

# Error and Error Mitigation in Low-Coverage Genomes

M.J. Hubisz, M.F. Lin, M. Kellis, A. Siepel

Table S2: Summary of alignments of 2x genomes and corresponding ENCODE sequences

2x assembly	ENCODE species	ENCODE regions	Alignment length (Mb)	Percent coverage <sup>a</sup>
dasNov2	armadillo	44	13.79	38.3
echTel1	tenrec	42	11.15	37.7
eriEur1	hedgehog	39	10.97	39.1
felCat3	cat	43	12.69	46.5
loxAfr2	elephant	44	16.90	48.8
micMur1	mouse_lemur	43	19.18	64.4
myoLuc1	sbbat	41	13.99	59.7
oryCun1	rabbit	43	16.46	53.3
otoGar1	galago	44	20.05	53.7
proCap1	rock_hyrax	6	3.79	55.6
pteVam1	flying_fox	5	4.30	83.9
sorAra1	shrew	41	13.90	57.0
speTri1	st_squirrel	44	14.42	53.5
tupBel1	tree_shrew	4	2.14	53.5
<i>cavPor3<sup>b</sup></i>	<i>guinea_pig</i>	<i>41</i>	<i>25.05</i>	<i>95.9</i>

<sup>a</sup>Fraction of ENCODE bases aligned to 2x assembly

<sup>b</sup>Sequenced to ~7x coverage and shown for comparison.

**Table S1.** Estimates of  $d_N/d_S$  for chr22 genes and four primates

<b>branch</b>	<b>full data</b>	<b>high-quality only</b>	<b>SEM</b>
tarsier	0.179	0.164	0.165
mouse lemur	0.173	0.166	0.162
bushbaby	0.189	0.179	0.171
tree shrew	0.135	0.123	0.124
<i>internal branch</i>	0.161	0.142	0.156

**Table S2.** CONGO performance with and without SEM

<b>Gene Set</b>	<b>Statistic<sup>a</sup></b>	<b>Baseline (%)<sup>b</sup></b>	<b>B+SEM (%)<sup>c</sup></b>	<b>SEM (%)<sup>d</sup></b>
CCDS <sup>e</sup>	Exon Sn	82.84	82.88	82.96
	Exon Sp	76.86	77.18	77.38
	Missed Exons	12.66	12.71	12.99
	Wrong Exons	17.01	16.95	16.86
	Nuc Sn	84.81	85.10	85.21
	Nuc Sp	82.13	82.08	82.02
R+E+U+G <sup>f</sup>	Exon Sn	74.26	74.31	74.36
	Exon Sp	89.88	90.28	90.49
	Missed Exons	21.86	21.90	22.17
	Wrong Exons	3.01	2.94	2.88
	Nuc Sn	76.01	76.30	76.40
	Nuc Sp	96.89	96.88	96.80

<sup>a</sup>As defined by [?]. Performance is measured against the whole genome, excluding the ENCODE “random” regions (~0.5% of the genome), which were used for training.

<sup>b</sup>Training and testing on original alignments.

<sup>c</sup>Training on original alignments, testing on alignments processed by SEM.

<sup>d</sup>Training and testing on alignments processed by SEM.

<sup>e</sup>“Consensus CDS” gene set (more conservative).

<sup>f</sup>Union of RefSeq, ENSEMBL, UCSC, and GENCODE gene sets (less conservative).

**Table S3.** CONGO performance by chromosome

Chrom.	Exon Sp (%) <sup>a</sup>		Exon Sn (%) <sup>b</sup>	
	Baseline	SEM <sup>c</sup>	Baseline	SEM <sup>c</sup>
1	90.10	<b>90.67</b>	83.00	<b>83.11</b>
2	90.34	<b>90.75</b>	85.52	85.43
3	90.43	<b>90.81</b>	86.27	86.05
4	89.43	<b>89.79</b>	84.75	<b>84.76</b>
5	89.33	<b>89.54</b>	85.75	85.69
6	89.16	<b>90.21</b>	84.05	<b>84.48</b>
7	90.61	<b>91.42</b>	81.99	<b>82.31</b>
8	89.53	<b>90.07</b>	85.05	<b>85.18</b>
9	89.97	<b>90.72</b>	84.53	<b>84.61</b>
10	90.02	<b>90.43</b>	83.94	83.66
11	86.51	<b>87.35</b>	84.52	<b>84.86</b>
12	91.11	<b>91.64</b>	85.83	85.83
13	90.62	<b>90.97</b>	81.85	<b>82.05</b>
14	88.55	<b>89.43</b>	86.19	<b>86.38</b>
15	91.28	<b>91.98</b>	83.00	<b>83.07</b>
16	89.99	<b>90.61</b>	79.69	<b>79.86</b>
17	90.93	<b>91.65</b>	83.53	<b>83.83</b>
18	89.24	<b>89.85</b>	88.07	<b>88.40</b>
19	89.93	<b>90.45</b>	71.70	<b>71.83</b>
20	91.38	<b>92.13</b>	85.24	<b>85.70</b>
21	91.36	<b>92.09</b>	81.76	<b>82.04</b>
22	90.83	<b>91.53</b>	80.33	<b>80.59</b>
X	88.06	<b>88.88</b>	74.80	<b>75.04</b>
Y	68.42	68.42	5.13	5.13

<sup>a</sup>Computed from union of RefSeq, ENSEMBL, UCSC, and GENCODE gene sets (R+E+U+G).

<sup>b</sup>Computed from CCDS gene set.

<sup>c</sup>SEM was used for training and testing. Bold indicates an improvement.