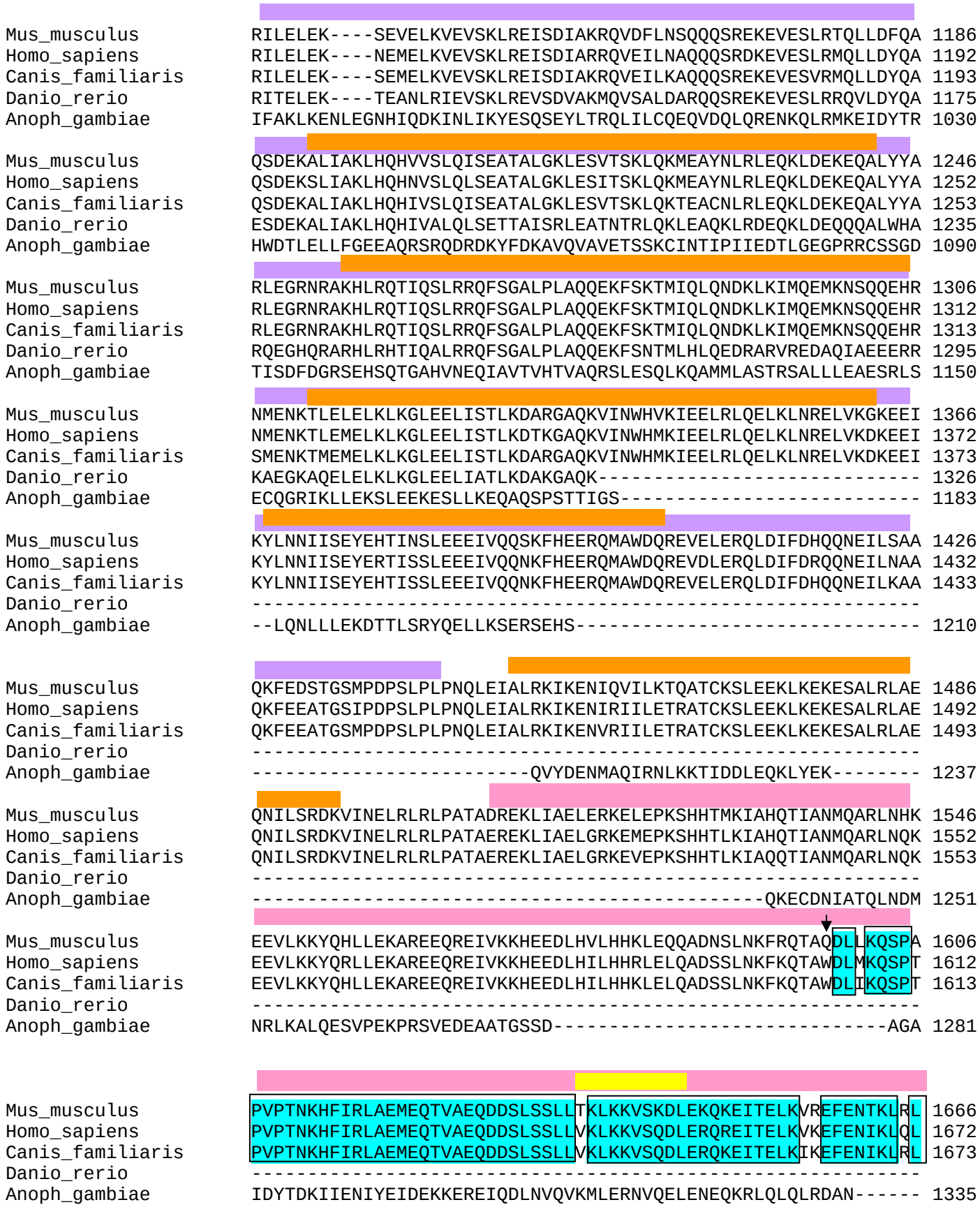
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Canis_familiaris PVPTNKHFIRLAEMEQTVAEQDDSLSSLLVKLKKVSQDLERQKEITELKIKEFENIKLRL 1673
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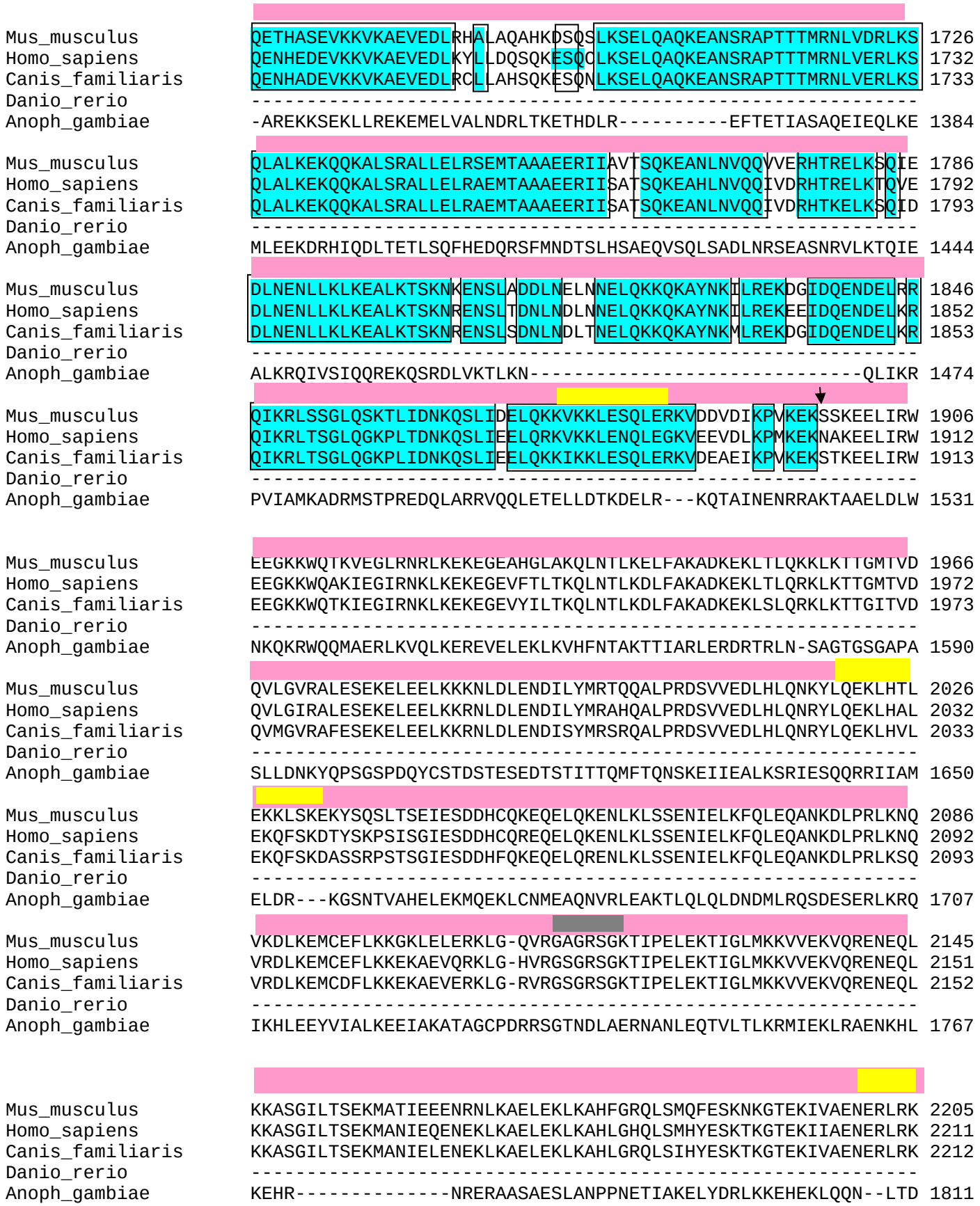
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Homo_sapiens QIKRLTSGLQGKPLTDNKQSLIEELQRKVKKLENQLEGKVEEVDLKPMKEKNAKEELIRW 1912
Canis_familiaris QIKRLTSGLQGKPLIDNKQSLIEELQKKIKKLESQLERKVDEAEIKPVKEKSTKEELIRW 1913
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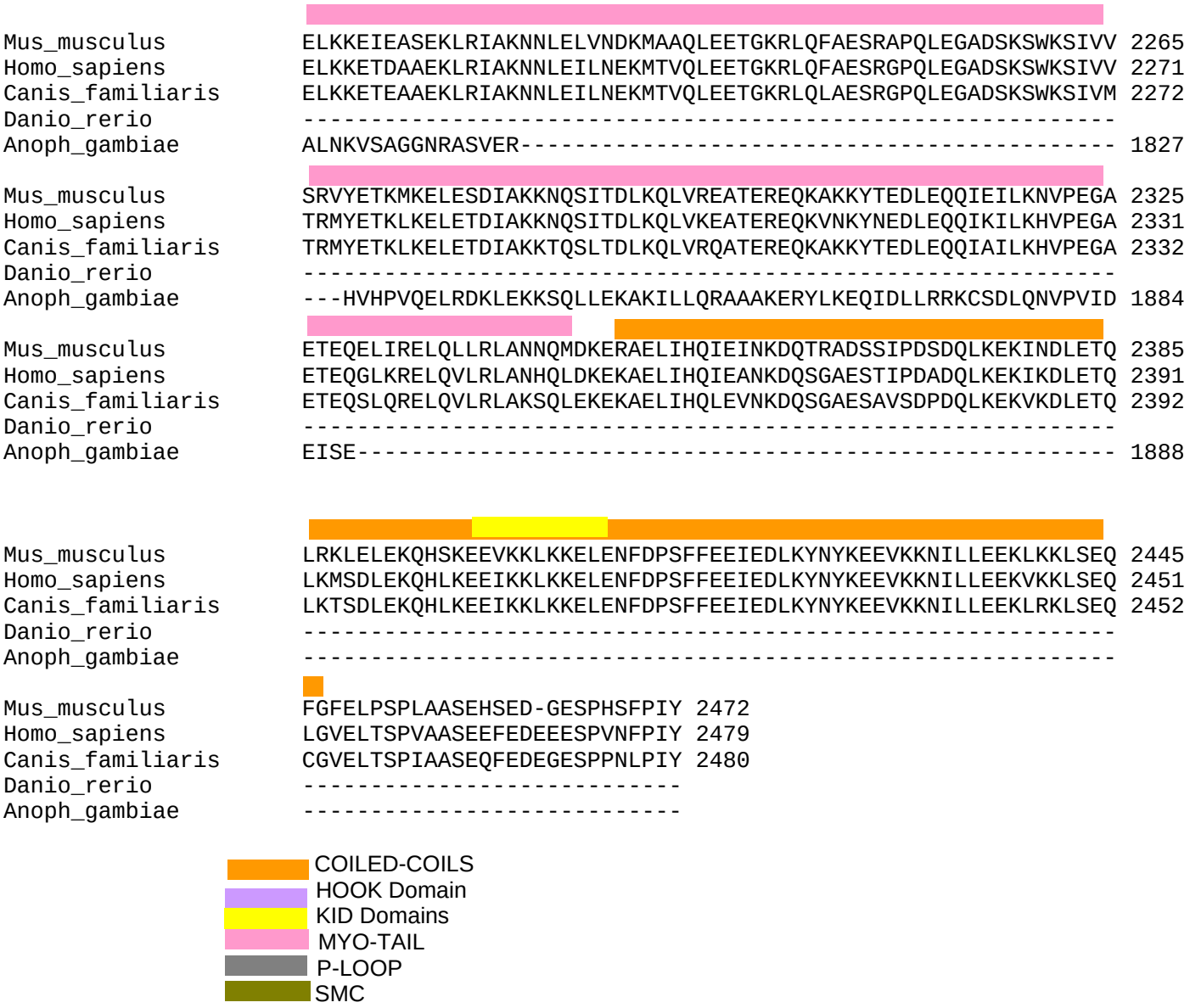
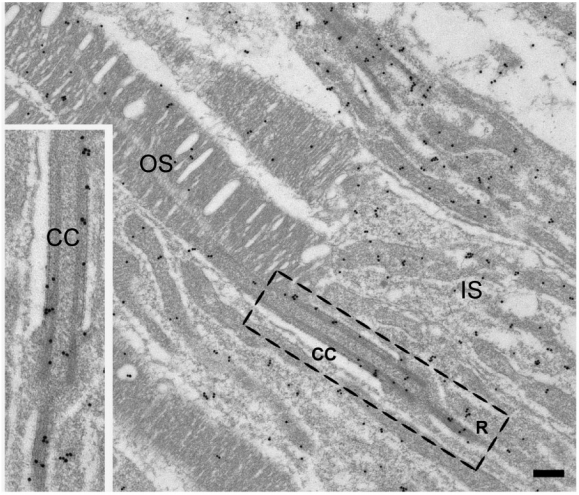
Anoph_gambiae SLLDNKYQPSGSPDQYCSTDSTESEDTSTITTQMFTQNSKEIIEALKSRIESQQRRIIAM 1650
Mus_musculus EKKLSKEKYSQSLTSEIESDDHCQKEQELQKENLKLSSENIELKFQLEQANKDLPRLKNQ 2086
Homo_sapiens EKQFSKDTYSKPSISGIESDDHCQREQELQKENLKLSSENIELKFQLEQANKDLPRLKNQ 2092
Canis_familiaris EKQFSKDASSRPSTSGIESDDHFQKEQELQRENLKLSSENIELKFQLEQANKDLPRLKSQ 2093
Danio_rerio ----------
Anoph_gambiae ELDR---KGSNTVAHELEKMQEKLCNMEAQNVRLEAKTLQLQLDNDMLRQSDESERLKRQ 1707
Mus_musculus VKDLKEMCEFLKKGKLELERKLG-QVRGAGRSGKTIPELEKTIGLMKKVVEKVQRENEQL 2145
Homo_sapiens VRDLKEMCEFLKKEKAEVQRKLG-HVRGSGRSGKTIPELEKTIGLMKKVVEKVQRENEQL 2151
Canis_familiaris VRDLKEMCDFLKKEKAEVERKLG-RVRGSGRSGKTIPELEKTIGLMKKVVEKVQRENEQL 2152
Danio_rerio ----------
Anoph_gambiae IKHLEEYVIALKEEIAKATAGCPDRRSGTNDLAERNANLEQTVLTLKRMIEKLRAENKHL 1767
Mus_musculus KKASGILTSEKMATIEEENRNLKAELEKLKAHFGRQLSMQFESKNKGTEKIVAENERLRK 2205
Homo_sapiens KKASGILTSEKMANIEQENEKLKAELEKLKAHLGHQLSMHYESKTKGTEKIIAENERLRK 2211
Canis_familiaris KKASGILTSEKMANIELENEKLKAELEKLKAHLGRQLSIHYESKTKGTEKIVAENERLRK 2212
Danio_rerio ----------
Anoph_gambiae KEHR----------NRERAASAESLANPPNETIAKELYDRLKKEHEKLQQN--LTD 1811



DELETED IN *rd16*

Name	Amino-acids	Name	Amino-acids	Score
Mus_musculus	1599-1897	Homo_sapiens	1605-1903	83
Mus_musculus	1599-1897	Canis_familiaris	1596-1904	86
Homo_sapiens	1605-1903	Canis_familiaris	1596-1904	89

SUPPLEMENTARY FIGURE 1



SUPPLEMENTARY FIGURE 2