

**SUPPLEMENTAL MATERIAL: ESTIMATING ERROR MODELS FOR WHOLE GENOME SEQUENCING USING MIXTURES OF DIRICHLET-MULTINOMIAL DISTRIBUTIONS**

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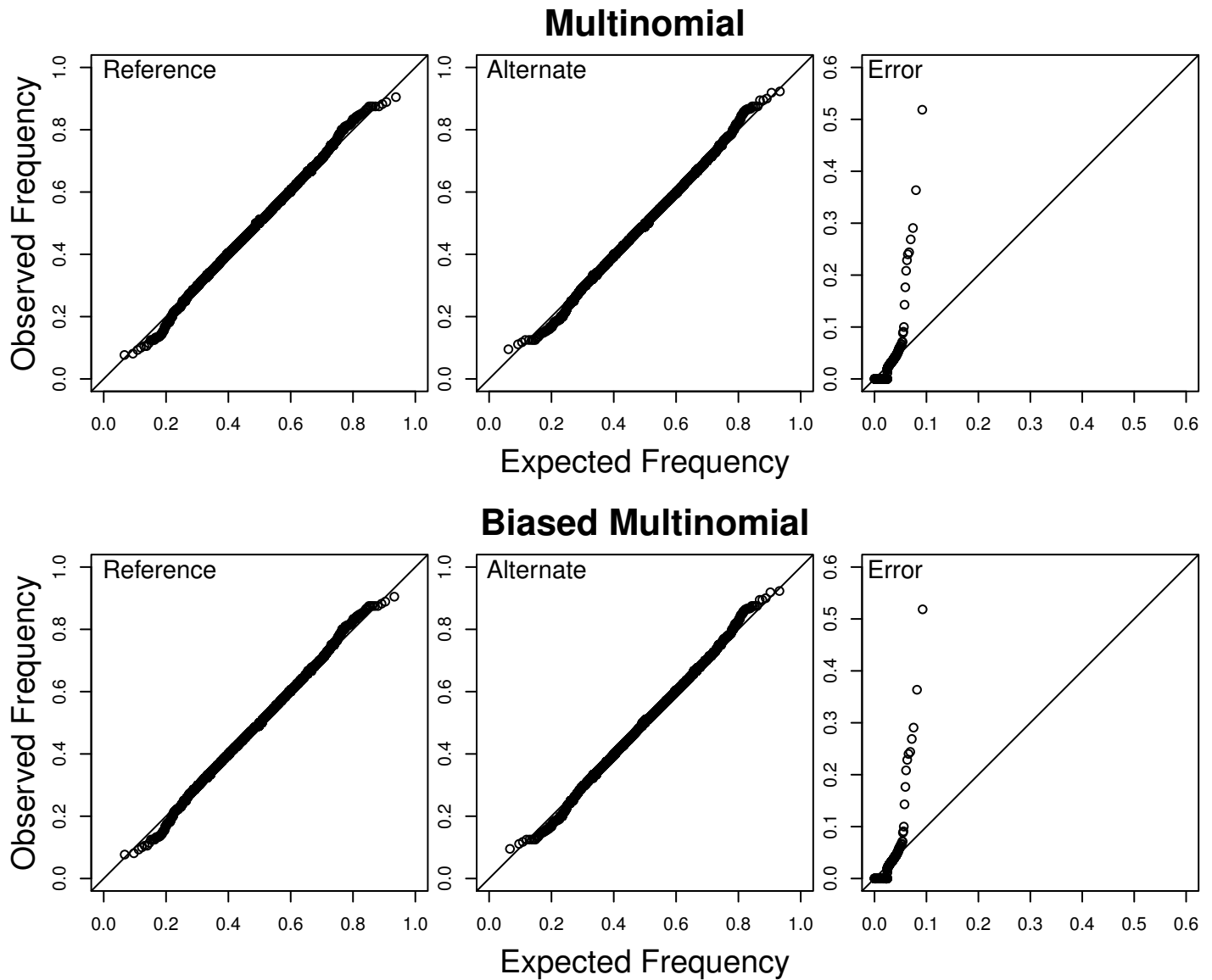
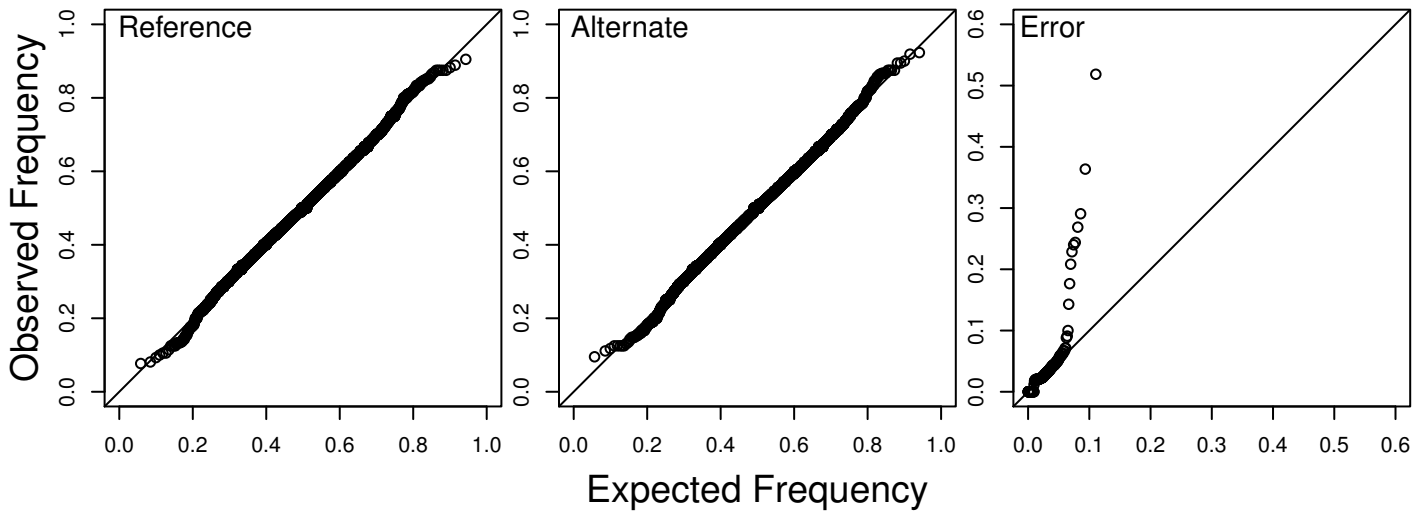
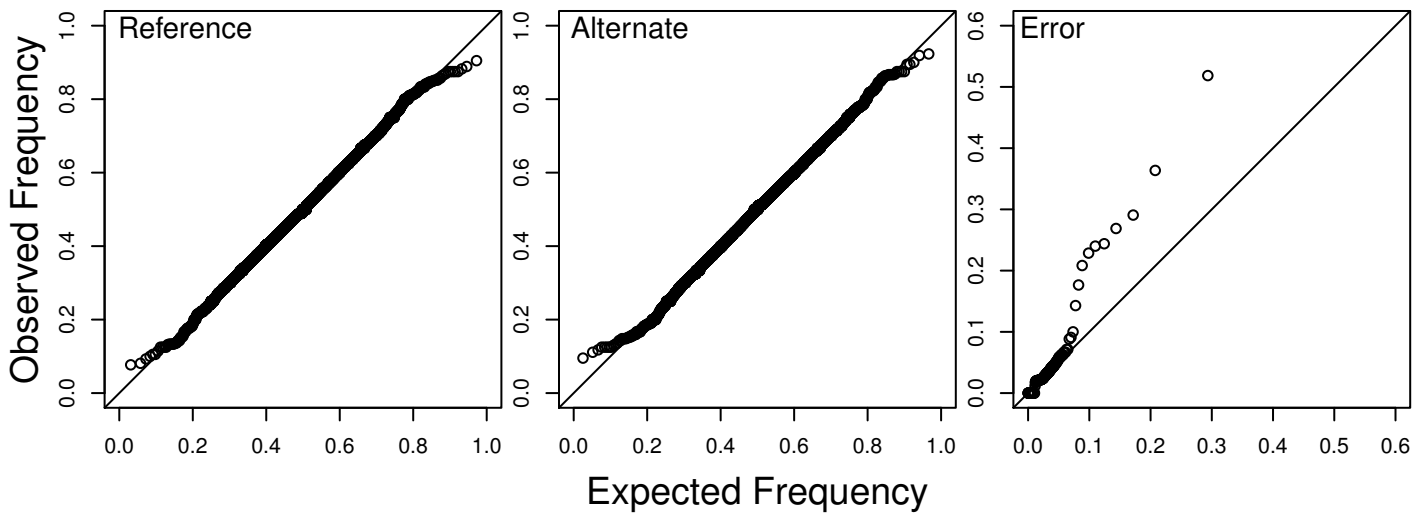


Figure S1: **Mixtures of Dirichlet-multinomials provide the best fits to genomic data: CEU13 Chr10 TH.** QQ plots evaluating the fits of several model to this dataset. The quantiles of the observed read count frequencies are calculated from the datasets, and the quantiles of the expected read count frequencies are estimated from the fitted model. A model that fits the data well produces points that fall along the diagonal.

## Dirichlet-Multinomial



## Mixture of 2 Dirichlet-Multinomials



## Mixture of 3 Dirichlet-Multinomials

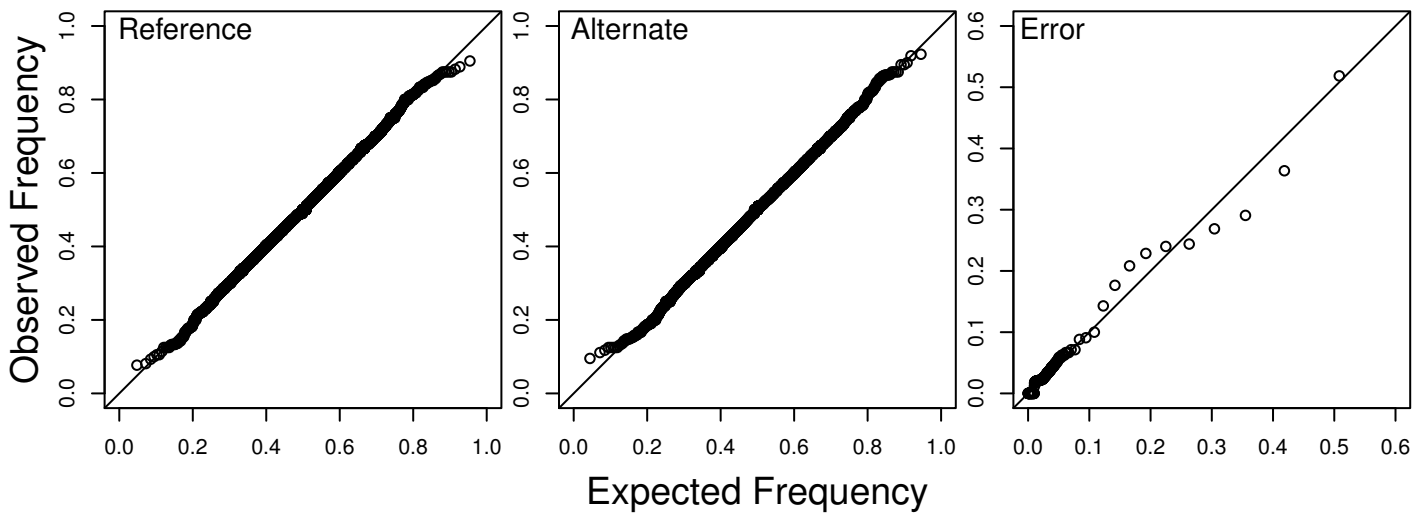
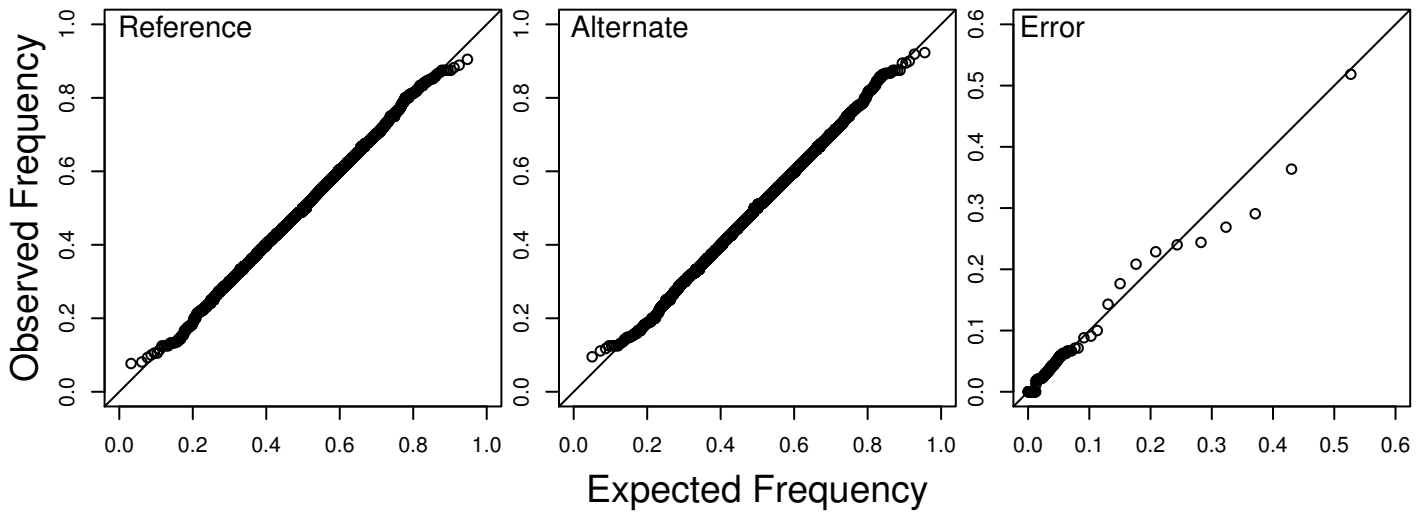
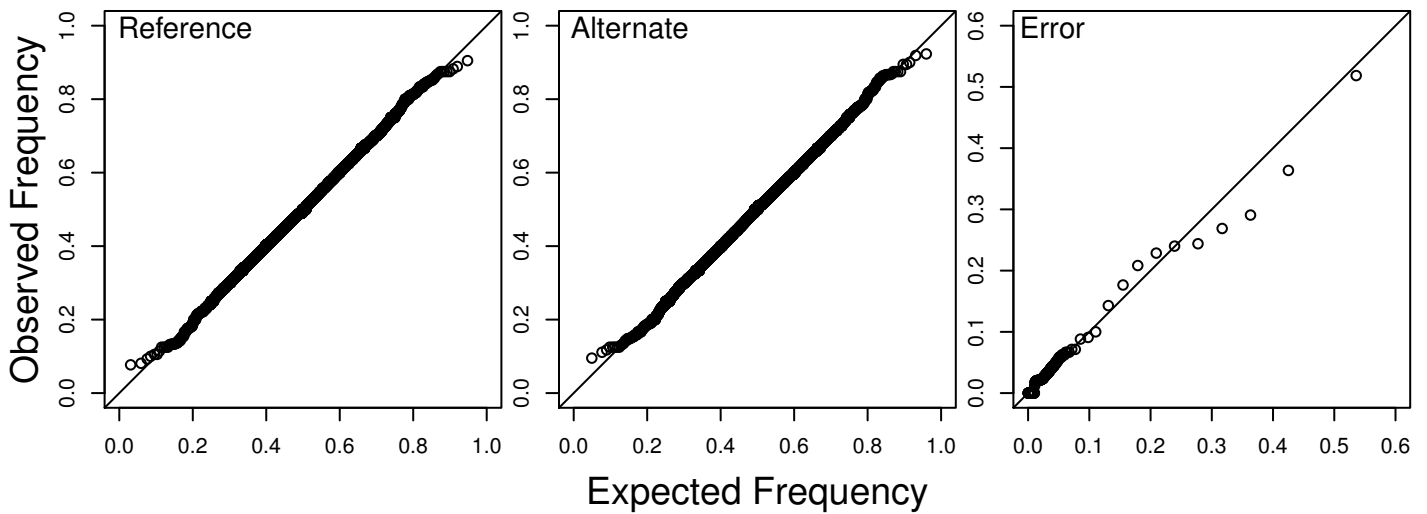


Figure S1 (Continued): **Mixtures of Dirichlet-multinomials provide the best fits to genomic data: CEU13 Chr10 TH.**

### Mixture of 4 Dirichlet-Multinomials



### Mixture of 5 Dirichlet-Multinomials



### Mixture of 6 Dirichlet-Multinomials

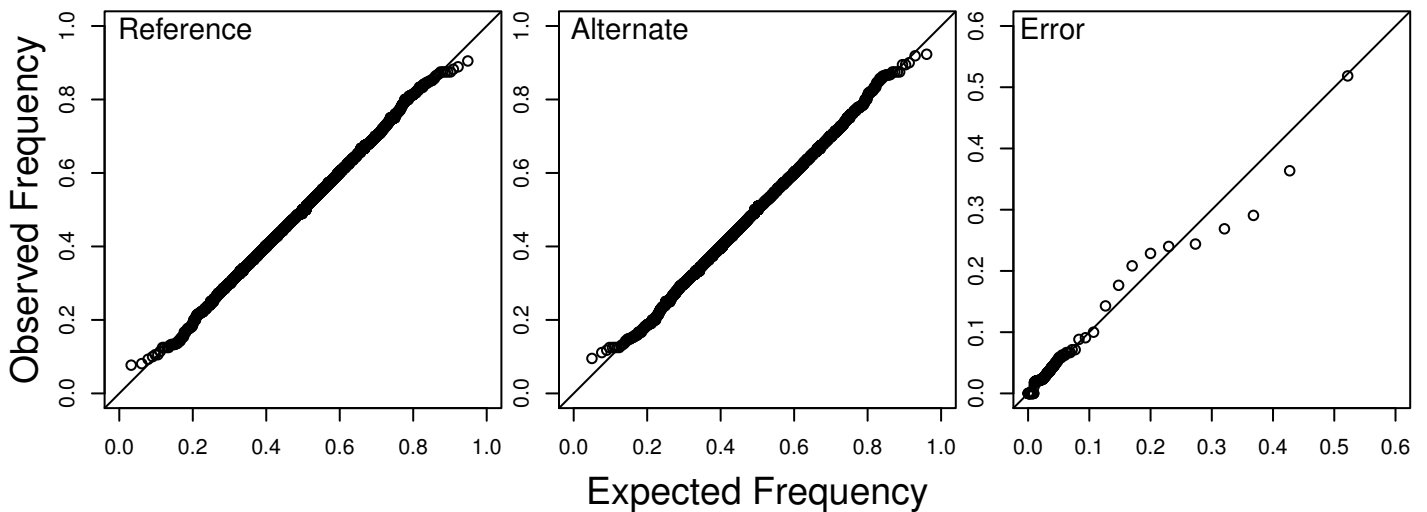
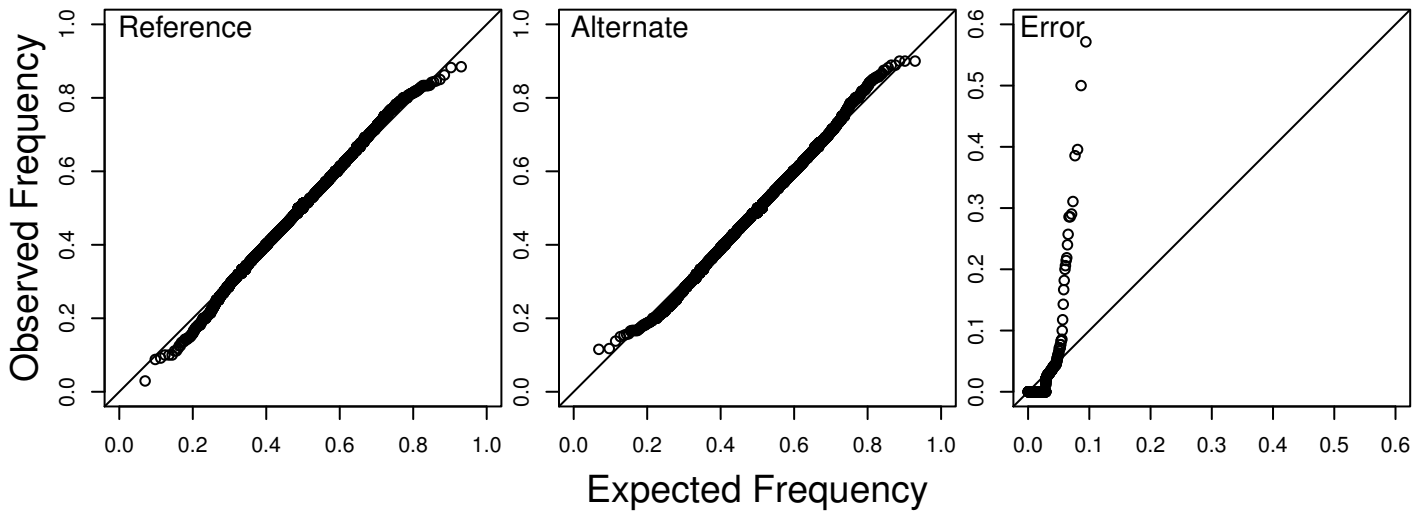
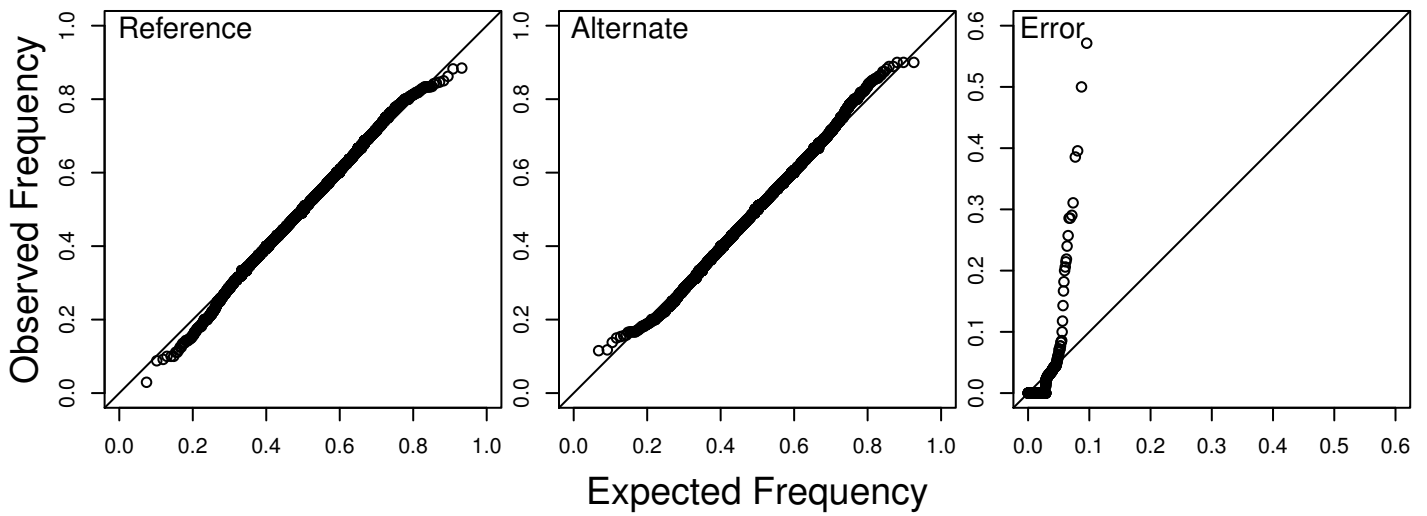


Figure S1 (Continued): **Mixtures of Dirichlet-multinomials provide the best fits to genomic data: CEU13 Chr10 TH.**

## Multinomial



## Biased Multinomial



## Dirichlet-Multinomial

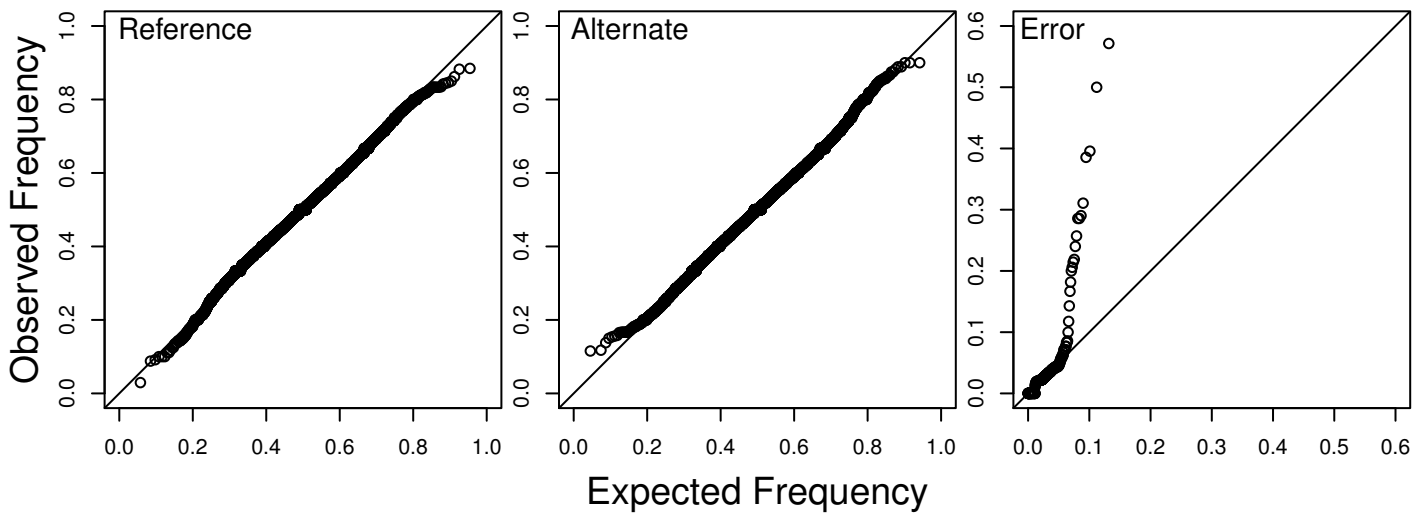
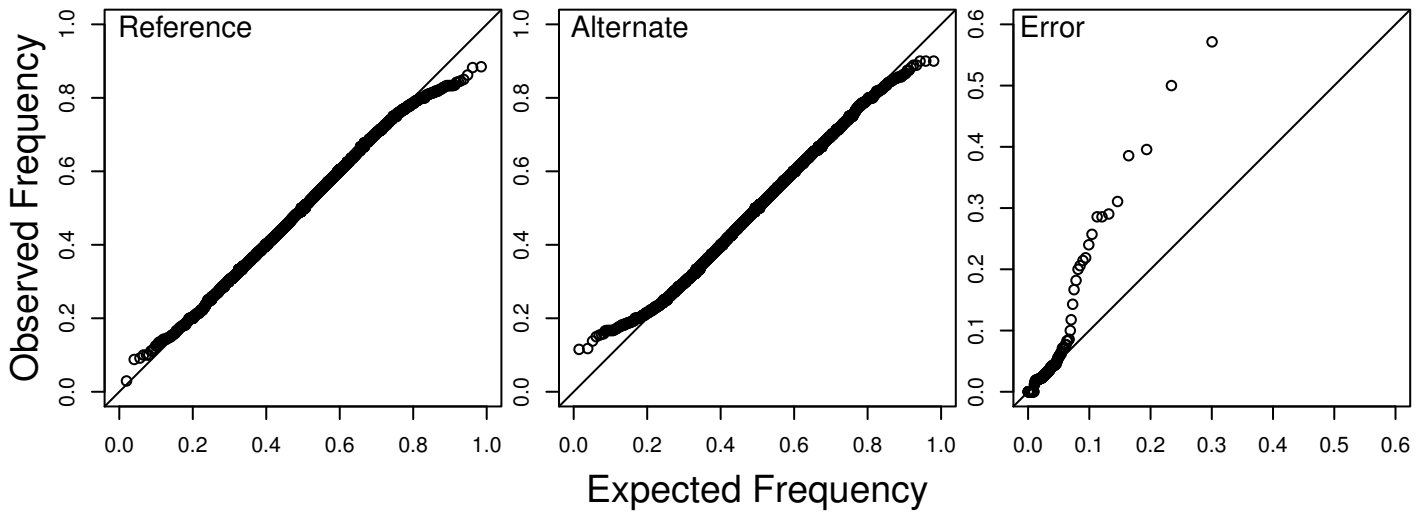
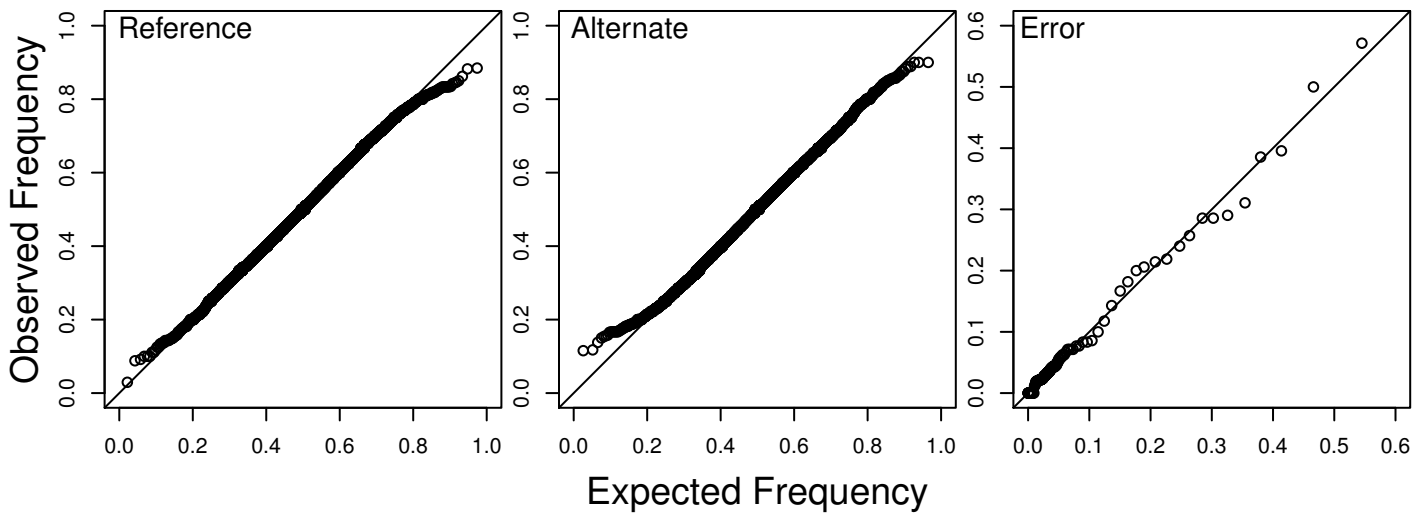


Figure S2: Mixtures of Dirichlet-multinomials provide the best fits to genomic data: CEU13 Chr21 TH.

### Mixture of 2 Dirichlet-Multinomials



### Mixture of 3 Dirichlet-Multinomials



### Mixture of 4 Dirichlet-Multinomials

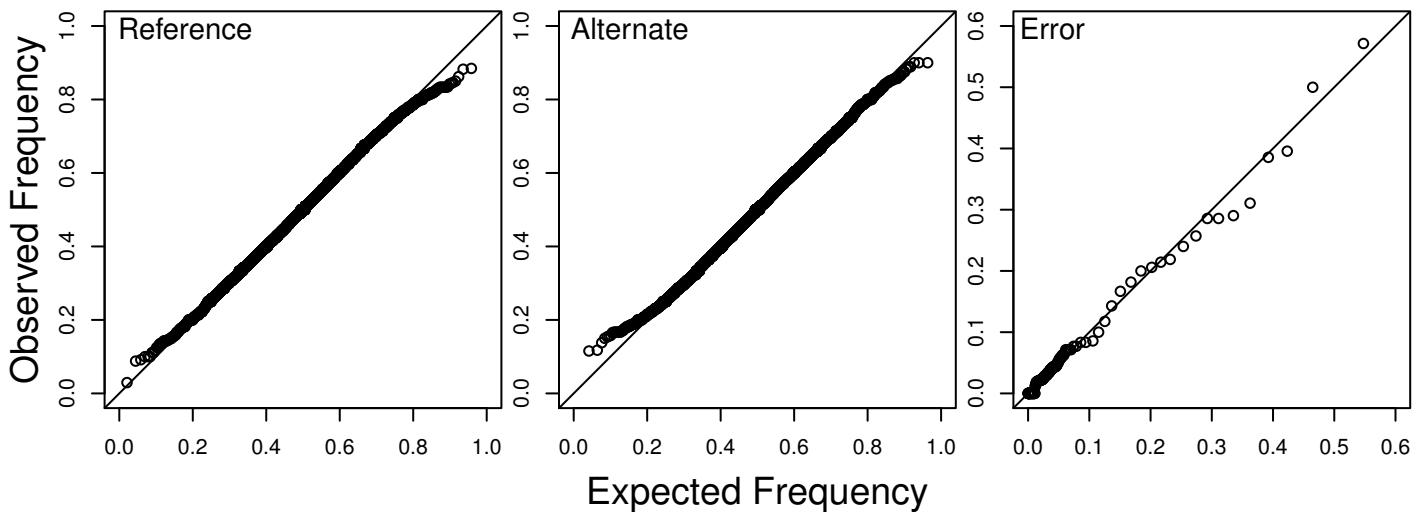
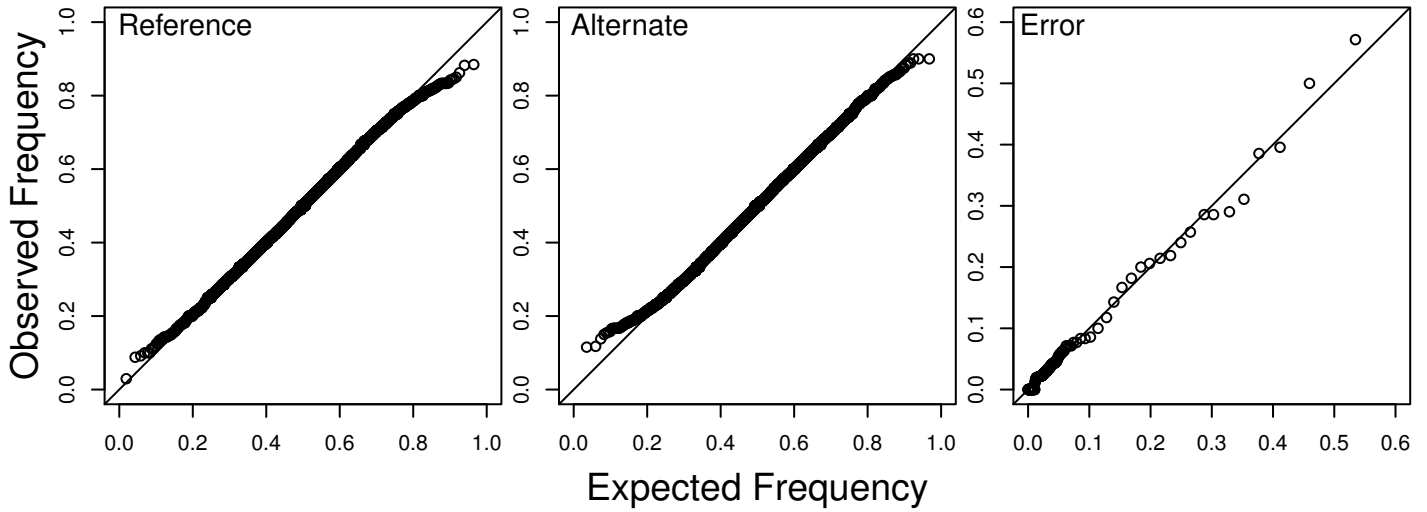


Figure S2 (Continued): **Mixtures of Dirichlet-multinomials provide the best fits to genomic data: CEU13 Chr21 TH.**

### Mixture of 5 Dirichlet–Multinomials



### Mixture of 6 Dirichlet–Multinomials

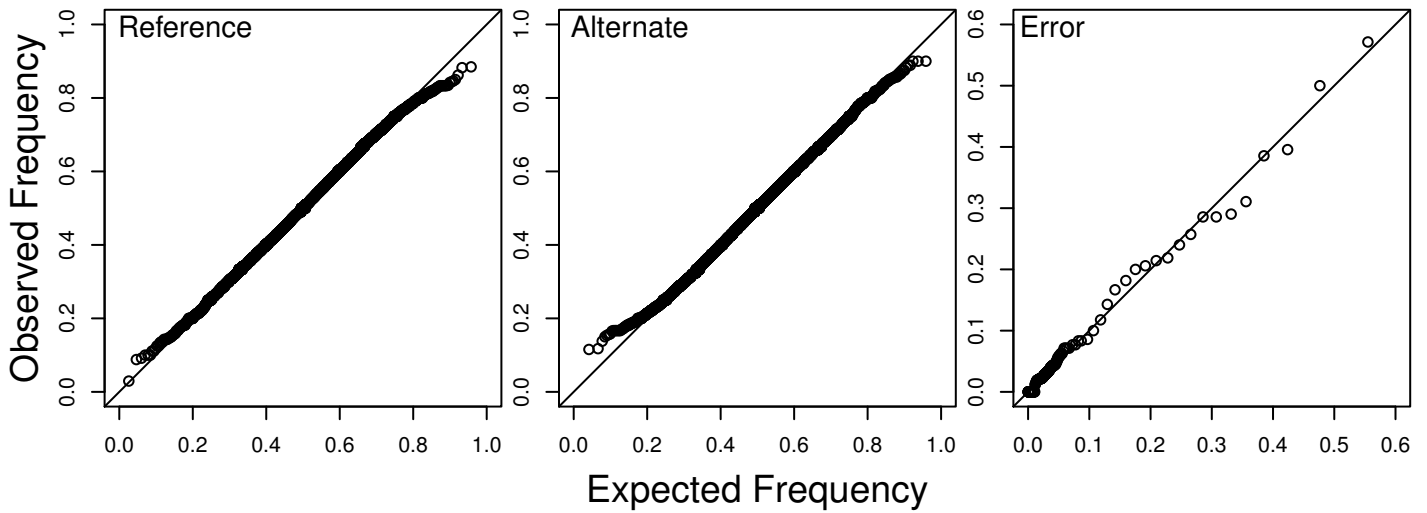
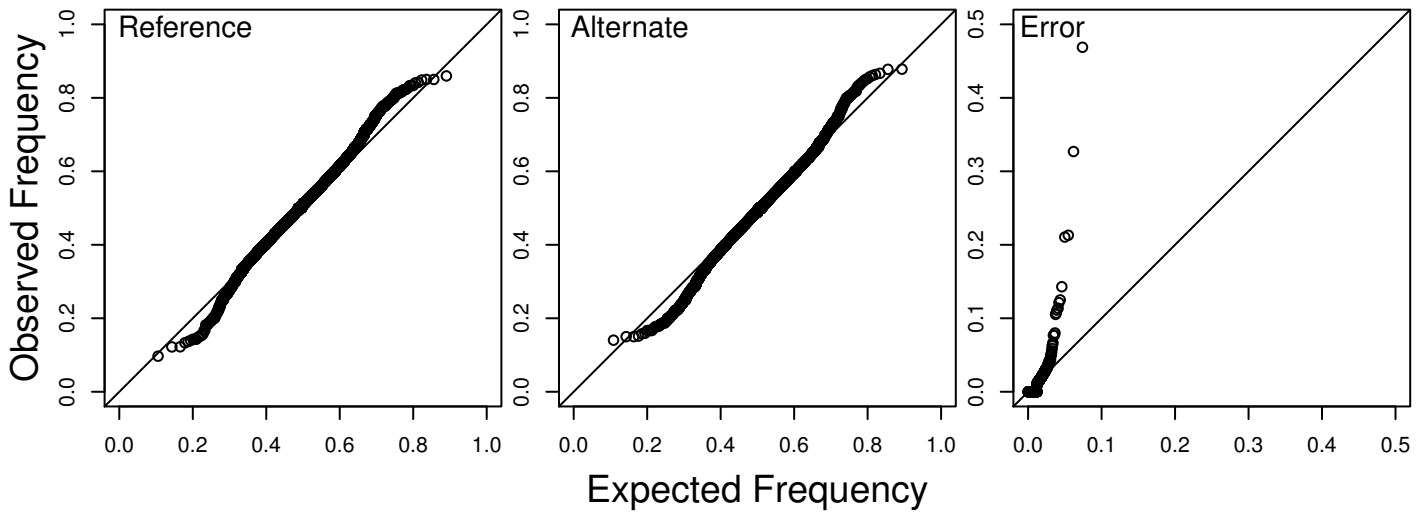
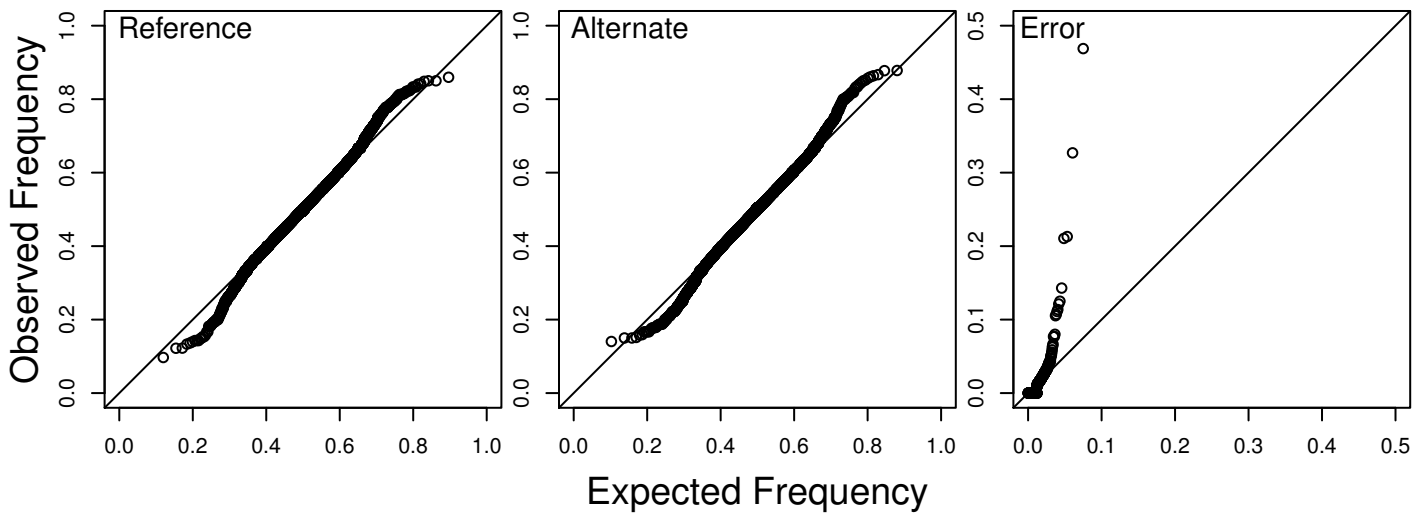


Figure S2 (Continued): **Mixtures of Dirichlet-multinomials provide the best fits to genomic data: CEU13 Chr21 TH.**

## Multinomial



## Biased Multinomial



## Dirichlet-Multinomial

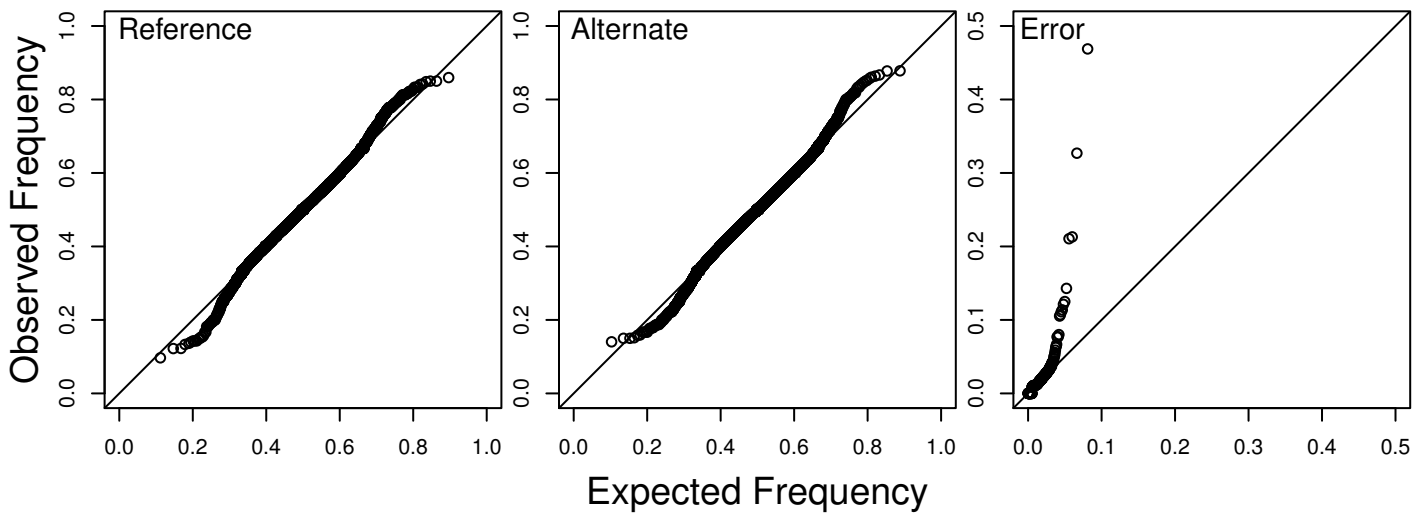
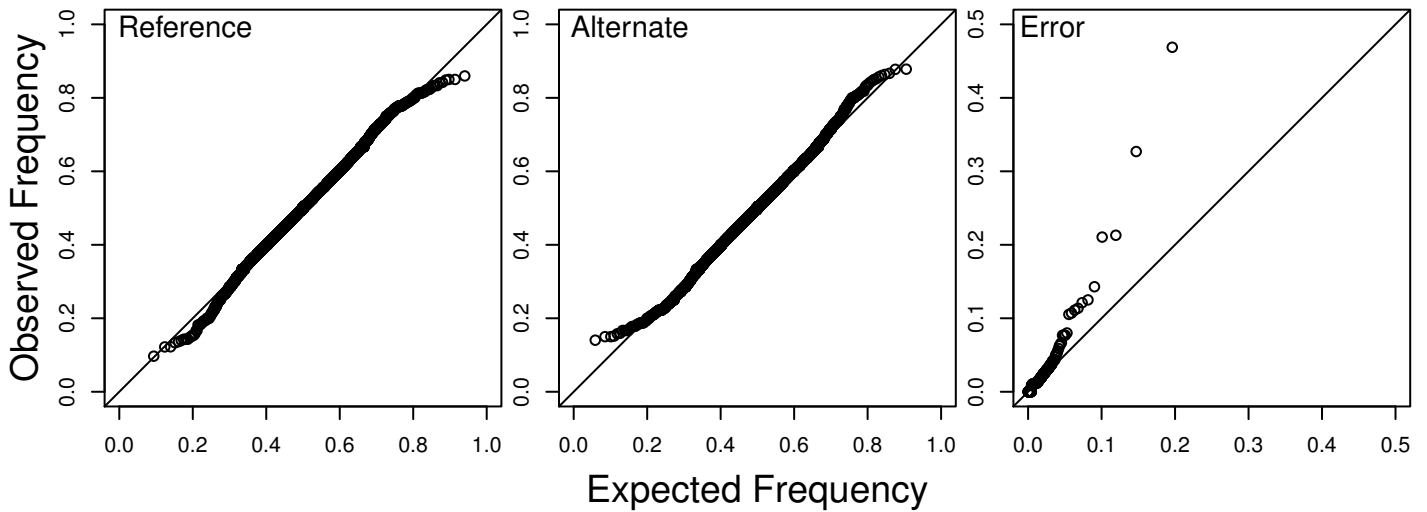
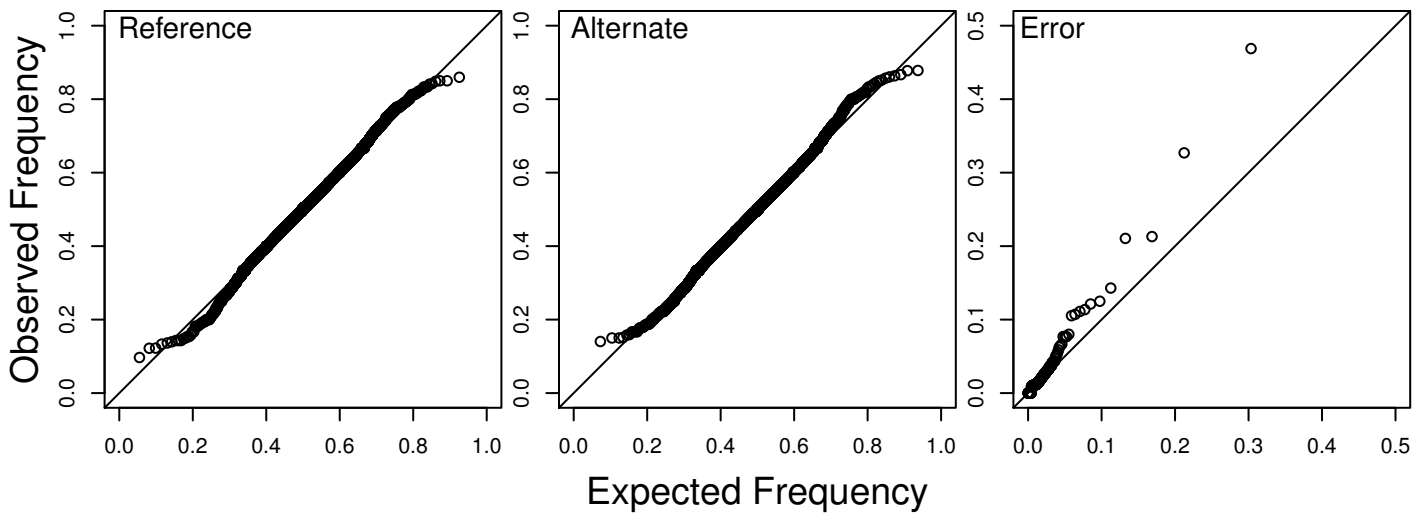


Figure S3: Mixtures of Dirichlet-multinomials provide the best fits to genomic data: CEU12 Chr10 TH.

### Mixture of 2 Dirichlet-Multinomials



### Mixture of 3 Dirichlet-Multinomials



### Mixture of 4 Dirichlet-Multinomials

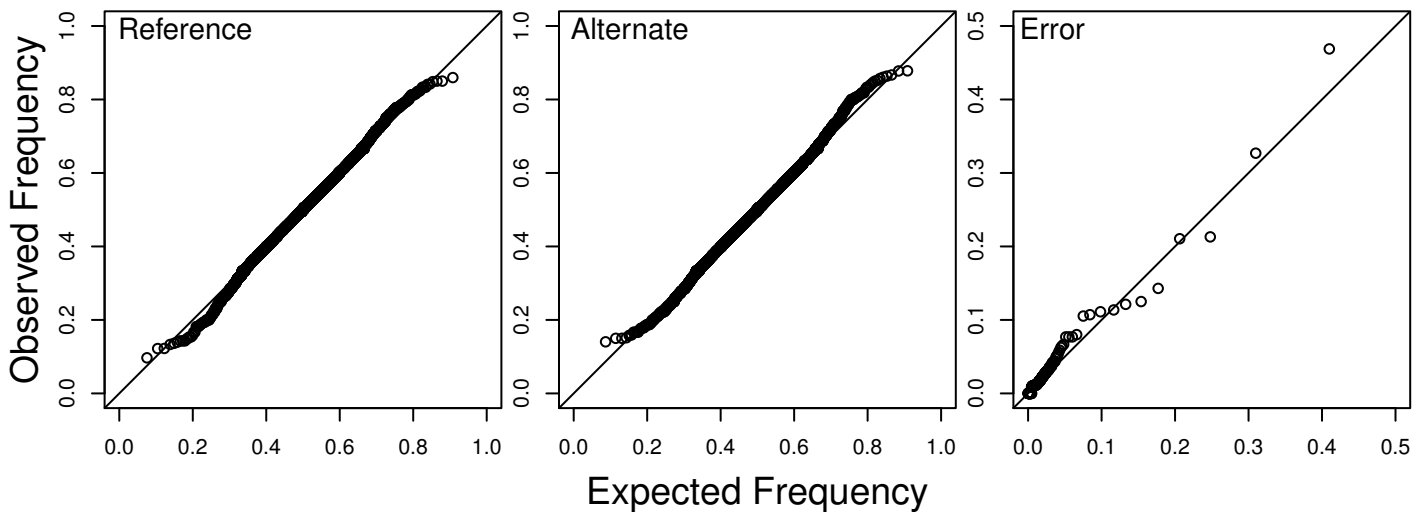
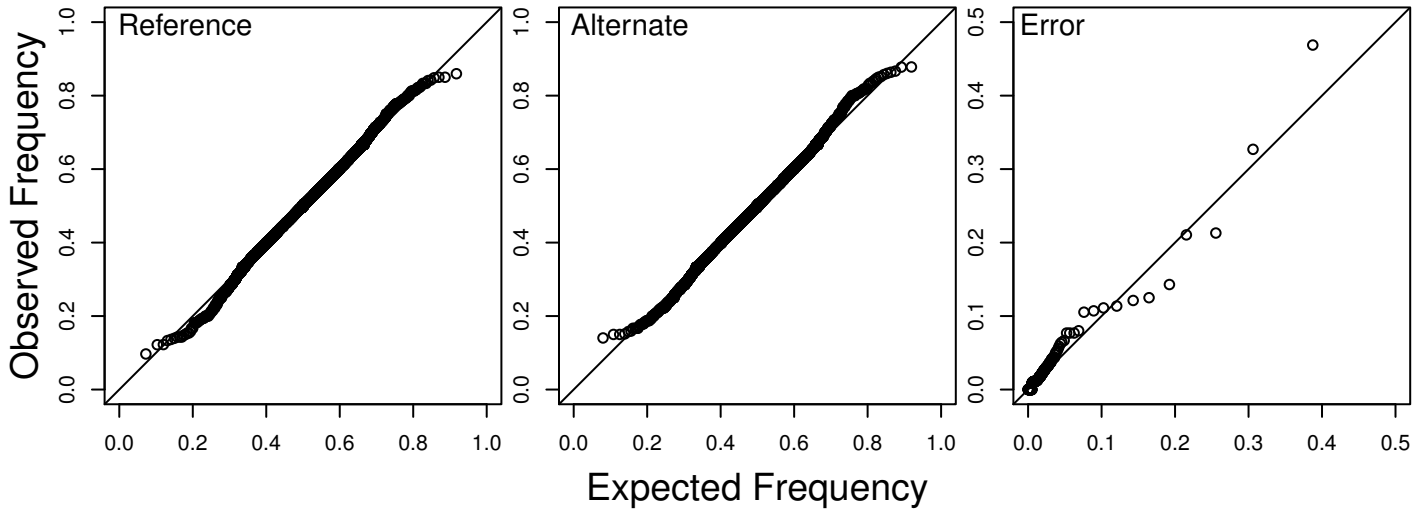


Figure S3 (Continued): **Mixtures of Dirichlet-multinomials provide the best fits to genomic data: CEU12 Chr10 TH.**



### Mixture of 5 Dirichlet–Multinomials



### Mixture of 6 Dirichlet–Multinomials

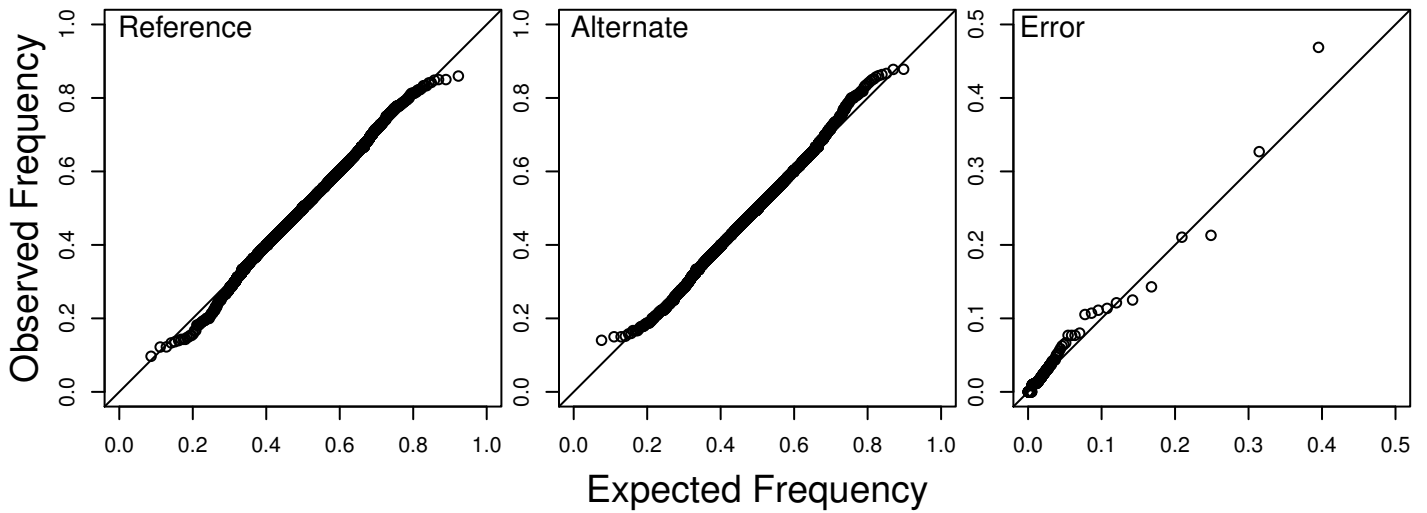
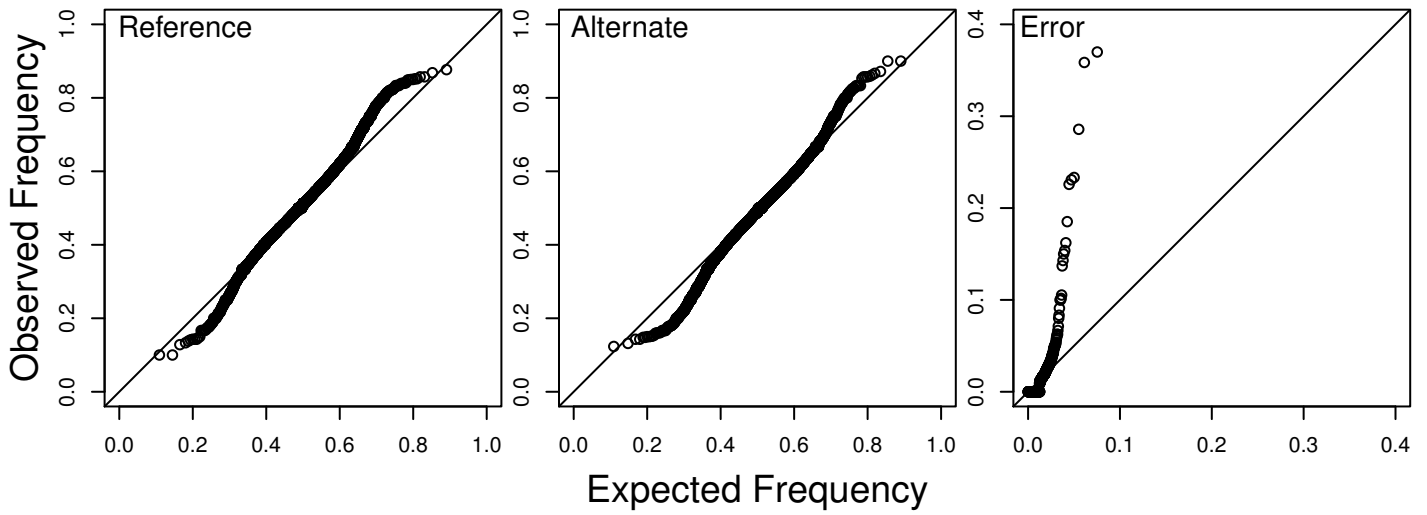
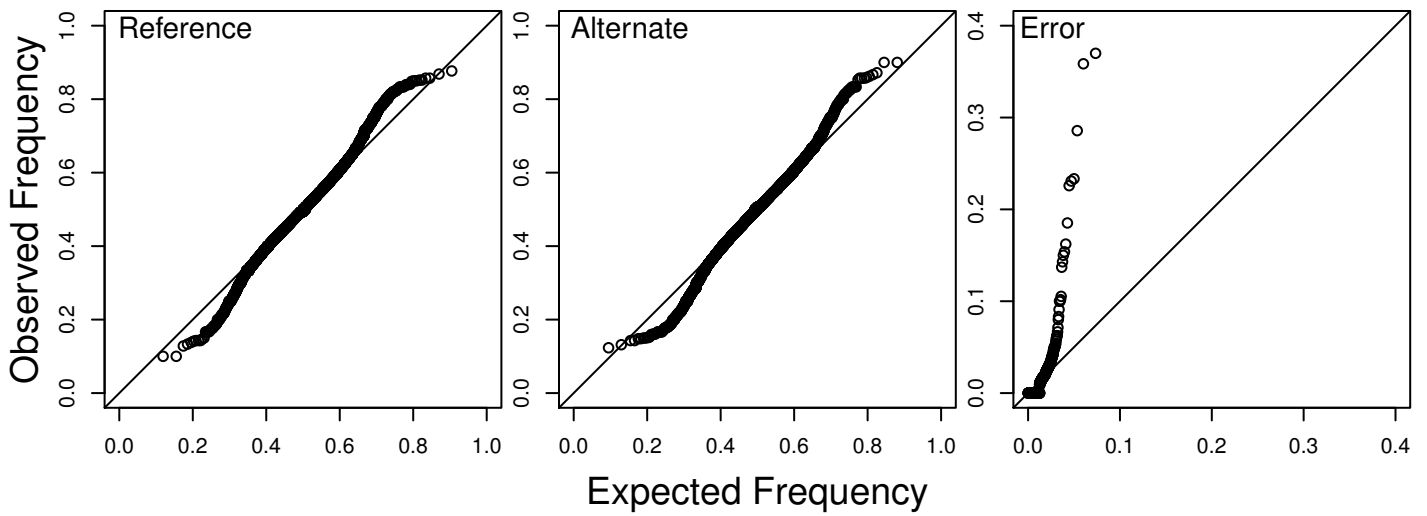


Figure S3 (Continued): **Mixtures of Dirichlet-multinomials provide the best fits to genomic data: CEU12 Chr10 TH.**

## Multinomial



## Biased Multinomial



## Dirichlet-Multinomial

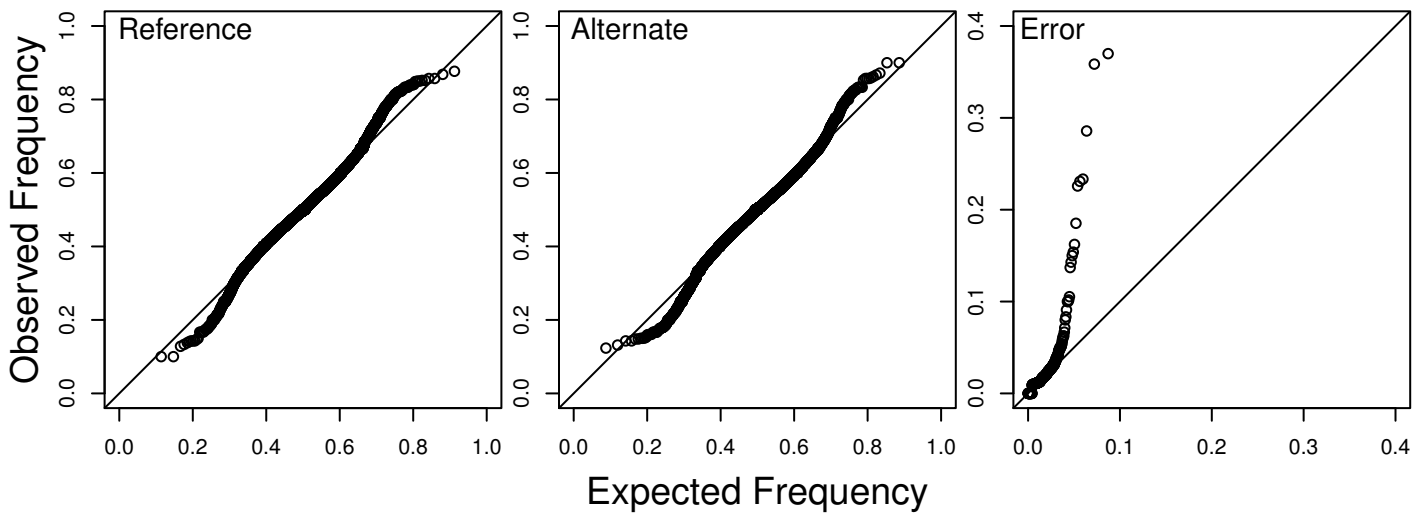
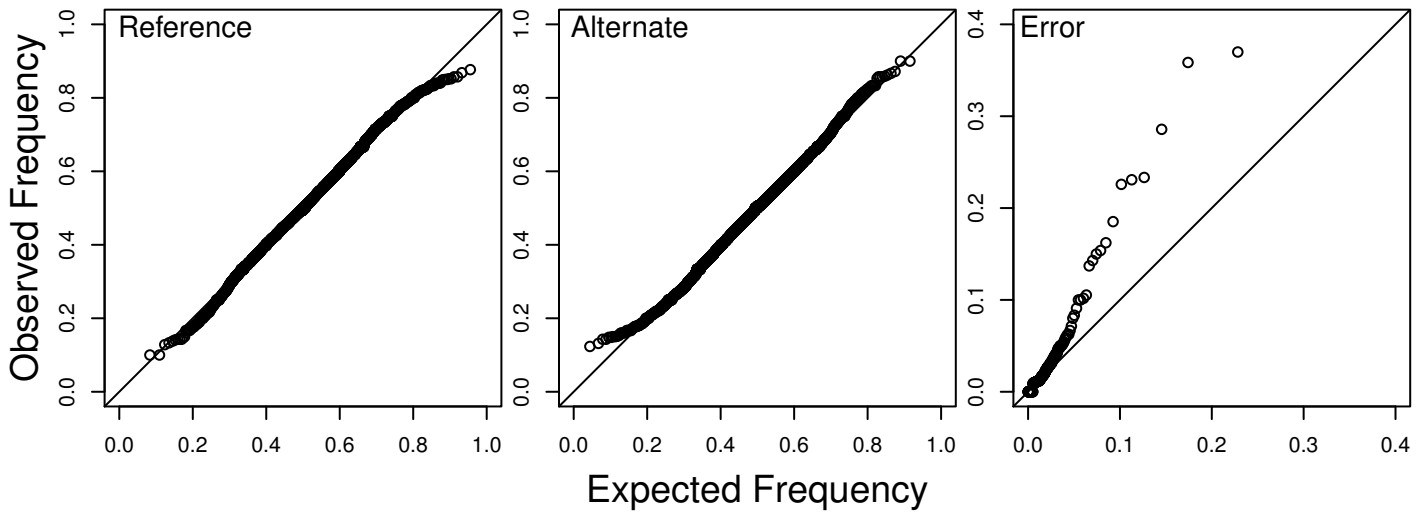
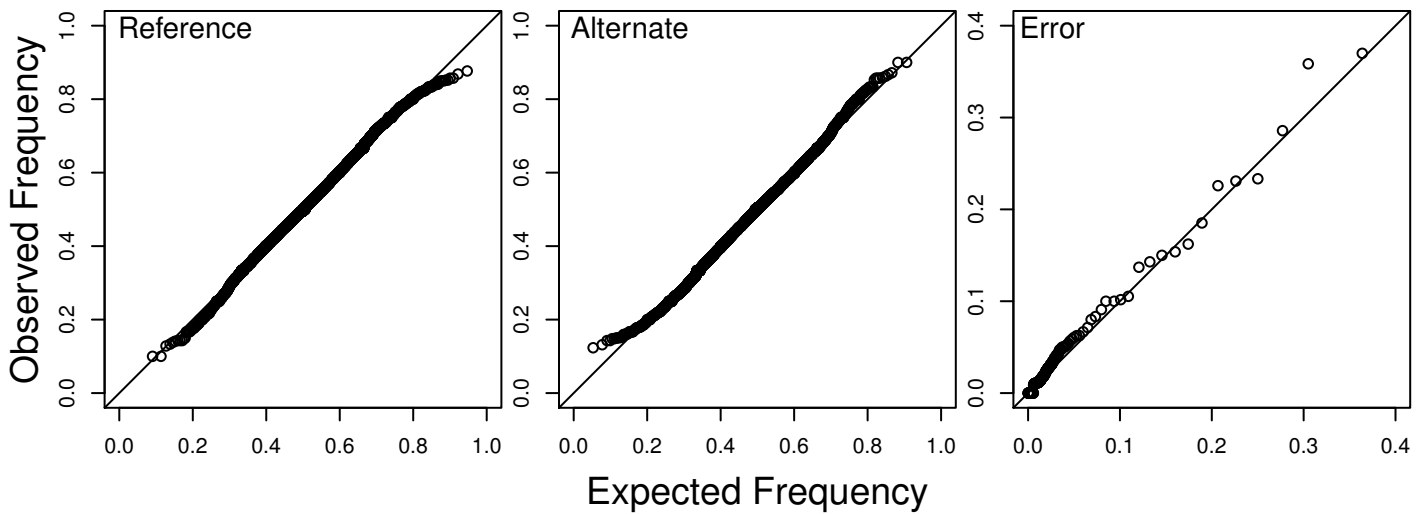


Figure S4: Mixtures of Dirichlet-multinomials provide the best fits to genomic data: CEU12 Chr21 TH.

### Mixture of 2 Dirichlet-Multinomials



### Mixture of 3 Dirichlet-Multinomials



### Mixture of 4 Dirichlet-Multinomials

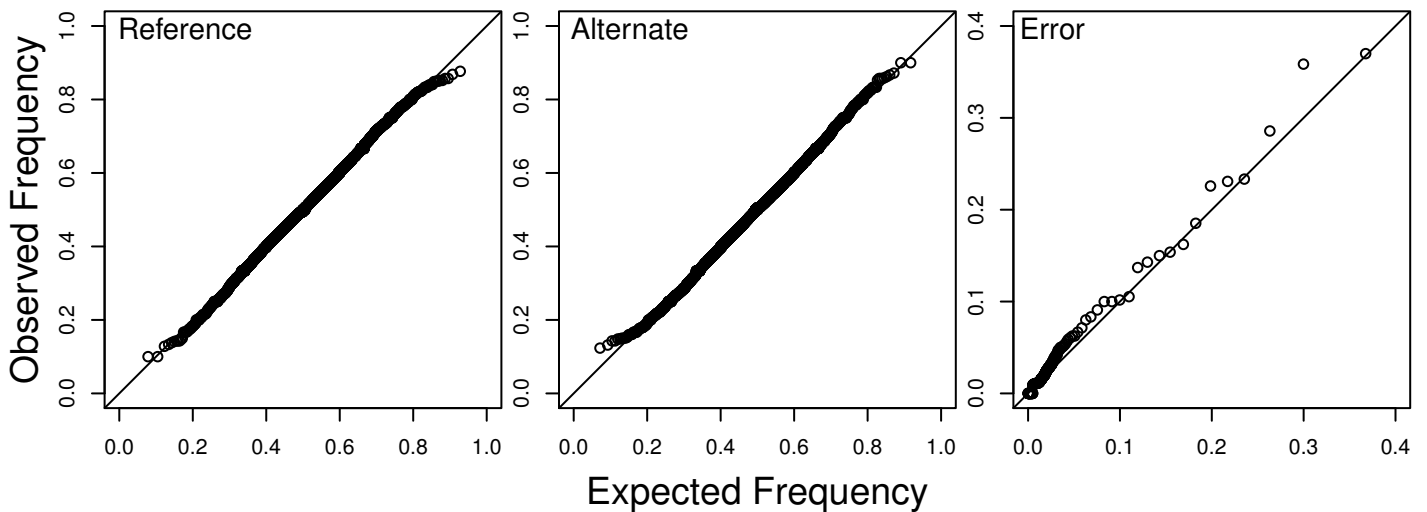
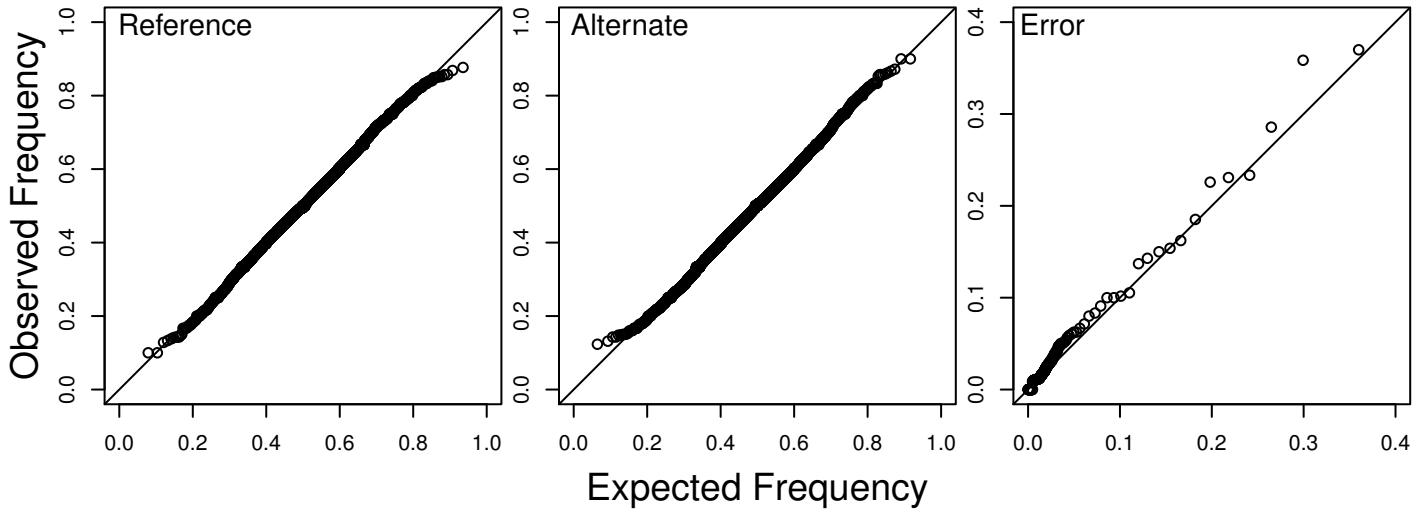


Figure S4 (Continued): **Mixtures of Dirichlet-multinomials provide the best fits to genomic data: CEU12 Chr21 TH.**

### Mixture of 5 Dirichlet–Multinomials



### Mixture of 6 Dirichlet–Multinomials

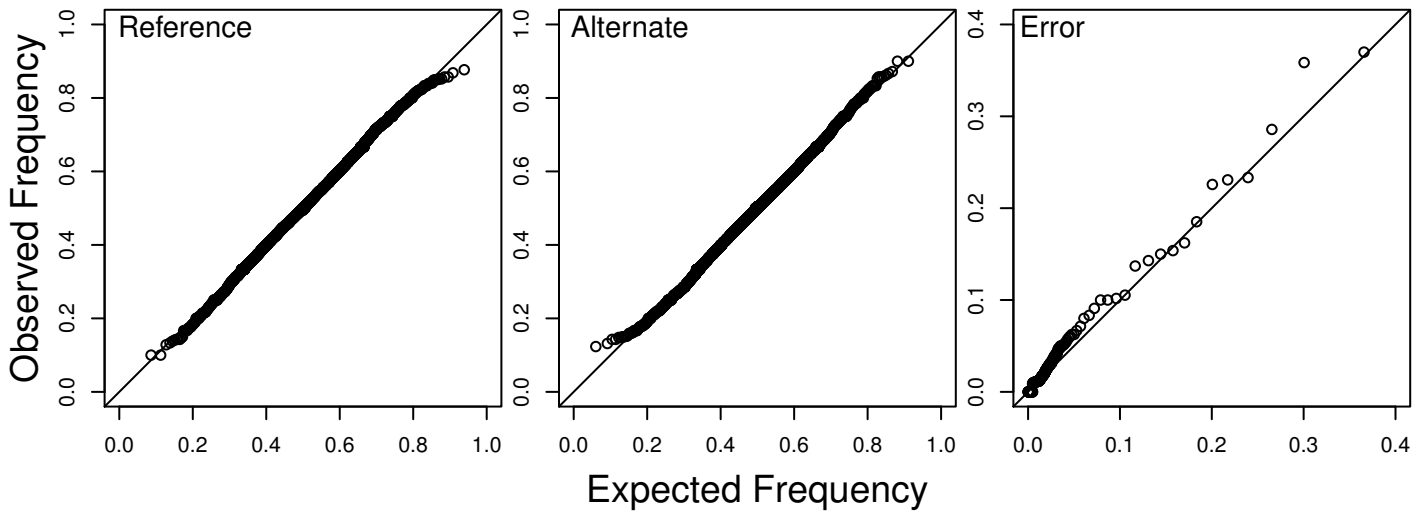
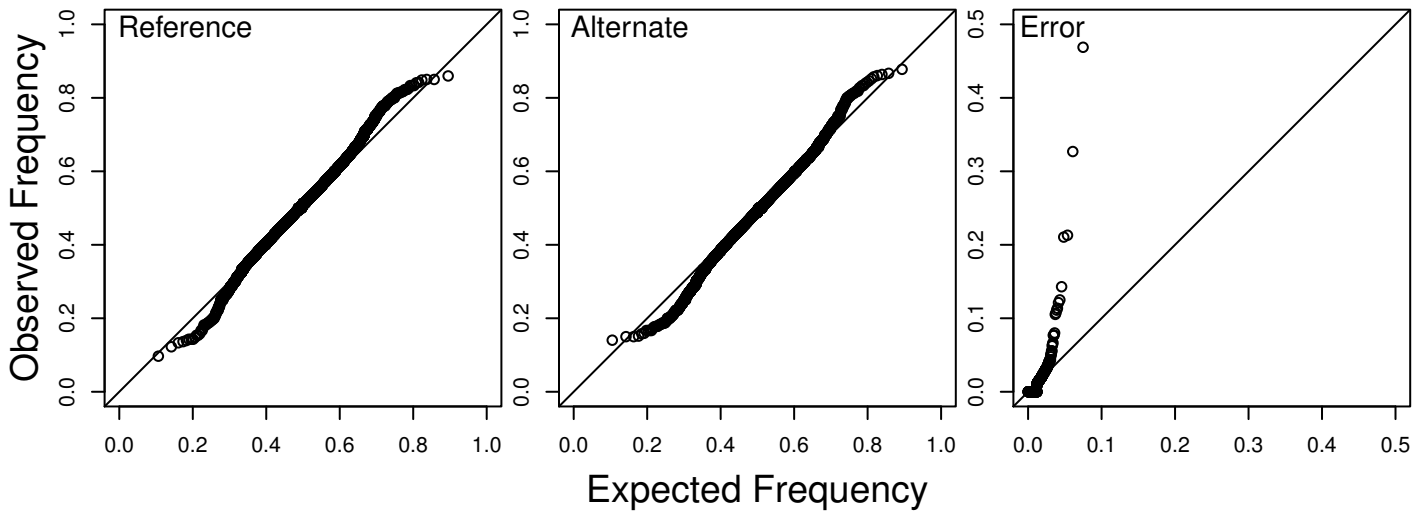
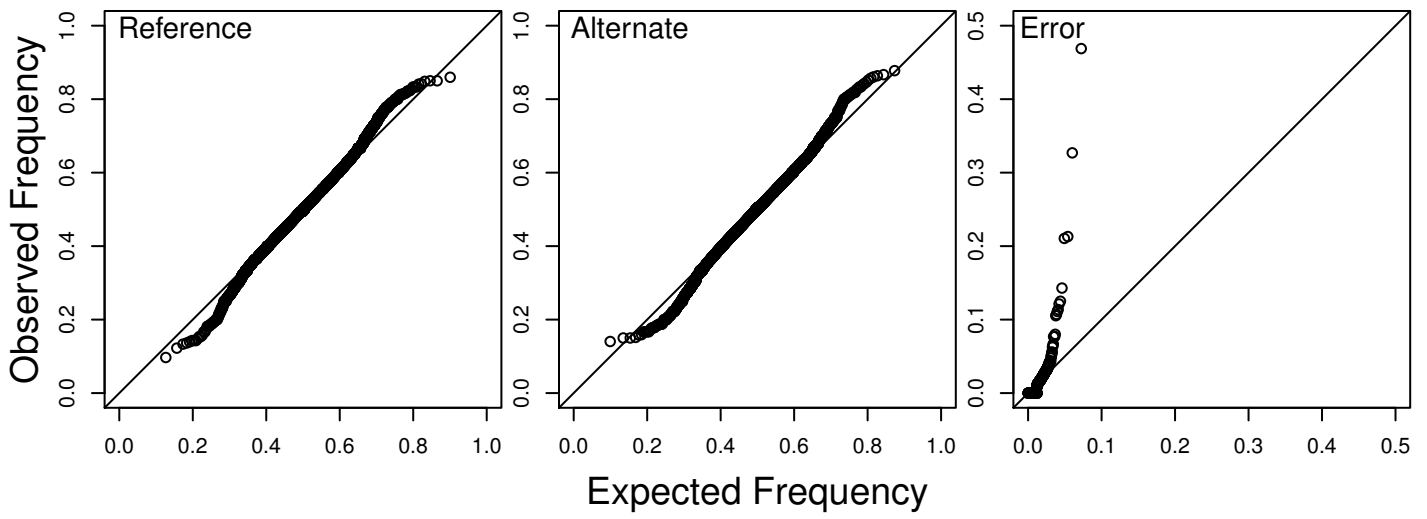


Figure S4 (Continued): **Mixtures of Dirichlet-multinomials provide the best fits to genomic data: CEU12 Chr21 TH.**

## Multinomial



## Biased Multinomial



## Dirichlet-Multinomial

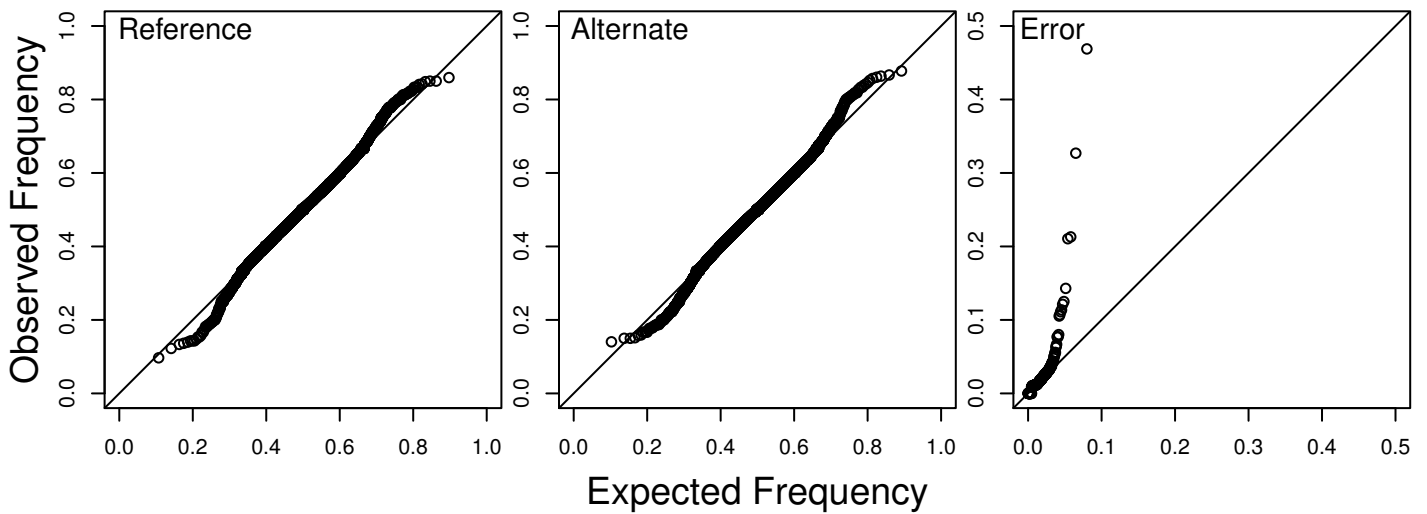
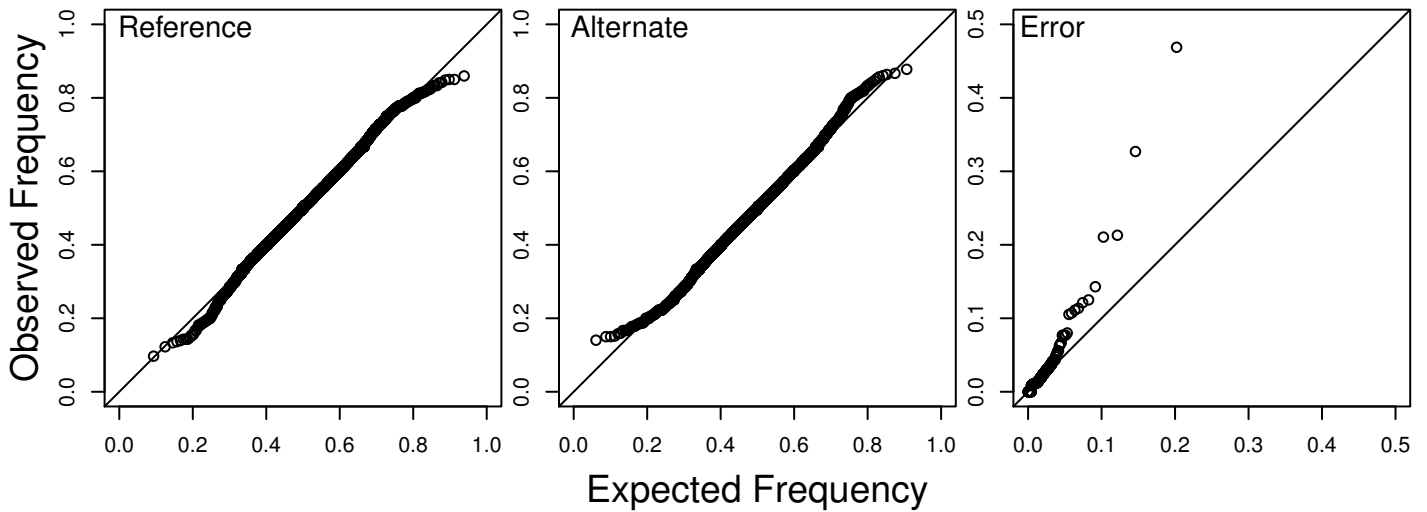
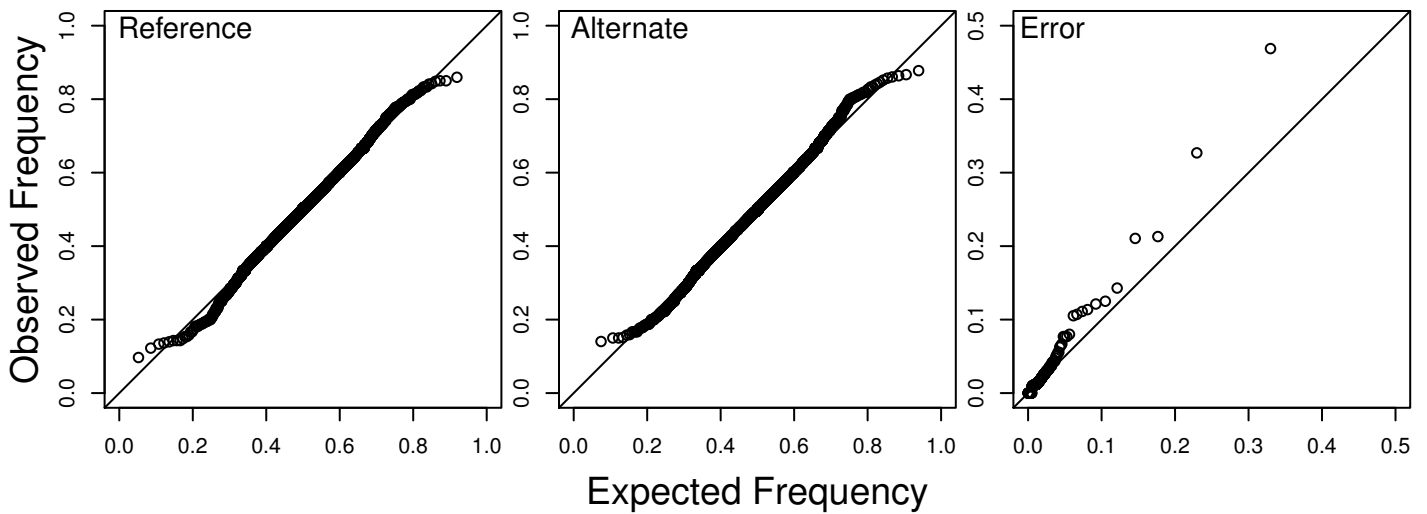


Figure S5: Mixtures of Dirichlet-multinomials provide the best fits to genomic data: CEU11 Chr10 TH.

### Mixture of 2 Dirichlet-Multinomials



### Mixture of 3 Dirichlet-Multinomials



### Mixture of 4 Dirichlet-Multinomials

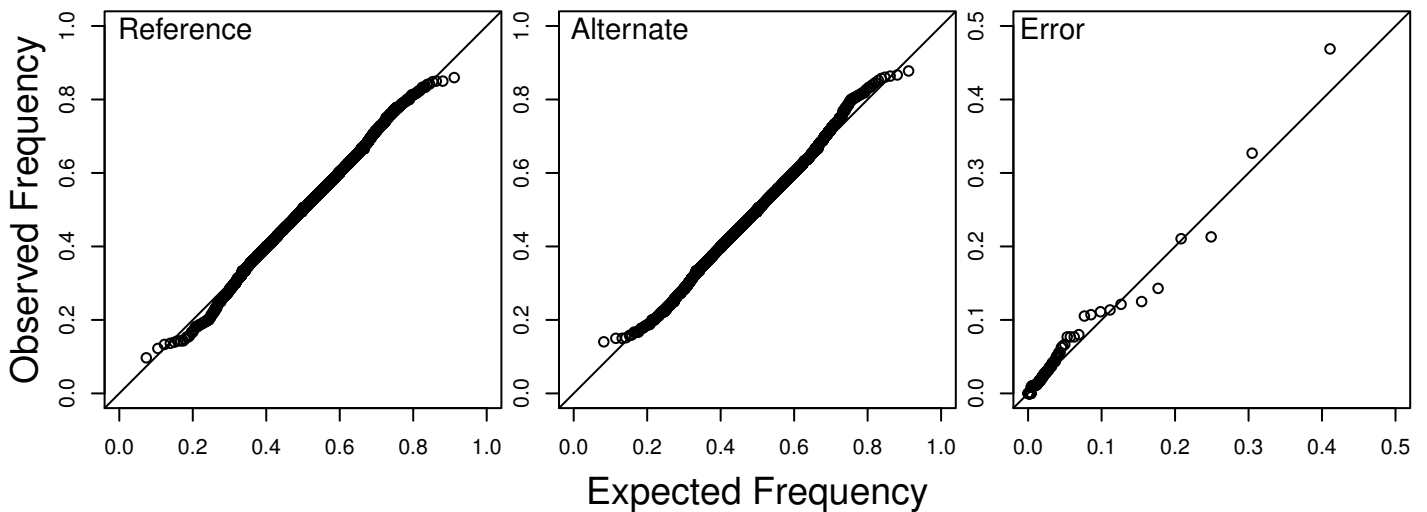
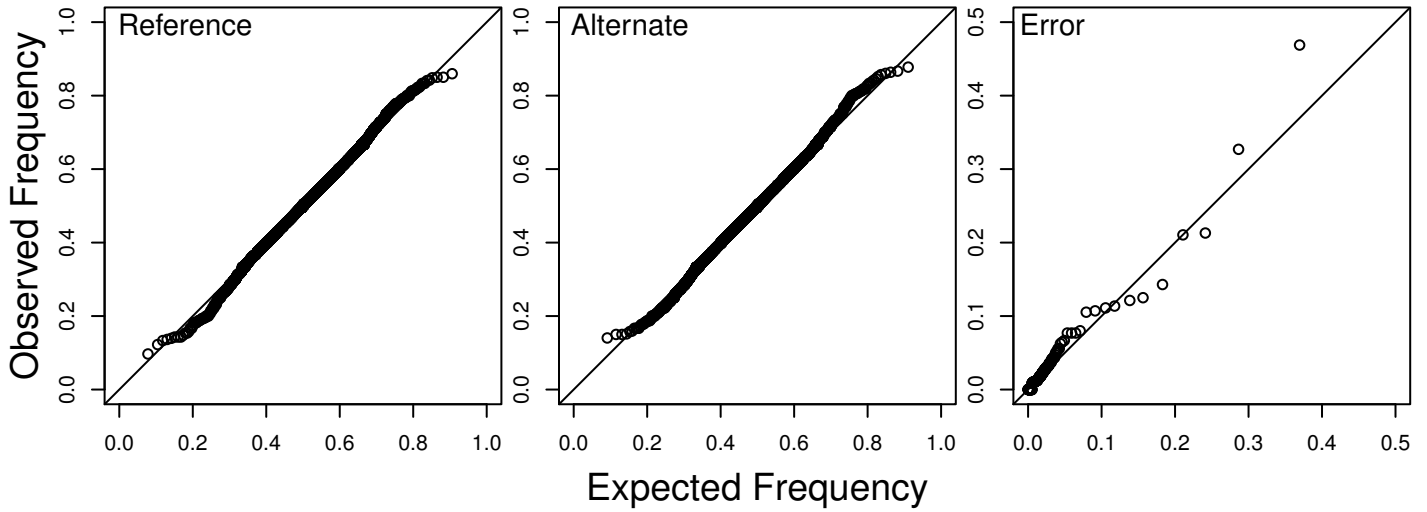


Figure S5 (Continued): **Mixtures of Dirichlet-multinomials provide the best fits to genomic data: CEU11 Chr10 TH.**

### Mixture of 5 Dirichlet–Multinomials



### Mixture of 6 Dirichlet–Multinomials

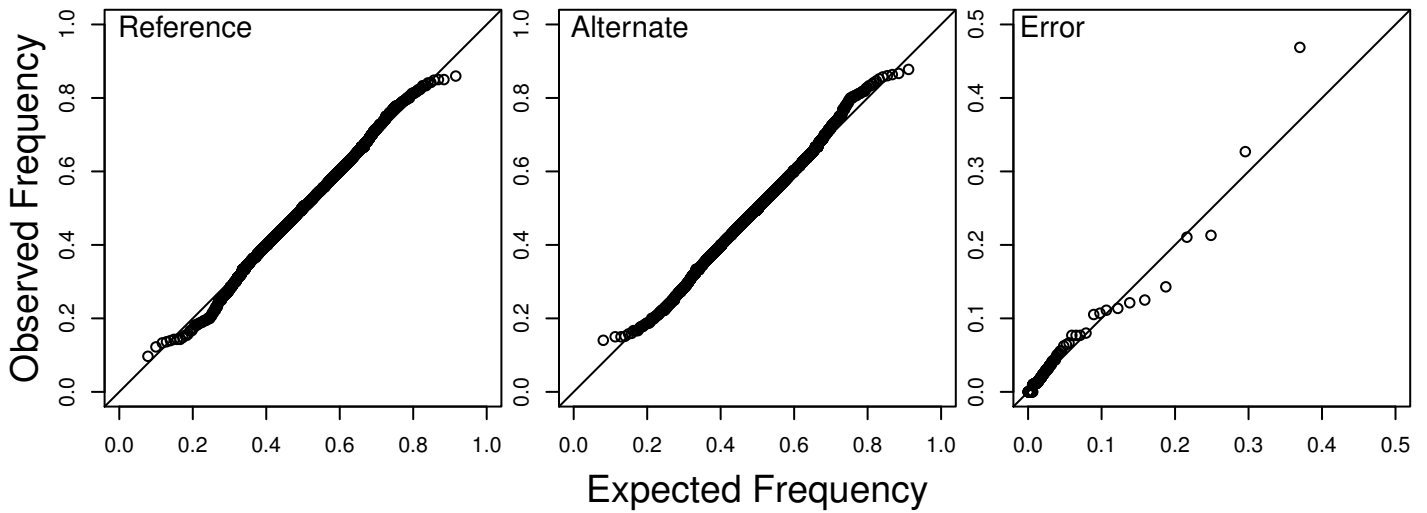
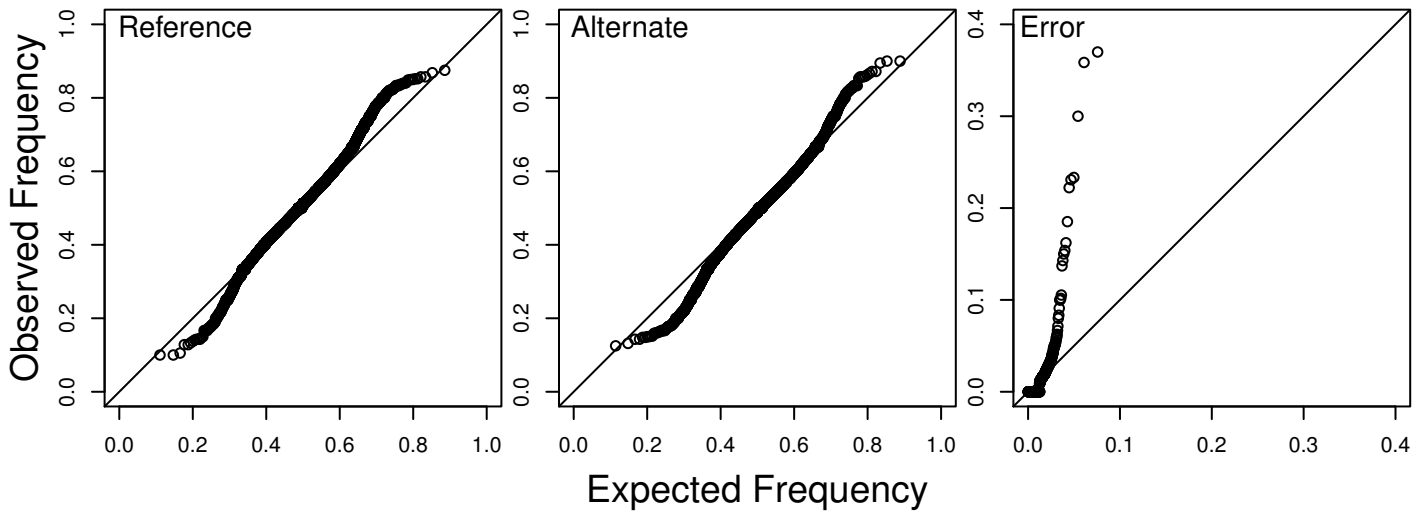
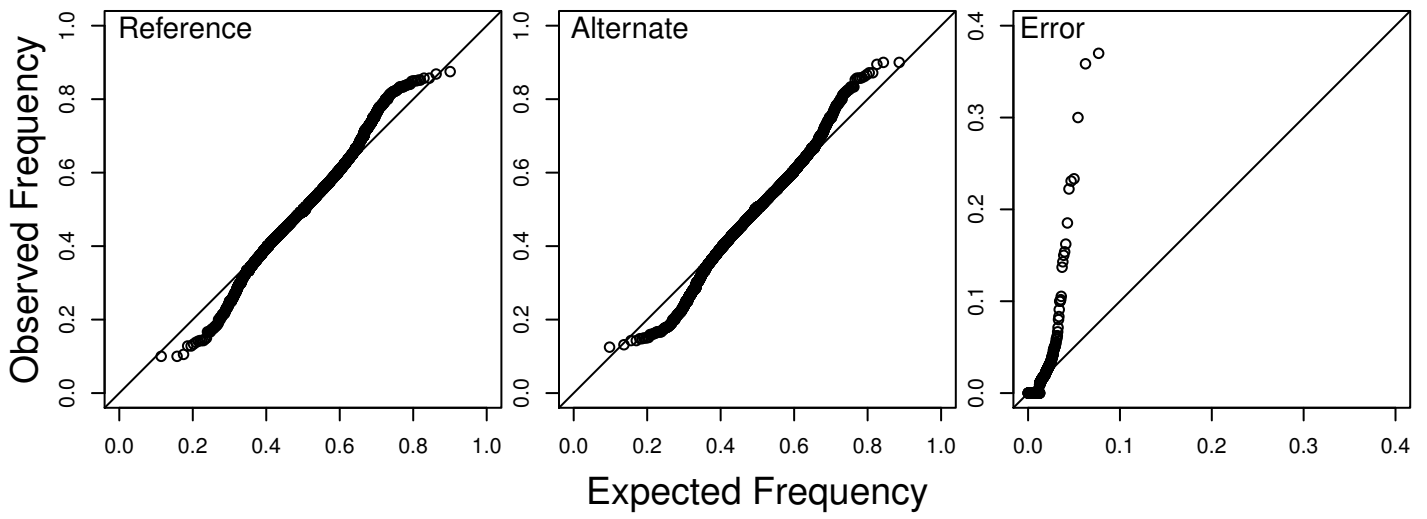


Figure S5 (Continued): **Mixtures of Dirichlet-multinomials provide the best fits to genomic data: CEU11 Chr10 TH.**

## Multinomial



## Biased Multinomial



## Dirichlet-Multinomial

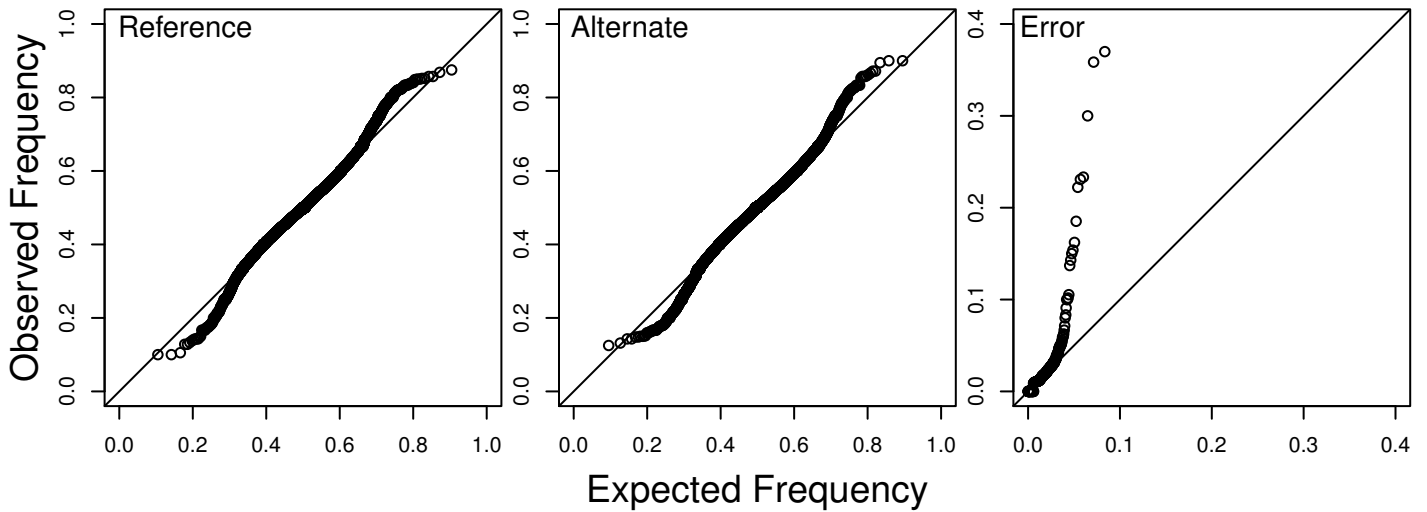
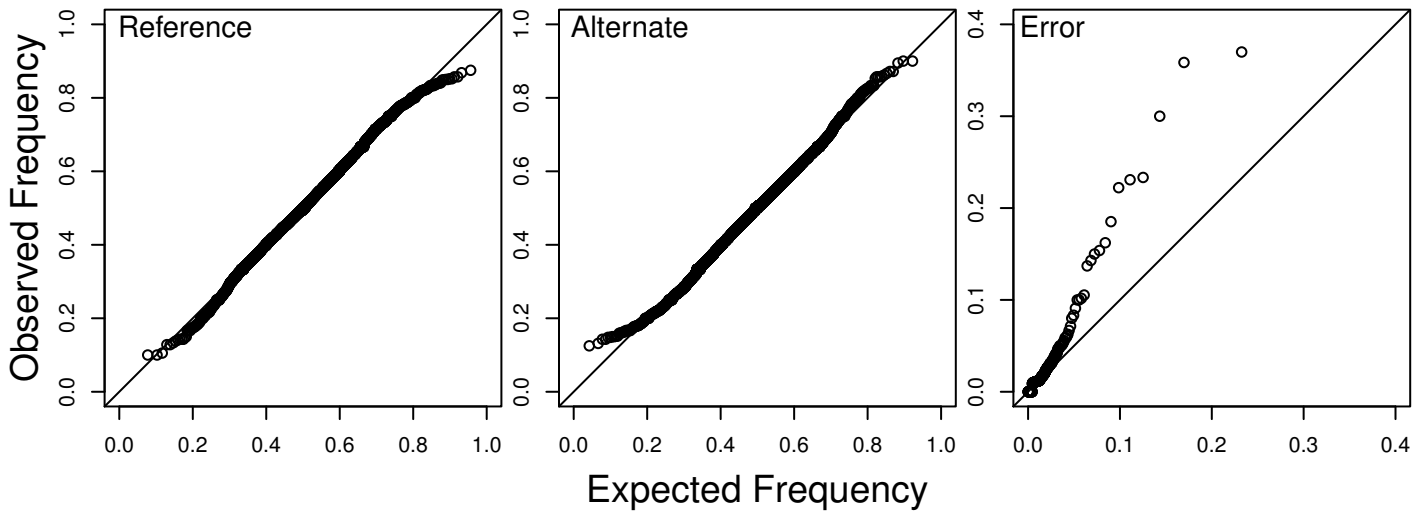


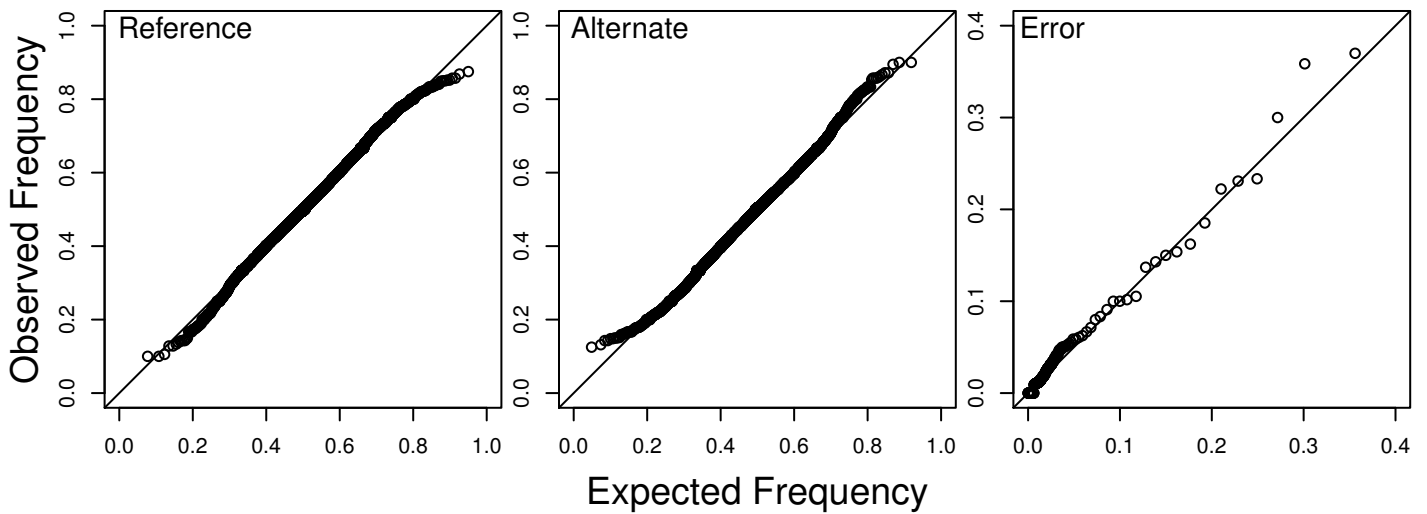
Figure S6: Mixtures of Dirichlet-multinomials provide the best fits to genomic data: CEU11 Chr21 TH.



### Mixture of 2 Dirichlet-Multinomials



### Mixture of 3 Dirichlet-Multinomials



### Mixture of 4 Dirichlet-Multinomials

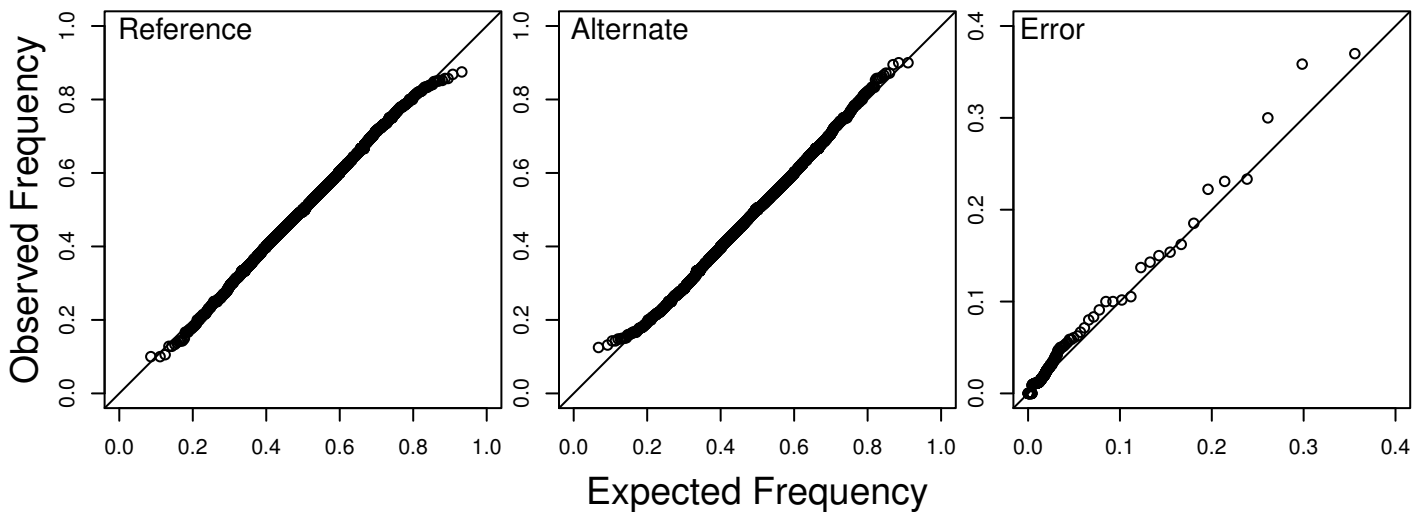
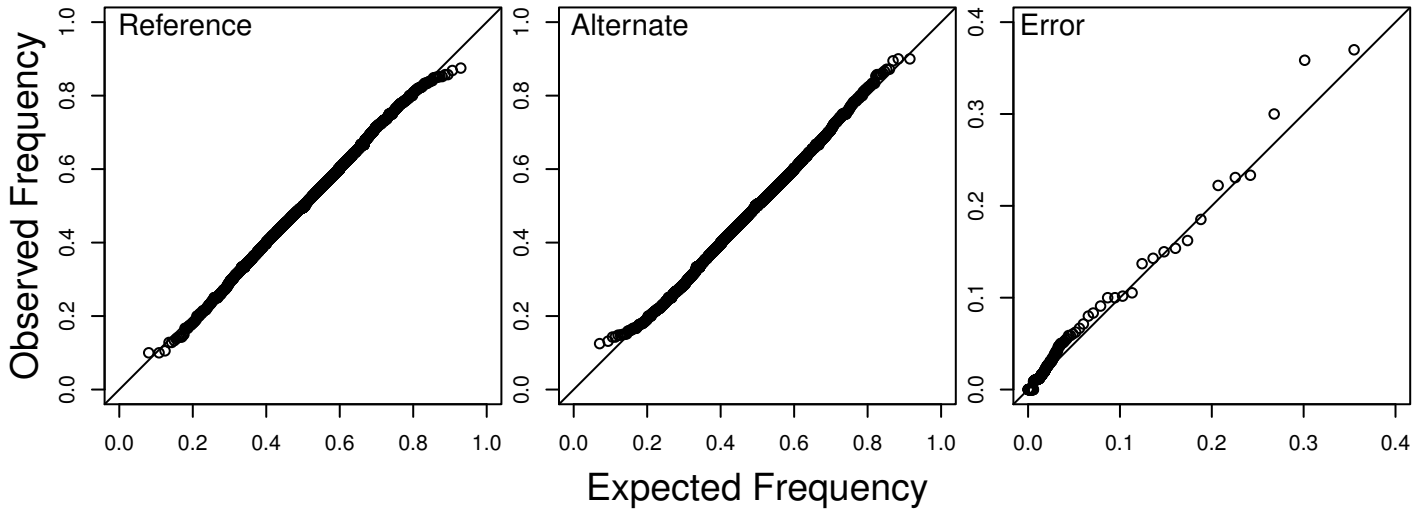


Figure S6 (Continued): **Mixtures of Dirichlet-multinomials provide the best fits to genomic data: CEU11 Chr21 TH.**

### Mixture of 5 Dirichlet–Multinomials



### Mixture of 6 Dirichlet–Multinomials

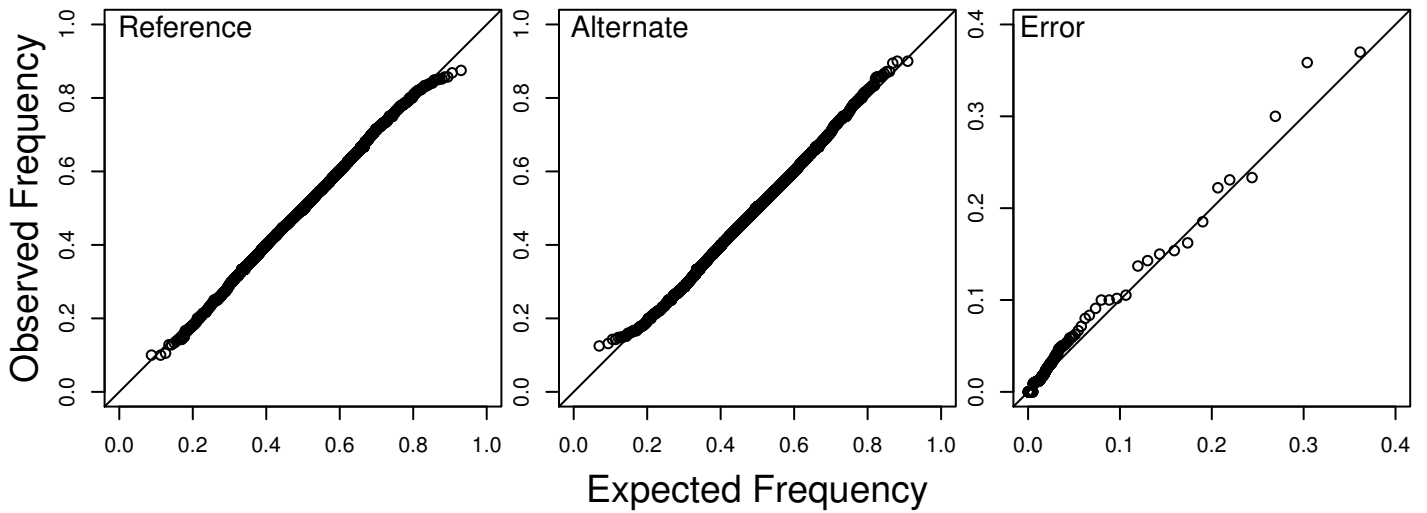
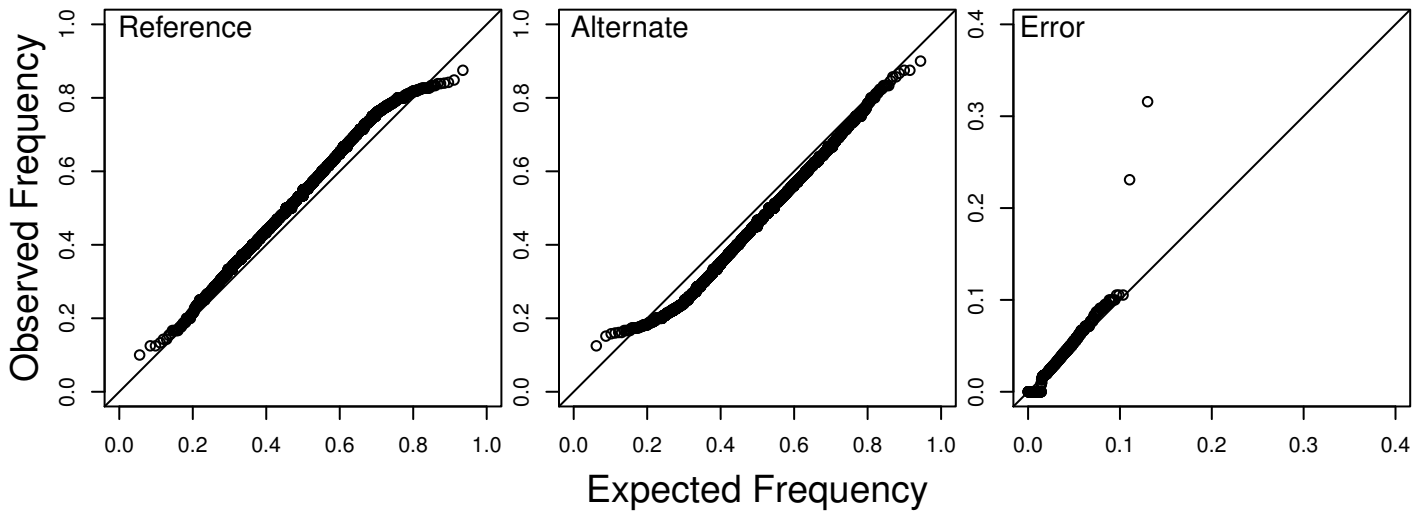
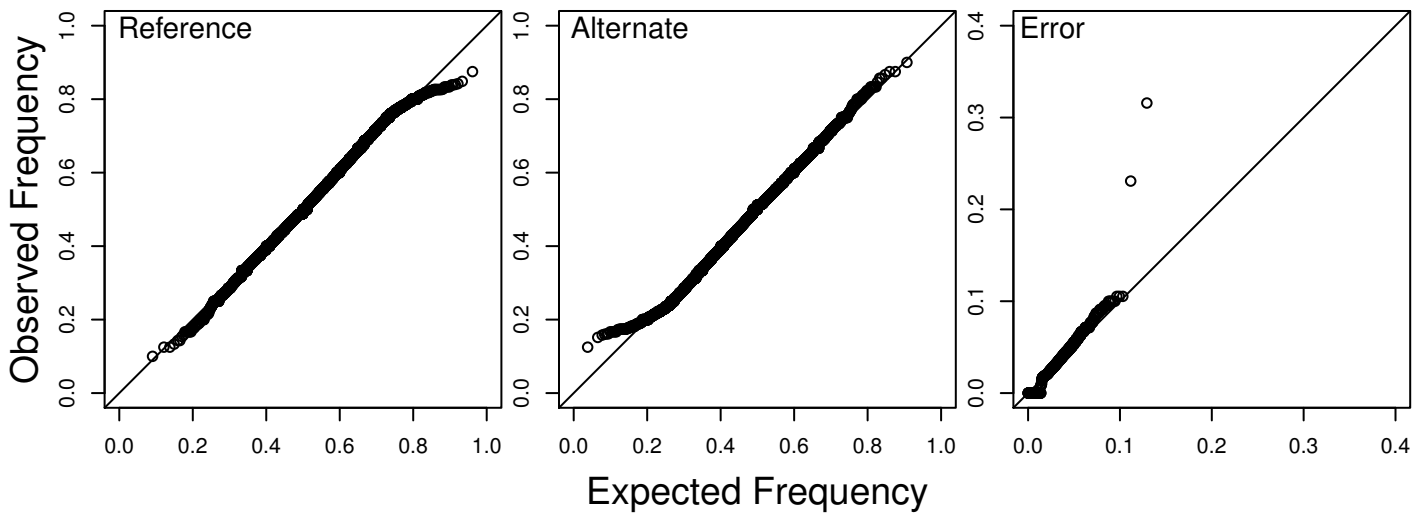


Figure S6 (Continued): **Mixtures of Dirichlet-multinomials provide the best fits to genomic data: CEU11 Chr21 TH.**

## Multinomial



## Biased Multinomial



## Dirichlet-Multinomial

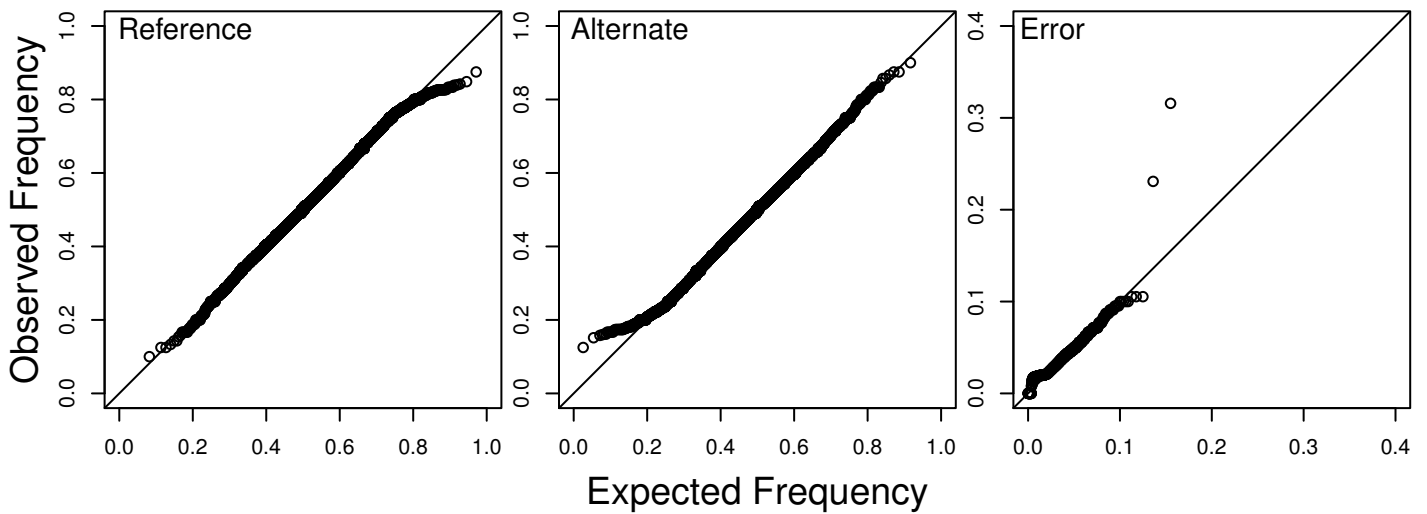
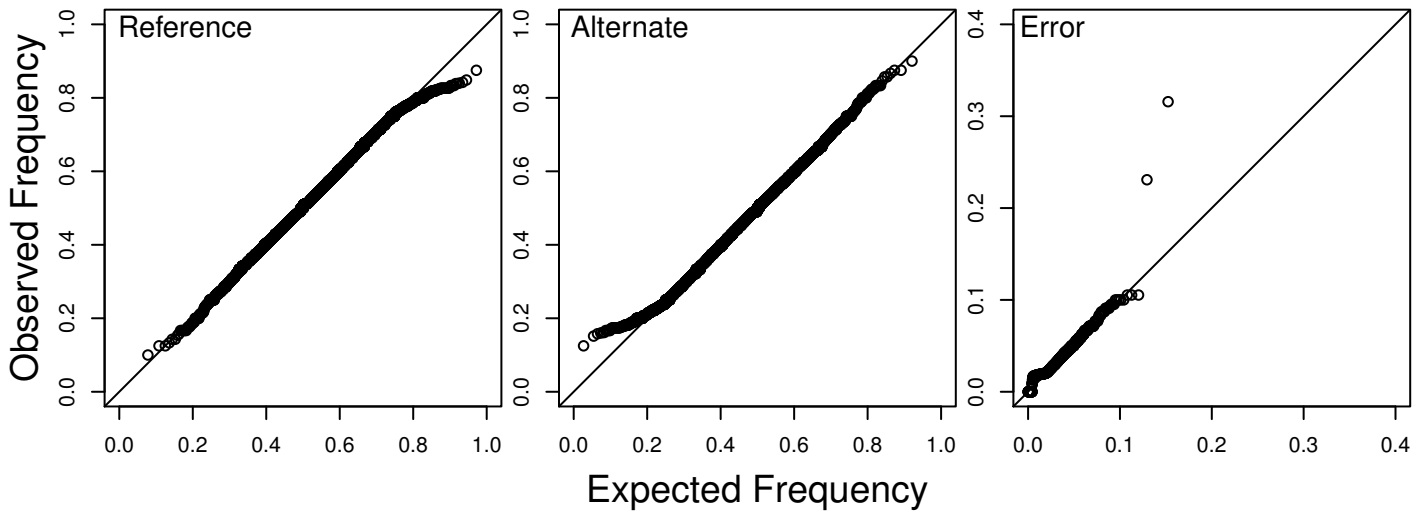
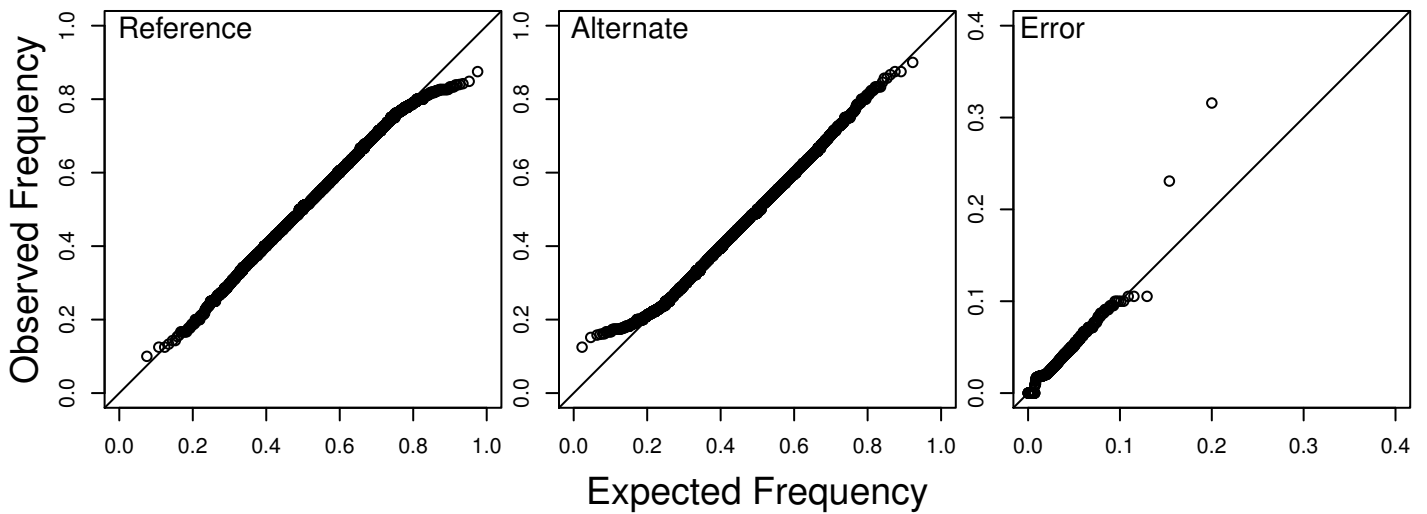


Figure S7: Mixtures of Dirichlet-multinomials provide the best fits to genomic data: CEU10 Chr10 TH.

### Mixture of 2 Dirichlet-Multinomials



### Mixture of 3 Dirichlet-Multinomials



### Mixture of 4 Dirichlet-Multinomials

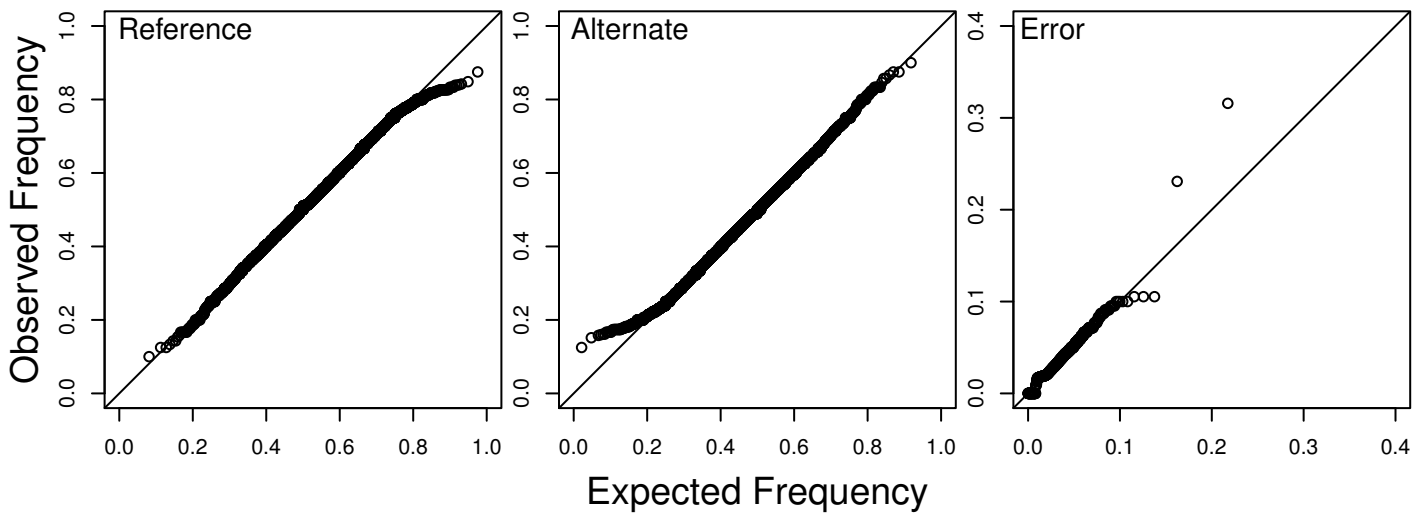
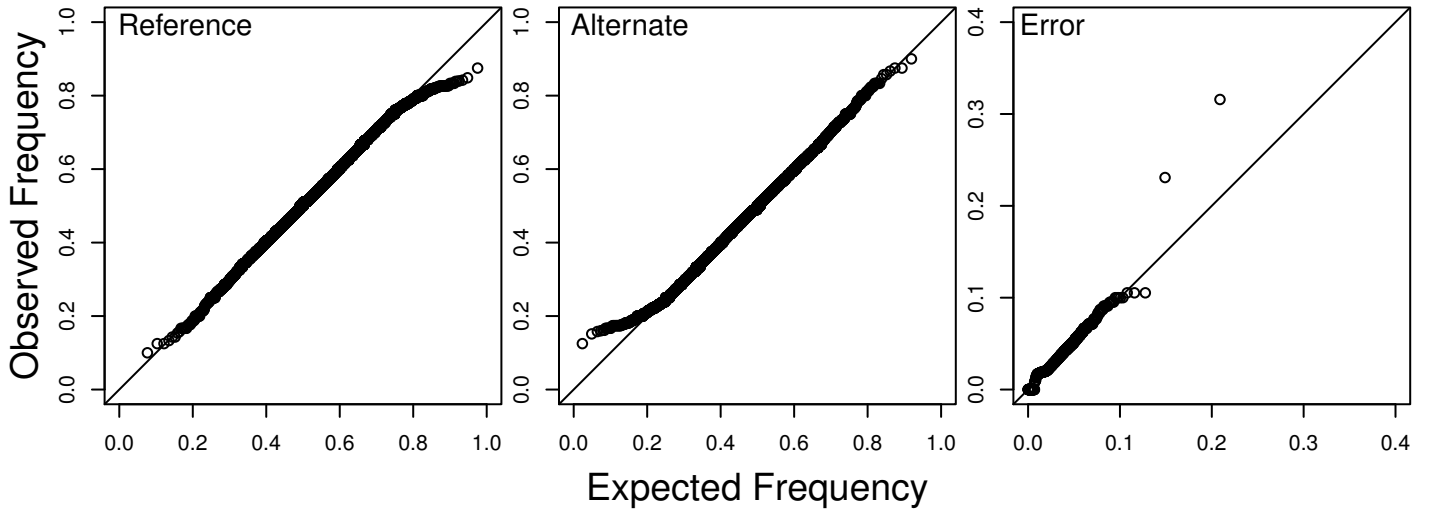


Figure S7 (Continued): **Mixtures of Dirichlet-multinomials provide the best fits to genomic data: CEU10 Chr10 TH.**

### Mixture of 5 Dirichlet–Multinomials



### Mixture of 6 Dirichlet–Multinomials

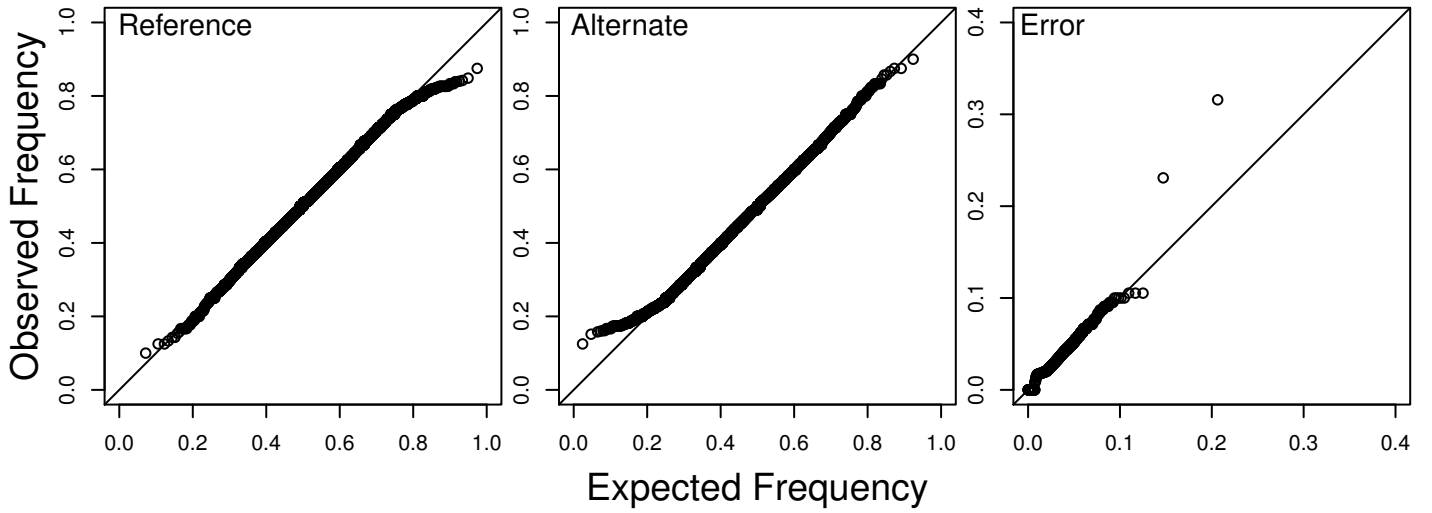
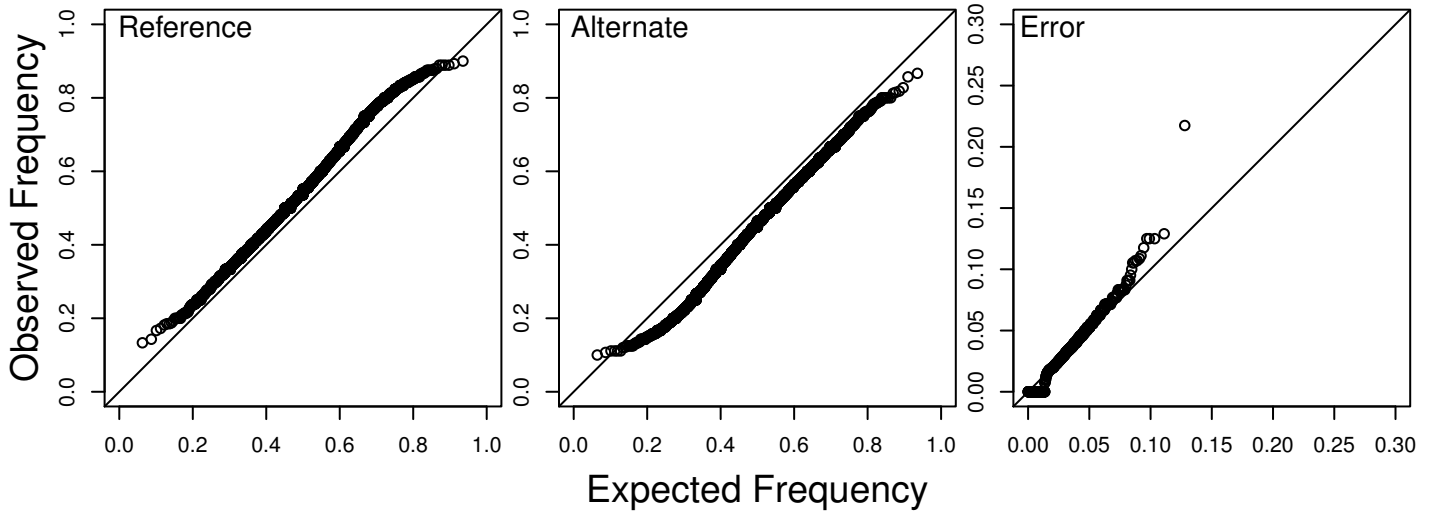
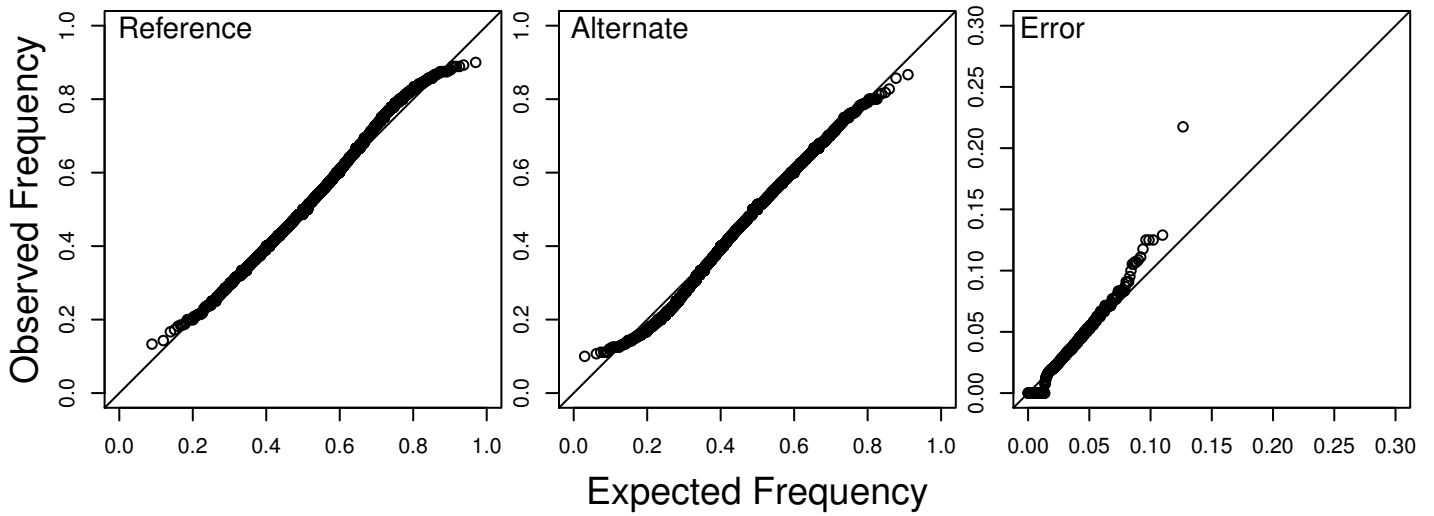


Figure S7 (Continued): **Mixtures of Dirichlet-multinomials provide the best fits to genomic data: CEU10 Chr10 TH.**

## Multinomial



## Biased Multinomial



## Dirichlet-Multinomial

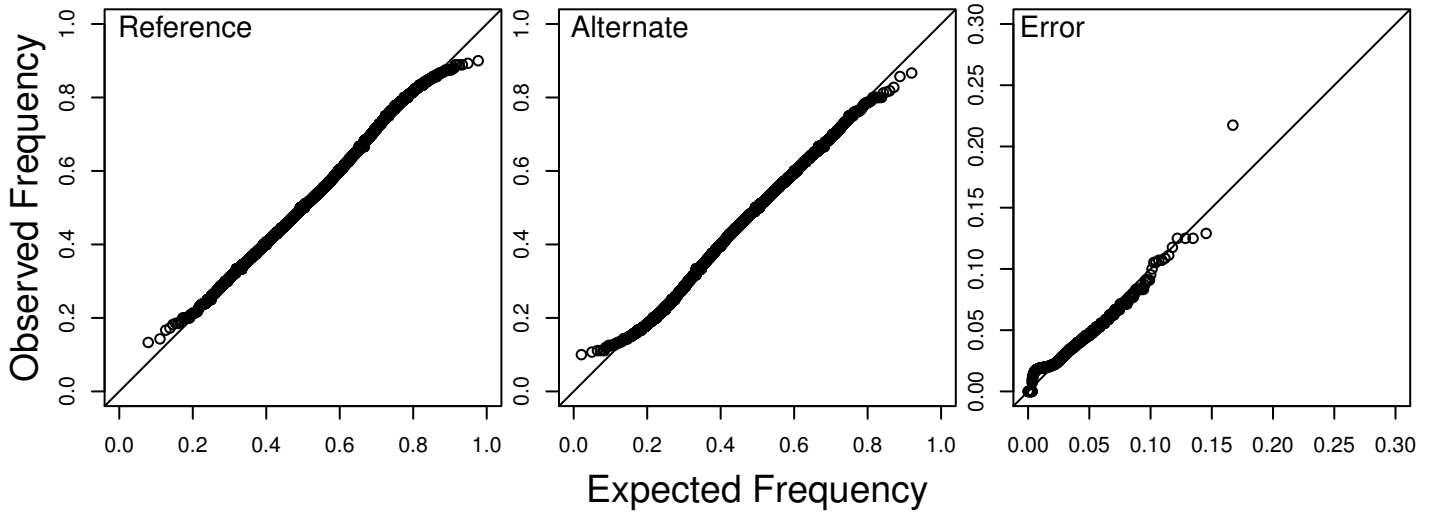
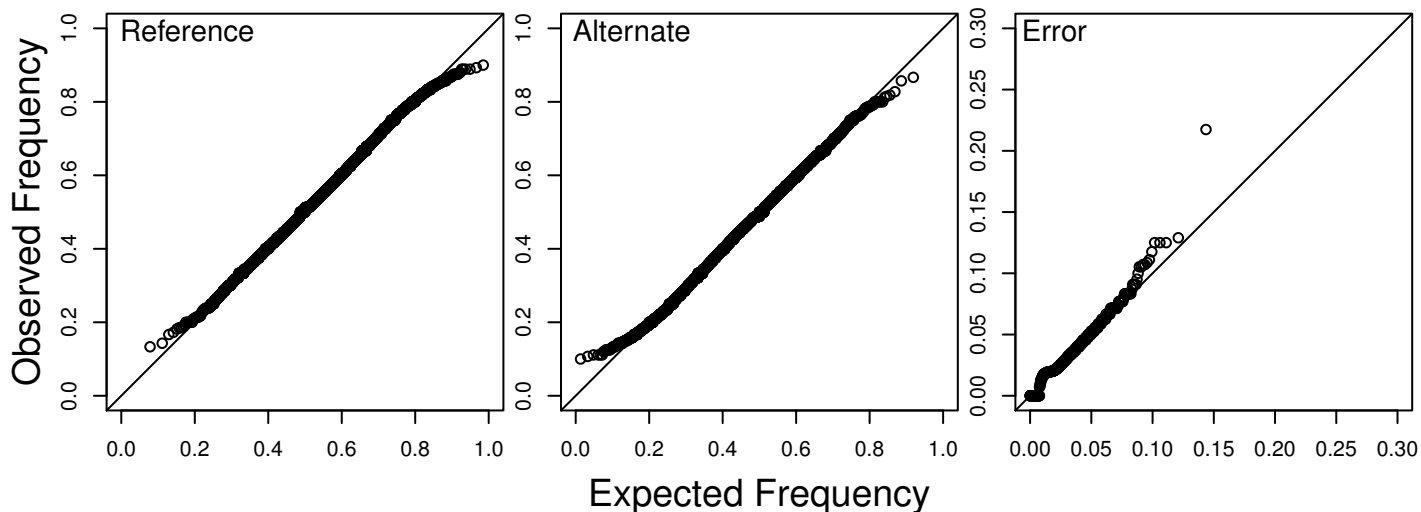
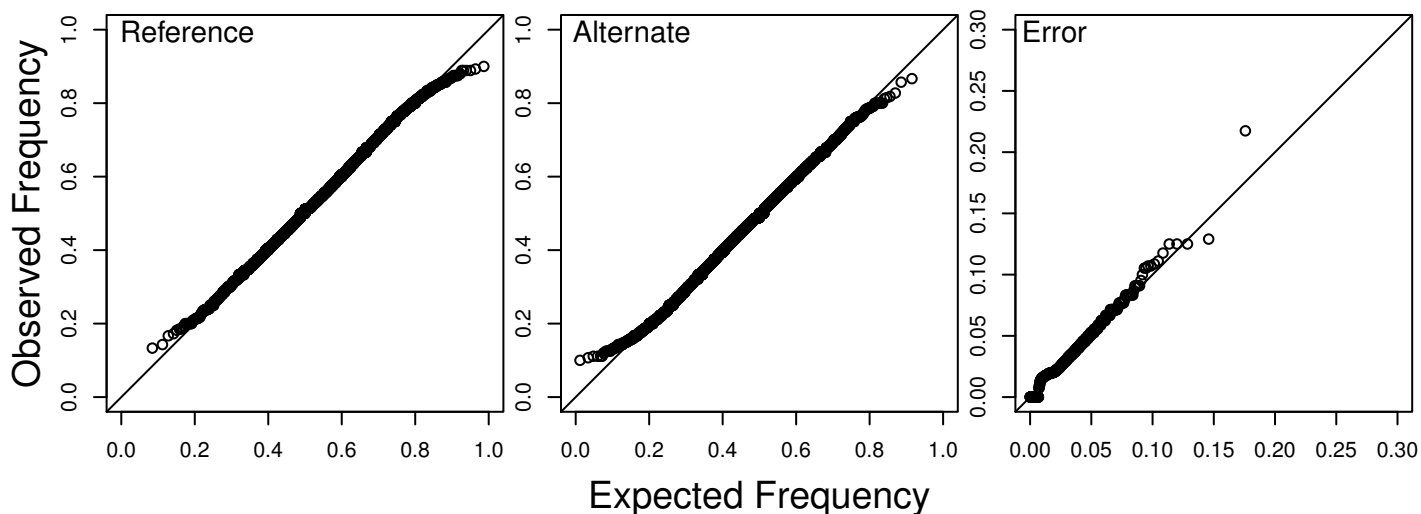


Figure S8: Mixtures of Dirichlet-multinomials provide the best fits to genomic data: CEU10 Chr21 TH.

### Mixture of 2 Dirichlet-Multinomials



### Mixture of 3 Dirichlet-Multinomials



### Mixture of 4 Dirichlet-Multinomials

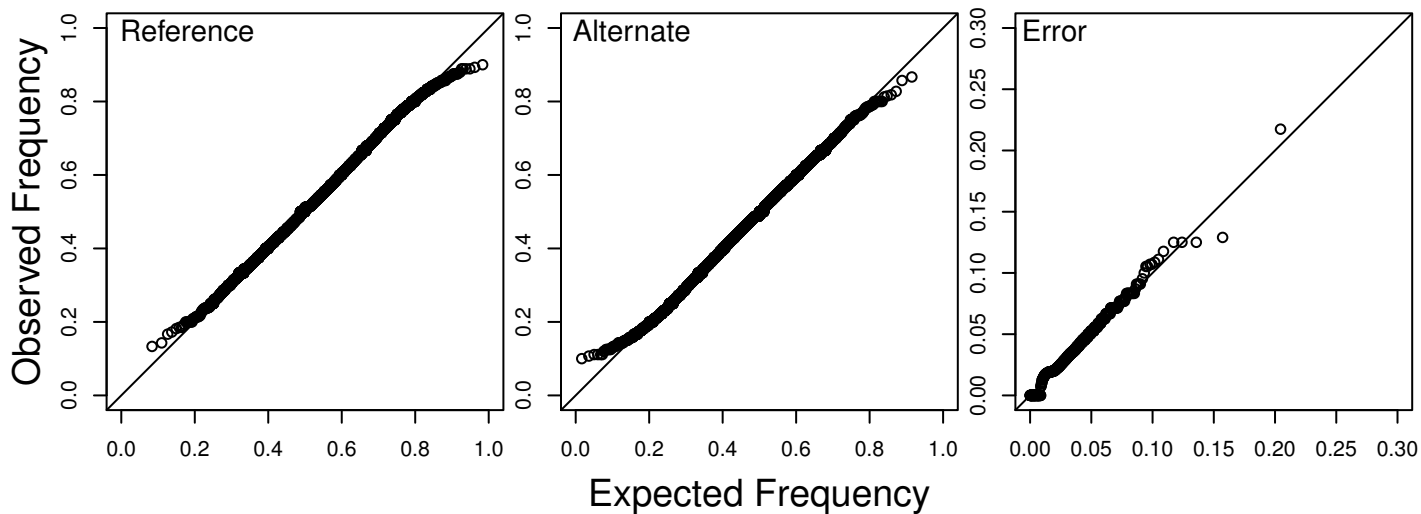
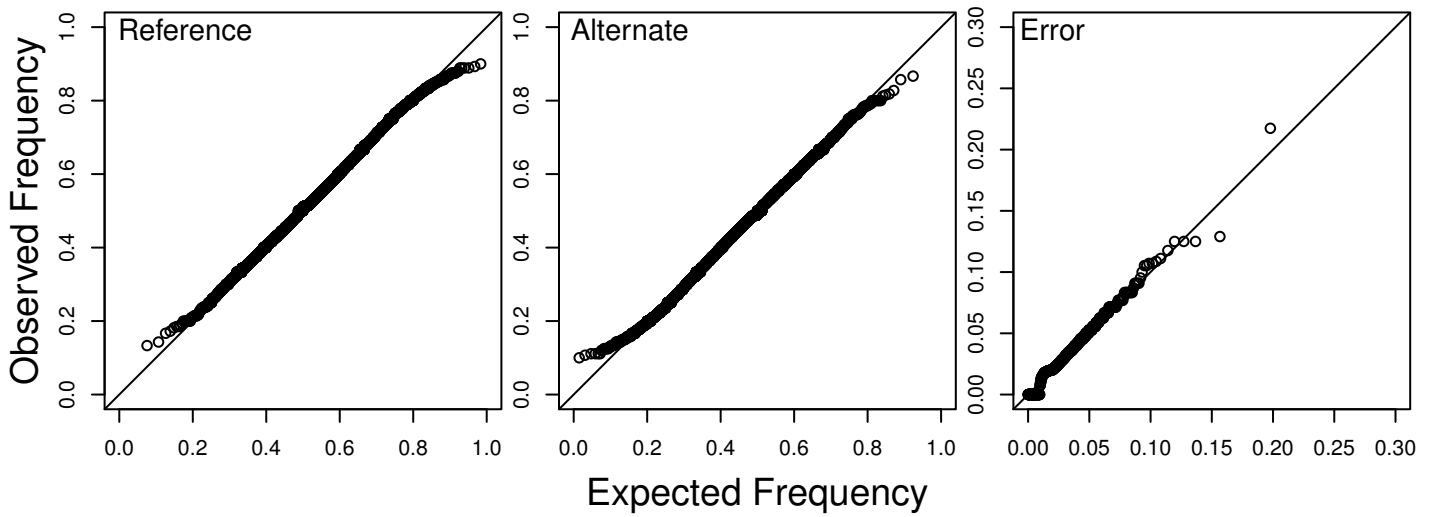


Figure S8 (Continued): **Mixtures of Dirichlet-multinomials provide the best fits to genomic data: CEU10 Chr21 TH.**

### Mixture of 5 Dirichlet–Multinomials



### Mixture of 6 Dirichlet–Multinomials

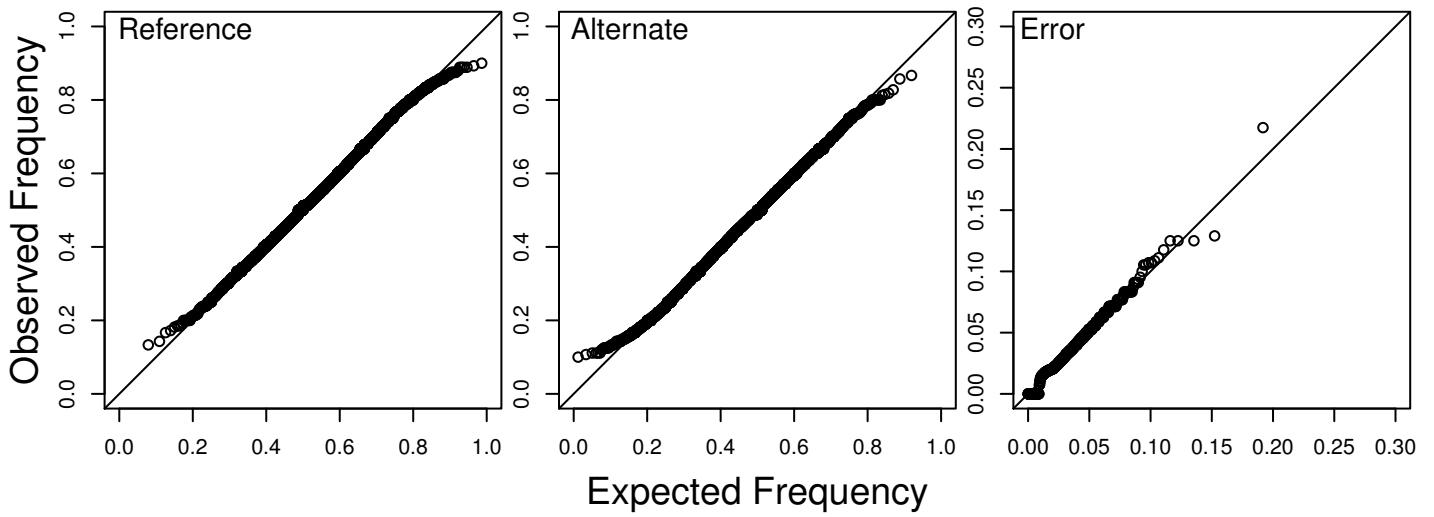


Figure S8 (Continued): **Mixtures of Dirichlet-multinomials provide the best fits to genomic data: CEU10 Chr21 TH.**



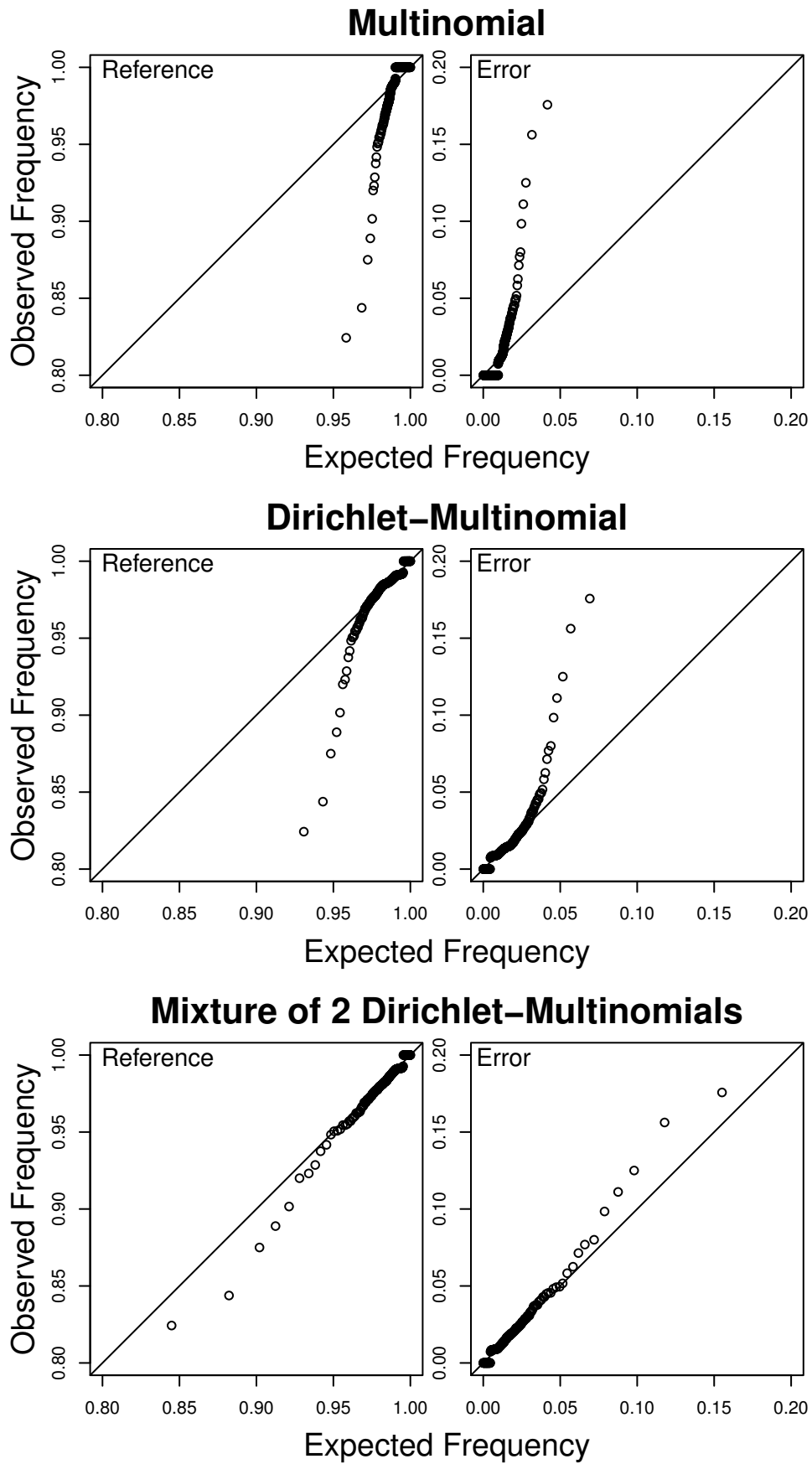
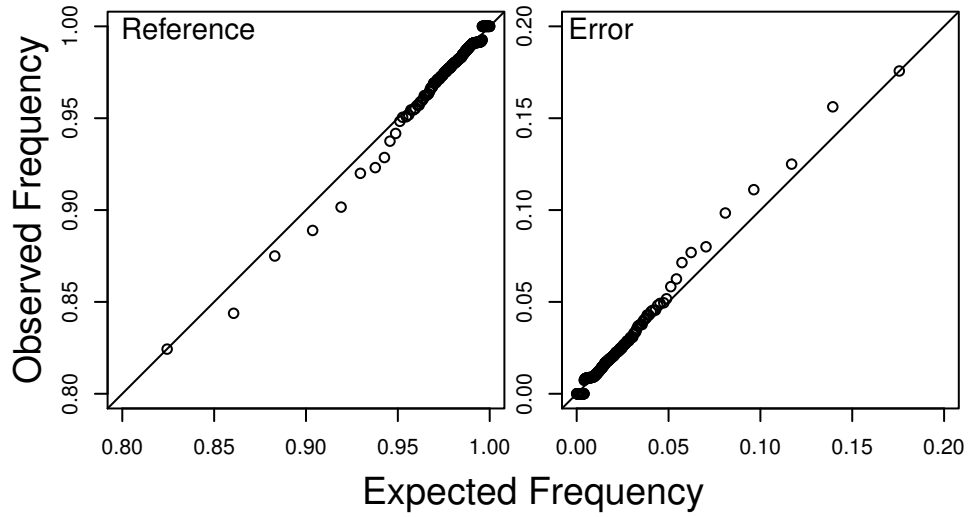
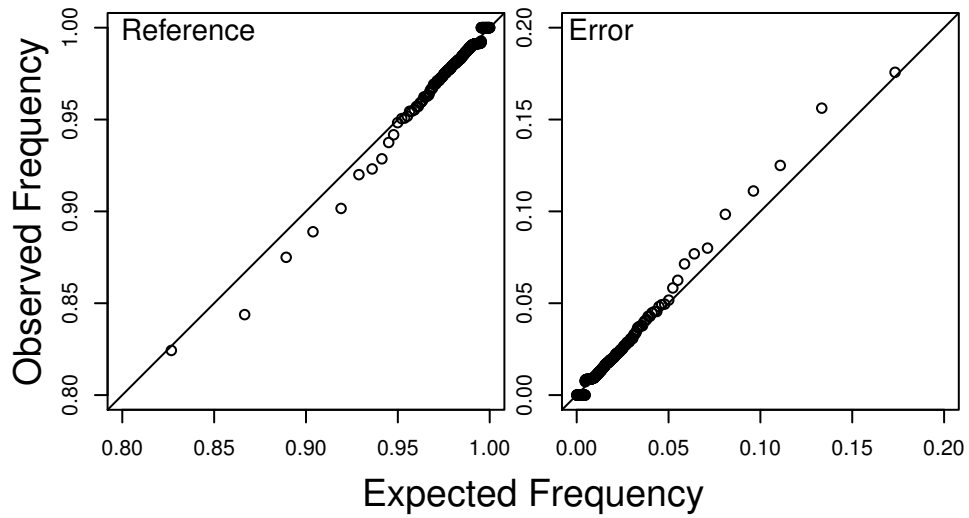


Figure S9: **Mixtures of Dirichlet-multinomials provide the best fits to genomic data: CHM1 Chr10 RD.**

### Mixture of 3 Dirichlet-Multinomials



### Mixture of 4 Dirichlet-Multinomials



### Mixture of 5 Dirichlet-Multinomials

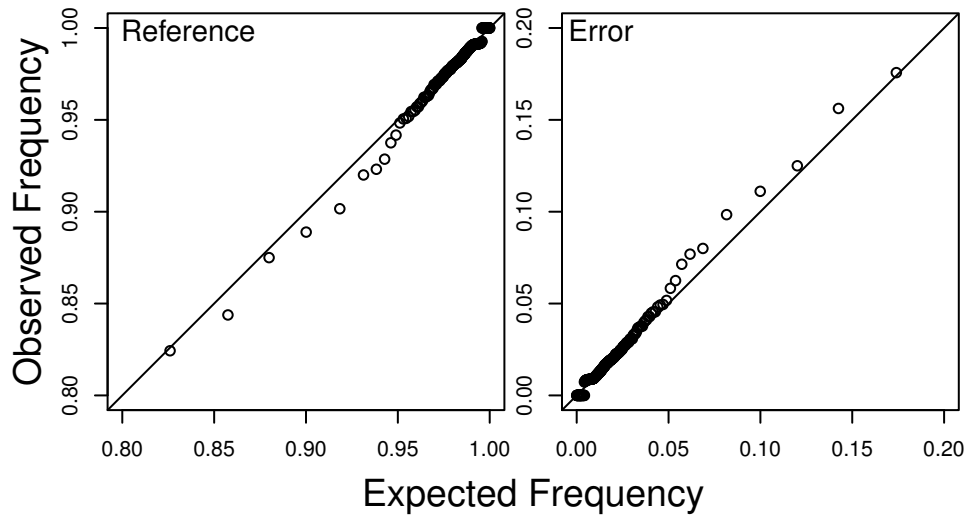


Figure S9 (Continued): **Mixtures of Dirichlet-multinomials provide the best fits to genomic data: CHM1 Chr10 RD.**

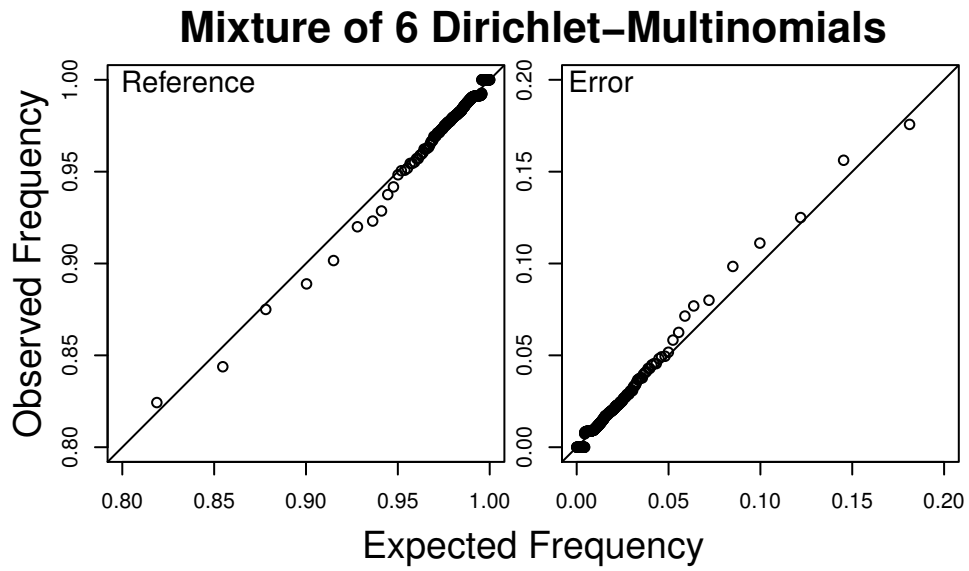
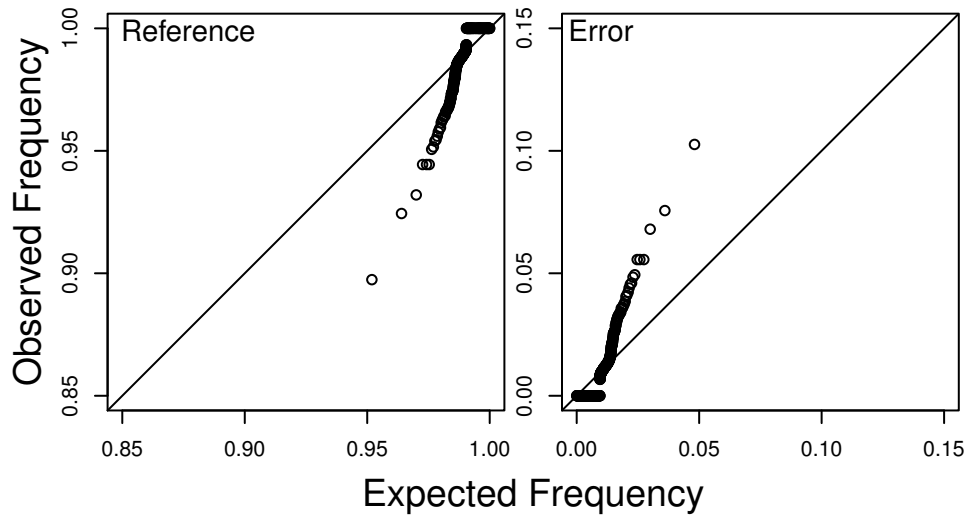
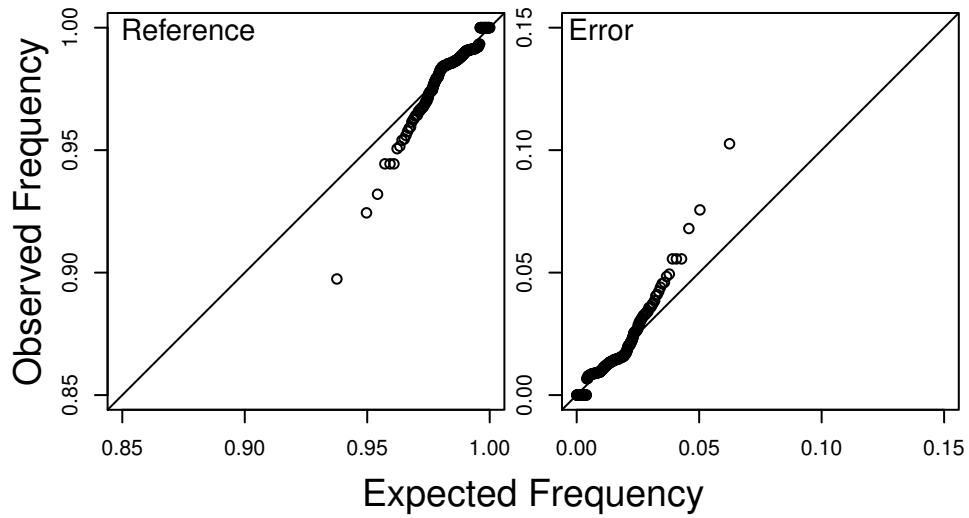


Figure S9 (Continued): **Mixtures of Dirichlet-multinomials provide the best fits to genomic data: CHM1 Chr10 RD.**

## Multinomial



## Dirichlet-Multinomial



## Mixture of 2 Dirichlet-Multinomials

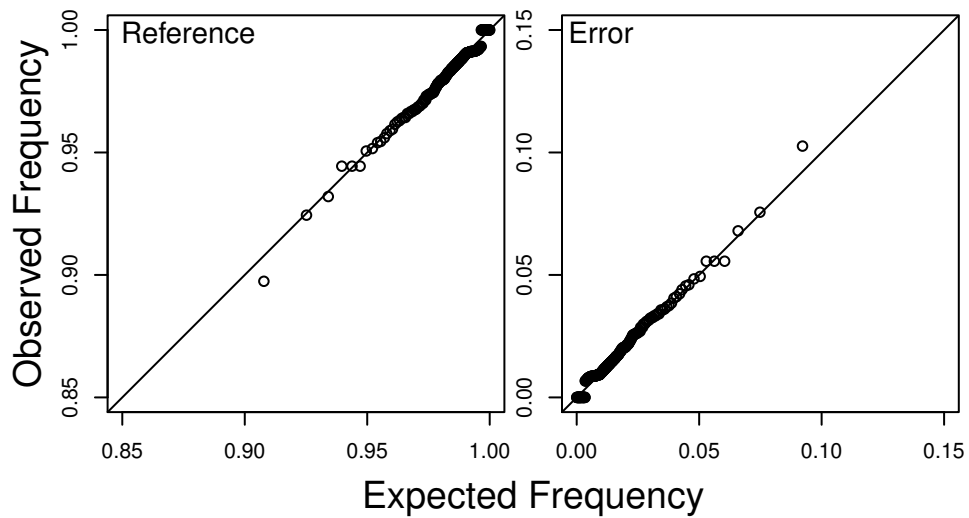
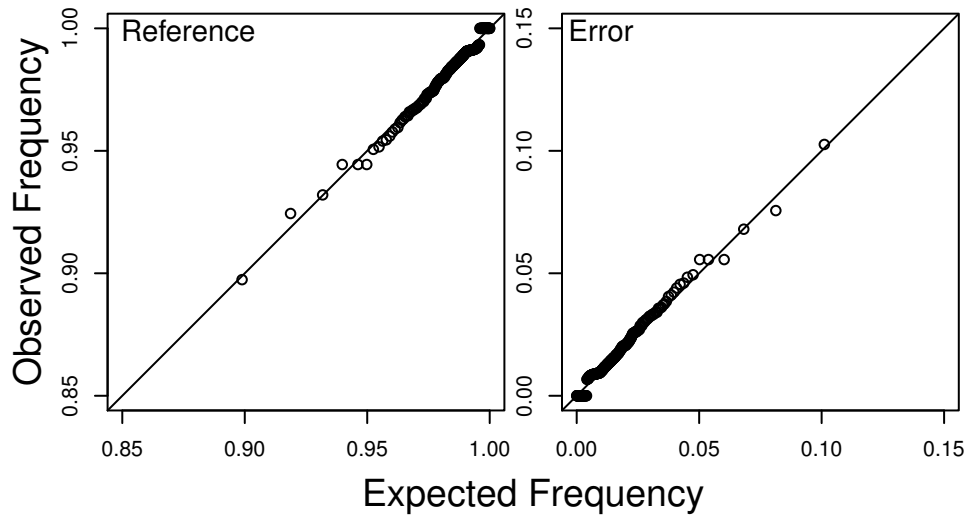
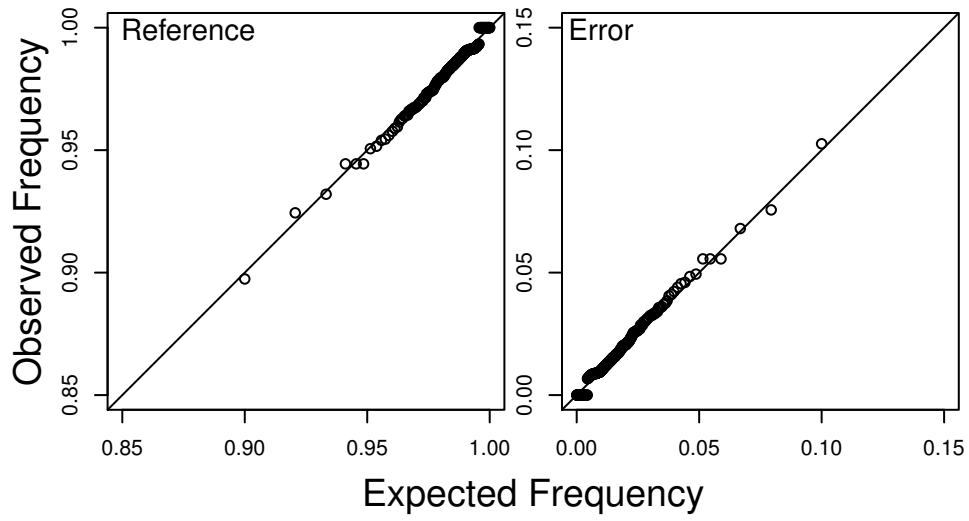


Figure S10: Mixtures of Dirichlet-multinomials provide the best fits to genomic data: CHM1 Chr21 RD.

### Mixture of 3 Dirichlet-Multinomials



### Mixture of 4 Dirichlet-Multinomials



### Mixture of 5 Dirichlet-Multinomials

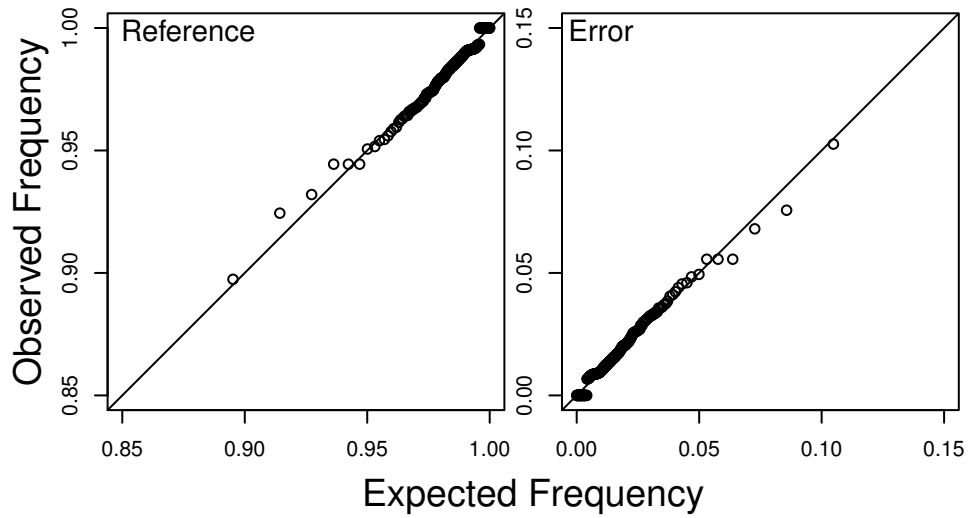


Figure S10 (Continued): **Mixtures of Dirichlet-multinomials provide the best fits to genomic data: CHM1 Chr21 RD.**

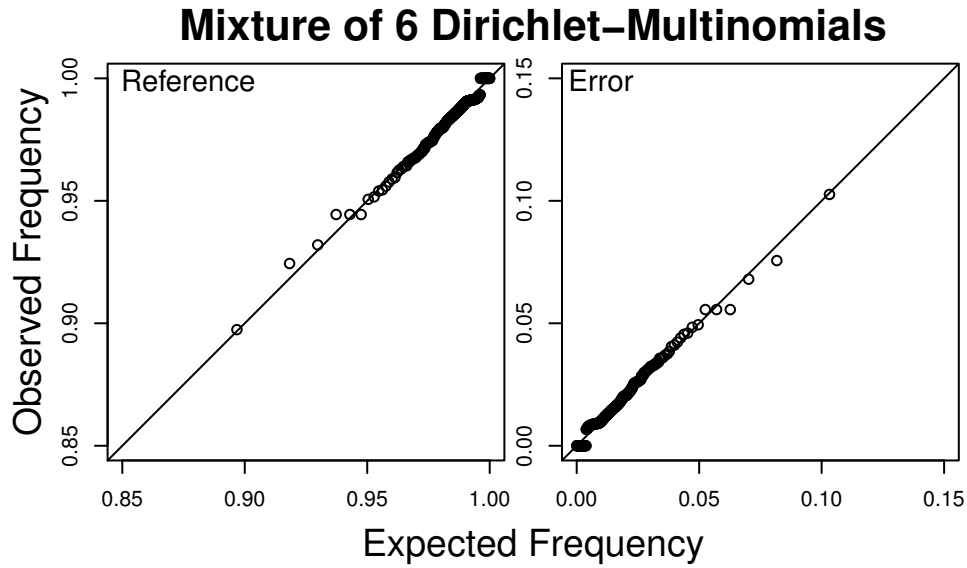


Figure S10 (Continued): **Mixtures of Dirichlet-multinomials provide the best fits to genomic data: CHM1 Chr21 RD.**

Table S1: **Number of heterozygous sites identified by different methods and the number of true heterozygous sites from 1000 Genomes Project.**

Dataset	Individual caller only	Trio caller only	Both callers (PH)	True heterozygotes (TH)
CEU13 Chr21	143	1604	40956	30120
CEU13 Chr10	1652	2645	38542	36590
CEU12 Chr21	151	1171	38180	29983
CEU12 Chr10	106	880	40144	36818
CEU11 Chr21	145	1190	38107	29991
CEU11 Chr10	114	867	40156	36825
CEU10 Chr21	6447	4971	31773	28197
CEU10 Chr10	194	1173	37108	35062

Table S2: **Maximum likelihood results for different Dirichlet-Multinomial mixture models on homozygous data.**

FD: Full homozygous dataset; RD: Reference homozygous dataset;  $k$ : Number of components in the mixture; Bolded: Best model by AIC or BIC.

	$k$	FD lnL	RD lnL	FD AIC	RD AIC	FD BIC	RD BIC
CHM1 Chr10	1	-9741.81	-8820.24	19489.62	17646.48	19515.40	17672.26
	2	-8941.66	-8797.67	17897.32	<b>17609.34</b>	<b>17957.48</b>	<b>17669.49</b>
	3	-8932.63	-8796.54	<b>17887.27</b>	17615.07	17981.80	17709.60
	4	-8931.64	-8796.51	17893.28	17623.02	18022.18	17751.91
	5	-8931.58	-8796.51	17901.17	17631.02	18064.44	17794.29
	6	-8931.63	-8796.53	17909.26	17639.07	18106.91	17836.71
CHM1 Chr21	1	-8884.14	-7705.50	17774.29	15417.01	17799.96	15442.68
	2	-8111.63	-7680.10	16237.26	<b>15374.20</b>	<b>16297.18</b>	<b>15434.12</b>
	3	-8101.03	-7678.67	16224.06	15379.34	16318.21	15473.49
	4	-8096.28	-7678.67	<b>16222.57</b>	15387.34	16350.96	15515.73
	5	-8096.14	-7678.67	16230.28	15395.34	16392.91	15557.96
	6	-8096.45	-7678.67	16238.90	15403.34	16435.77	15600.20

Table S3: **Maximum likelihood results for different Dirichlet-Multinomial mixture models on heterozygous data.**

TH: True heterozygote dataset; PH: Potential heterozygote dataset;  $k$ : Number of components in the mixture; Bolded: Best model by AIC or BIC.

	$k$	TH lnL	PH lnL	TH AIC	PH AIC	TH BIC	PH BIC
CEU13 Chr21	1	-680227.22	-1049127.29	1360460.44	2098260.57	1360485.35	2098286.28
	2	-679553.35	-1047190.51	1359120.71	2094395.02	1359178.84	2094455.01
	3	-679518.05	-1046976.22	1359058.10	2093974.43	<b>1359149.44</b>	2094068.71
	4	-679503.99	-1046863.10	<b>1359037.99</b>	2093756.19	1359162.55	2093884.75
	5	-679500.25	-1046801.13	1359038.49	2093640.26	1359196.27	2093803.09
	6	-679499.16	-1046759.39	1359044.32	<b>2093564.77</b>	1359235.31	<b>2093761.89</b>
CEU13 Chr10	1	-822125.53	-861877.92	1644257.06	1723761.83	1644282.55	1723787.47
	2	-821819.67	-861134.63	1643653.34	1722283.25	<b>1643712.83</b>	1722343.07
	3	-821806.53	-861095.35	<b>1643635.06</b>	<b>1722212.71</b>	1643728.54	<b>1722306.71</b>
	4	-821803.92	-861092.33	1643637.84	1722214.65	1643765.32	1722342.83
	5	-821802.52	-861088.23	1643643.03	1722214.47	1643804.51	1722376.83
	6	-821801.14	-861086.03	1643648.29	1722218.06	1643843.76	1722414.61
CEU12 Chr10	1	-1576846.58	-1681522.38	3153699.16	3363050.76	3153724.69	3363076.53
	2	-1576531.60	-1679609.04	3153077.20	3359232.08	<b>3153136.77</b>	3359292.21
	3	-1576517.22	-1679519.20	<b>3153056.44</b>	3359060.39	3153150.04	3359154.88
	4	-1576513.59	-1679490.33	3153057.18	3359010.66	3153184.83	<b>3359139.50</b>
	5	-1576510.84	-1679484.47	3153059.69	3359006.94	3153221.37	3359170.15
	6	-1576508.61	-1679473.47	3153063.22	<b>3358992.95</b>	3153258.94	3359190.51
CEU12 Chr21	1	-1295587.45	-1567598.52	2591180.90	3135203.04	2591205.81	3135228.44
	2	-1294765.87	-1564084.48	2589545.74	3128182.96	<b>2589603.87</b>	3128242.23
	3	-1294751.05	-1563981.27	2589524.11	3127984.54	2589615.44	3128077.69
	4	-1294738.61	-1563903.83	<b>2589507.23</b>	3127837.65	2589631.78	3127964.67
	5	-1294735.56	-1563870.23	2589509.11	3127778.45	2589666.88	<b>3127939.35</b>
	6	-1294732.85	-1563852.35	2589511.69	<b>3127750.70</b>	2589702.67	3127945.47
CEU11 Chr10	1	-1576118.74	-1680591.96	3152243.47	3361189.92	3152269.00	3361215.69
	2	-1575808.42	-1678712.33	3151630.84	3357438.67	<b>3151690.41</b>	3357498.80
	3	-1575793.38	-1678620.74	<b>3151608.77</b>	3357263.48	3151702.38	3357357.97
	4	-1575789.85	-1678591.80	3151609.70	3357213.59	3151737.34	<b>3357342.44</b>
	5	-1575786.15	-1678585.13	3151610.30	3357208.25	3151771.99	3357371.46
	6	-1575785.32	-1678576.67	3151616.63	<b>3357199.34</b>	3151812.36	3357396.90
CEU11 Chr21	1	-1295073.11	-1564321.59	2590152.22	3128649.18	2590177.13	3128674.58
	2	-1294248.59	-1560880.98	2588511.18	3121775.95	<b>2588569.31</b>	3121835.22
	3	-1294233.74	-1560781.60	2588489.49	3121585.20	2588580.83	3121678.34
	4	-1294220.12	-1560704.65	<b>2588470.24</b>	3121439.29	2588594.79	3121566.29
	5	-1294216.29	-1560669.63	2588470.58	3121377.27	2588628.35	<b>3121538.13</b>
	6	-1294214.05	-1560651.10	2588474.10	<b>3121348.19</b>	2588665.08	3121542.93
CEU10 Chr10	1	-786319.70	-838669.52	1572645.39	1677345.03	1572670.77	1677370.56
	2	-786283.09	-838295.21	1572580.18	1676604.41	<b>1572639.38</b>	1676663.98
	3	-786279.14	-838241.95	1572580.28	1676505.89	1572673.32	<b>1676599.49</b>
	4	-786272.06	-838223.33	<b>1572574.12</b>	1676476.66	1572700.98	1676604.30
	5	-786271.18	-838218.88	1572580.36	<b>1676475.77</b>	1572741.05	1676637.44
	6	-786271.55	-838215.94	1572589.10	1676477.87	1572783.63	1676673.58
CEU10 Chr21	1	-609177.81	-741752.33	1218361.61	1483510.67	1218386.32	1483535.69
	2	-609069.60	-740874.49	1218153.20	1481762.98	<b>1218210.86</b>	1481821.36
	3	-609057.91	-740685.21	<b>1218137.82</b>	1481392.42	1218228.43	1481484.15
	4	-609057.53	-740660.48	1218145.05	1481350.95	1218268.61	<b>1481476.04</b>
	5	-609057.09	-740641.97	1218152.19	1481321.95	1218308.69	1481480.39
	6	-609056.88	-740630.74	1218159.75	<b>1481307.48</b>	1218349.21	1481499.28

Table S4: **Maximum likelihood parameter estimates for several homozygous models on CHM1 Chr10.**

FD: Full homozygous dataset; RD: Reference homozygous dataset;  $k$ : Number of components in the mixture; M: Multinomial model;  $\pi_{ref}$ : frequency of reference base calls;  $\pi_{err}$ : frequency of error base calls;  $\alpha_{ref}$ : concentration parameter for reference base calls;  $\alpha_{err}$ : concentration parameter for error base calls;  $\varphi$ : dispersion parameter;  $\rho$ : frequency of component in mixture model; ML- $\rho$ : frequency of component when using maximum-likelihood assignment

$k$	Data	$\pi_{ref}$	$\pi_{err}$	$\alpha_{ref}$	$\alpha_{err}$	$\varphi$	$\rho$	ML- $\rho$
M	RD	1.00	0.000286	Inf	Inf	0.00	1.00	1.00
1	FD	0.999	0.00122	6.23	0.00761	0.138	1.00	1.00
1	RD	1.00	0.000289	190.	0.0551	0.00522	1.00	1.00
2	FD	1.00	0.000282	240.	0.0676	0.00415	0.984	0.999
		0.975	0.0254	0.0534	0.00139	0.948	0.0164	0.000978
2	RD	1.00	0.000260	370.	0.0961	0.00269	0.942	0.997
		0.999	0.000833	20.0	0.0167	0.0475	0.0585	0.00324
3	FD	1.00	0.000253	372.	0.0940	0.00268	0.964	0.979
		0.999	0.00134	21.3	0.0287	0.0447	0.0359	0.0205
		0.0836	0.916	0.0129	0.141	0.867	0.000379	0.000502
3	RD	1.00	0.000246	415.	0.102	0.00240	0.998	0.979
		0.976	0.0244	395.	9.87	0.00246	0.00147	0.0206
		0.859	0.141	Inf	Inf	0.00	9.34e-05	0.000201
4	FD	1.00	0.000464	0.0115	5.35e-06	0.989	0.745	0.979
		0.999	0.000964	656.	0.633	0.00152	0.253	0.00
		0.974	0.0259	1.93e+03	51.4	0.000504	0.00139	0.0206
		0.862	0.138	Inf	Inf	0.00	9.50e-05	0.000201
4	RD	1.00	0.000228	465.	0.106	0.00215	0.920	0.979
		0.999	0.000623	127.	0.0794	0.00779	0.0790	0.00
		0.968	0.0318	Inf	Inf	0.00	0.000684	0.0206
		0.859	0.141	Inf	Inf	0.00	9.35e-05	0.000176
5	FD	1.00	0.000265	382.	0.101	0.00261	0.950	0.00364
		0.993	0.00714	0.0115	8.29e-05	0.989	0.0485	0.976
		0.970	0.0301	Inf	Inf	0.00	0.000943	0.00750
		0.860	0.140	Inf	Inf	0.00	9.01e-05	0.000176
		0.996	0.00395	569.	2.26	0.00175	4.06e-07	0.0131
5	RD	1.00	0.000249	357.	0.0889	0.00279	0.980	0.979
		1.00	0.000487	Inf	Inf	0.00	0.0187	0.00
		0.969	0.0306	Inf	Inf	0.00	0.000904	0.00316
		0.860	0.140	Inf	Inf	0.00	9.42e-05	0.000176
		0.987	0.0129	205.	2.69	0.00478	4.12e-06	0.0174
6	FD	1.00	0.000194	74.2	0.0144	0.0133	0.502	0.979
		1.00	0.000368	713.	0.263	0.00140	0.498	0.00
		0.113	0.887	0.0122	0.0955	0.903	0.000389	0.000351
		0.995	0.00506	778.	3.96	0.00128	0.000159	0.0203
		0.860	0.140	Inf	Inf	0.00	8.85e-05	0.000426
		0.530	0.470	0.0149	0.0132	0.973	2.64e-06	0.00
6	RD	1.00	0.000250	389.	0.0972	0.00256	0.998	0.979
		0.996	0.00379	94.1	0.358	0.0105	0.000895	0.00113
		0.969	0.0307	2.57e+04	814.	3.77e-05	0.000729	0.000953
		0.972	0.0276	2.17e+03	61.4	0.000449	0.000204	0.0185
		0.855	0.145	Inf	Inf	0.00	9.05e-05	0.000176
		1.00	0.000316	2.05e+03	0.646	0.000489	4.29e-05	0.00



Table S5: **Maximum likelihood parameter estimates for several homozygous models on CHM1 Chr21.**

$k$	Data	$\pi_{ref}$	$\pi_{err}$	$\alpha_{ref}$	$\alpha_{err}$	$\varphi$	$\rho$	ML- $\rho$
M	RD	1.00	0.000255	Inf	Inf	0.00	1.00	1.00
1	FD	0.999	0.00129	5.18	0.00667	0.162	1.00	1.00
1	RD	1.00	0.000256	273.	0.0697	0.00365	1.00	1.00
2	FD	1.00	0.000249	396.	0.0987	0.00252	0.972	0.995
		0.982	0.0176	0.119	0.00214	0.892	0.0278	0.00485
2	RD	1.00	0.000220	Inf	Inf	0.00	0.751	0.808
		1.00	0.000361	64.5	0.0233	0.0153	0.249	0.192
3	FD	0.999	0.000615	0.100	6.16e-05	0.909	0.745	0.981
		0.999	0.000804	Inf	Inf	0.00	0.253	0.00
		0.981	0.0192	Inf	Inf	0.00	0.00224	0.0191
3	RD	1.00	0.000185	Inf	Inf	0.00	0.995	0.981
		0.986	0.0141	Inf	Inf	0.00	0.00446	0.0187
		0.926	0.0742	Inf	Inf	0.00	0.000101	0.000441
4	FD	1.00	0.000185	2.25e+04	4.16	4.44e-05	0.994	0.980
		0.987	0.0130	512.	6.77	0.00192	0.00497	0.0191
		0.00453	0.995	0.274	60.3	0.0162	0.000363	0.000363
		0.766	0.234	6.39	1.96	0.107	0.000234	0.000259
4	RD	1.00	0.000185	Inf	Inf	0.00	0.995	0.981
		0.986	0.0141	Inf	Inf	0.00	0.00446	0.0187
		0.926	0.0742	Inf	Inf	0.00	0.000101	0.000441
		1.00	0.000405	384.	0.156	0.00259	1.00e-06	0.00
5	FD	1.00	0.000188	1.39e+04	2.62	7.17e-05	0.995	0.980
		0.985	0.0146	Inf	Inf	0.00	0.00401	0.0191
		0.984	0.0156	3.81	0.0604	0.205	0.000842	0.00
		0.00453	0.995	0.274	60.3	0.0162	0.000363	0.000363
		0.754	0.246	6.13	2.00	0.110	0.000181	0.000234
5	RD	1.00	0.000185	Inf	Inf	0.00	0.995	0.981
		0.986	0.0141	Inf	Inf	0.00	0.00446	0.0187
		1.00	0.000411	1.31e+03	0.540	0.000761	0.000130	0.00
		0.926	0.0742	Inf	Inf	0.00	0.000101	0.000441
		0.999	0.000912	447.	0.408	0.00223	4.17e-09	0.00
6	FD	1.00	0.000188	1.30e+04	2.45	7.68e-05	0.995	0.980
		0.985	0.0146	Inf	Inf	0.00	0.00320	0.0161
		0.985	0.0148	Inf	Inf	0.00	0.000800	0.00285
		0.00453	0.995	0.274	60.3	0.0162	0.000363	0.000363
		0.832	0.168	3.91	0.791	0.176	0.000337	0.000389
		0.999	0.000754	172.	0.130	0.00576	0.000117	0.00
6	RD	1.00	0.000185	Inf	Inf	0.00	0.995	0.00
		0.986	0.0141	Inf	Inf	0.00	0.00444	0.0163
		1.00	2.58e-05	Inf	Inf	0.00	0.000851	0.981
		0.926	0.0742	Inf	Inf	0.00	0.000101	0.000389
		0.985	0.0154	Inf	Inf	0.00	1.73e-05	0.00252
		1.00	0.000423	Inf	Inf	0.00	8.07e-06	0.00

Table S6: **Maximum likelihood parameter estimates for several heterozygous models on CEU13 Chr10.**

TH: True heterozygous dataset; PH: Potential heterozygous dataset;  $k$ : Number of components in the mixture; M: Multinomial model; BM: Biased multinomial model;  $\pi_{ref}$ : frequency of reference base calls;  $\pi_{alt}$ : frequency of alternative base calls;  $\pi_{err}$ : frequency of error base calls;  $\alpha_{ref}$ : concentration parameter for reference base calls;  $\alpha_{alt}$ : concentration parameter for alternative base calls;  $\alpha_{err}$ : concentration parameter for error base calls;  $\varphi$ : dispersion parameter;  $\rho$ : frequency of component in mixture model; ML- $\rho$ : frequency of component when using maximum-likelihood assignment

$k$	Data	$\pi_{ref}$	$\pi_{alt}$	$\pi_{err}$	$\alpha_{ref}$	$\alpha_{alt}$	$\alpha_{err}$	$\varphi$	$\rho$	ML- $\rho$
M	TH	0.4998	0.4998	0.0004489	Inf	Inf	Inf	0.000	1.000	1.000
BM	TH	0.5021	0.4974	0.0004489	Inf	Inf	Inf	0.000	1.000	1.000
1	TH	0.5022	0.4974	0.0003694	149.4	148.0	0.1099	0.003350	1.000	1.000
1	PH	0.5031	0.4964	0.0004475	73.28	72.30	0.06517	0.006819	1.000	1.000
2	TH	0.5022	0.4975	0.0003361	1174.	1163.	0.7857	0.0004276	0.9925	0.8482
		0.5041	0.4897	0.006212	3.551	3.450	0.04377	0.1243	0.007537	0.1518
2	PH	0.5029	0.4968	0.0003283	767.8	758.6	0.5013	0.0006545	0.9761	0.8449
		0.5112	0.4797	0.009143	3.886	3.647	0.06951	0.1162	0.02391	0.1551
3	TH	0.5021	0.4976	0.0003426	4868.	4825.	3.321	0.0001031	0.9808	0.7949
		0.5130	0.4865	0.0004879	7.228	6.855	0.006875	0.06627	0.01897	0.1983
		0.3012	0.4273	0.2715	3.192	4.528	2.877	0.08622	0.0002831	0.006757
3	PH	0.5023	0.4974	0.0003417	Inf	Inf	Inf	0.000	0.9496	0.7835
		0.5229	0.4757	0.001465	7.662	6.970	0.02147	0.06389	0.04922	0.2055
		0.2993	0.4848	0.2159	3.121	5.056	2.252	0.08749	0.001208	0.01100
4	TH	0.5021	0.4975	0.0003420	1139.	1128.	0.7755	0.0004408	0.9937	0.7739
		0.4078	0.5910	0.001191	5.389	7.810	0.01574	0.07035	0.003917	0.1360
		0.7484	0.2516	1.349e-13	5.610e+07	1.886e+07	1.011e-05	1.334e-08	0.002079	0.08268
		0.3080	0.4266	0.2654	3.202	4.435	2.758	0.08776	0.0002939	0.007418
4	PH	0.5023	0.4974	0.0003456	Inf	Inf	Inf	0.000	0.9511	0.7415
		0.5290	0.4700	0.0009964	7.912	7.029	0.01490	0.06267	0.04643	0.1320
		0.3191	0.4786	0.2023	3.101	4.652	1.967	0.09328	0.001372	0.009973
		0.2978	0.6981	0.004057	Inf	Inf	Inf	0.000	0.001127	0.1165
5	TH	0.5021	0.4976	0.0003412	1359.	1347.	0.9237	0.0003692	0.9937	0.7490
		0.7279	0.2721	1.968e-13	1350.	504.5	3.650e-10	0.0005390	0.002797	0.1022
		0.3923	0.6077	9.512e-12	4.240	6.569	1.028e-10	0.08469	0.002008	0.0006894
		0.3172	0.6811	0.001642	Inf	Inf	Inf	0.000	0.001216	0.1413
		0.3193	0.4333	0.2473	3.219	4.368	2.493	0.09026	0.0003254	0.006867
5	PH	0.5015	0.4982	0.0003431	6676.	6633.	4.567	7.511e-05	0.9634	0.6692
		0.6009	0.3983	0.0008334	16.63	11.02	0.02307	0.03487	0.02926	0.1958
		0.3000	0.6985	0.001459	Inf	Inf	Inf	0.000	0.004281	0.1191
		0.3362	0.4773	0.1864	3.153	4.476	1.748	0.09637	0.001591	0.01197
		0.3572	0.6428	1.207e-11	3.672	6.609	1.241e-10	0.08864	0.001474	0.003921
6	TH	0.5019	0.4977	0.0003358	7033.	6974.	4.705	7.137e-05	0.9905	0.6602
		0.6759	0.3241	2.719e-13	55.93	26.82	2.250e-11	0.01194	0.005647	0.1619
		0.3330	0.6660	0.0009848	Inf	Inf	Inf	0.000	0.002714	0.1616
		0.2642	0.7358	6.057e-13	5.362	14.94	1.229e-11	0.04695	0.0006687	0.005129
		0.2956	0.4218	0.2826	3.261	4.654	3.117	0.08311	0.0002766	0.0003034
		0.4908	0.4708	0.03843	Inf	Inf	Inf	0.000	0.0002350	0.01081
6	PH	0.5015	0.4981	0.0003495	2.703e+04	2.685e+04	18.84	1.855e-05	0.9595	0.6548
		0.6041	0.3959	1.659e-10	18.70	12.26	5.137e-09	0.03129	0.02522	0.2078
		0.4696	0.5304	3.594e-08	5.491	6.203	4.203e-07	0.07878	0.01051	0.007184
		0.3007	0.6968	0.002499	Inf	Inf	Inf	0.000	0.002618	0.1185
		0.5582	0.3668	0.07494	11.30	7.427	1.517	0.04707	0.001127	0.01060
		0.2544	0.5173	0.2283	3.403	6.921	3.055	0.06955	0.0009985	0.001105

Table S7: Maximum likelihood parameter estimates for several heterozygous models on CEU13 Chr21.

$k$	Data	$\pi_{ref}$	$\pi_{alt}$	$\pi_{err}$	$\alpha_{ref}$	$\alpha_{alt}$	$\alpha_{err}$	$\varphi$	$\rho$	ML- $\rho$
M	TH	0.4996	0.4996	0.0007413	Inf	Inf	Inf	0.000	1.000	1.000
BM	TH	0.5035	0.4958	0.0007413	Inf	Inf	Inf	0.000	1.000	1.000
1	TH	0.5038	0.4958	0.0004158	57.43	56.52	0.04740	0.008696	1.000	1.000
1	PH	0.5279	0.4711	0.0009801	10.61	9.472	0.01970	0.04739	1.000	1.000
2	TH	0.5038	0.4958	0.0003342	682.3	671.4	0.4525	0.0007379	0.9603	0.8147
		0.5014	0.4959	0.002686	4.680	4.629	0.02507	0.09677	0.03968	0.1853
2	PH	0.4992	0.5005	0.0003619	Inf	Inf	Inf	0.000	0.5780	0.7012
		0.5608	0.4373	0.001863	5.983	4.666	0.01987	0.08569	0.4220	0.2988
3	TH	0.5036	0.4960	0.0003526	2049.	2018.	1.435	0.0002457	0.9390	0.7829
		0.5080	0.4914	0.0005265	6.862	6.638	0.007111	0.06893	0.06044	0.2130
		0.2391	0.4825	0.2784	3.407	6.876	3.967	0.06558	0.0005865	0.004120
3	PH	0.5047	0.4949	0.0003515	122.6	120.2	0.08539	0.004100	0.7425	0.7017
		0.4928	0.5034	0.003834	4.602	4.701	0.03580	0.09672	0.1627	0.1239
		0.7471	0.2524	0.0005183	108.2	36.56	0.07506	0.006858	0.09479	0.1744
4	TH	0.5032	0.4965	0.0003518	382.5	377.4	0.2674	0.001314	0.9857	0.8037
		0.7252	0.2748	1.877e-13	193.4	73.28	5.007e-11	0.003736	0.008887	0.1176
		0.2426	0.7567	0.0006698	66.99	209.0	0.1850	0.003608	0.004737	0.07333
		0.2543	0.4884	0.2572	3.393	6.517	3.432	0.06973	0.0006832	0.005393
4	PH	0.5036	0.4960	0.0004012	Inf	Inf	Inf	0.000	0.5961	0.6771
		0.5244	0.4744	0.001256	6.993	6.325	0.01674	0.06976	0.3464	0.1854
		0.7743	0.2252	0.0005180	Inf	Inf	Inf	0.000	0.05511	0.1269
		0.3176	0.4197	0.2627	3.514	4.644	2.907	0.08288	0.002399	0.01057
5	TH	0.5036	0.4961	0.0003458	386.5	380.7	0.2654	0.001301	0.9855	0.7382
		0.7407	0.2593	7.194e-14	2236.	783.1	2.172e-10	0.0003311	0.007506	0.09755
		0.2262	0.7738	7.870e-14	136.5	467.1	4.750e-11	0.001654	0.003360	0.01142
		0.3279	0.6690	0.003110	Inf	Inf	Inf	0.000	0.002962	0.1503
		0.2541	0.4880	0.2579	3.403	6.536	3.453	0.06948	0.0006817	0.002479
5	PH	0.4988	0.5009	0.0003695	360.7	362.3	0.2673	0.001381	0.7264	0.5190
		0.6432	0.3561	0.0007526	74.41	41.20	0.08706	0.008570	0.1300	0.2247
		0.3627	0.6364	0.0009410	9.229	16.20	0.02395	0.03781	0.07374	0.1588
		0.7812	0.2185	0.0003504	Inf	Inf	Inf	0.000	0.06494	0.08141
		0.4161	0.4103	0.1736	3.510	3.462	1.465	0.1060	0.004912	0.01607
6	TH	0.5078	0.4919	0.0003247	813.0	787.6	0.5198	0.0006242	0.9052	0.4932
		0.4533	0.5462	0.0005582	Inf	Inf	Inf	0.000	0.08075	0.3049
		0.7323	0.2677	2.838e-13	341.8	124.9	1.324e-10	0.002138	0.008270	0.1057
		0.2219	0.7781	8.791e-14	359.2	1260.	1.423e-10	0.0006174	0.003176	0.01002
		0.3160	0.6805	0.003556	Inf	Inf	Inf	0.000	0.001938	0.08318
		0.2584	0.4886	0.2530	3.378	6.389	3.308	0.07105	0.0007006	0.002914
6	PH	0.5010	0.4987	0.0003355	5937.	5910.	3.976	8.438e-05	0.7094	0.4596
		0.6385	0.3608	0.0007513	74.62	42.17	0.08781	0.008484	0.1378	0.2235
		0.3720	0.6274	0.0006599	169.3	285.6	0.3004	0.002192	0.06812	0.1939
		0.7806	0.2191	0.0003489	Inf	Inf	Inf	0.000	0.06624	0.08251
		0.2149	0.7848	0.0002883	97.88	357.5	0.1313	0.002191	0.01289	0.02454
		0.4194	0.4209	0.1598	3.492	3.504	1.330	0.1072	0.005488	0.01596

Table S8: Maximum likelihood parameter estimates for several heterozygous models on CEU12 Chr10.

$k$	Data	$\pi_{ref}$	$\pi_{alt}$	$\pi_{err}$	$\alpha_{ref}$	$\alpha_{alt}$	$\alpha_{err}$	$\varphi$	$\rho$	ML- $\rho$
M	TH	0.4998	0.4998	0.0003800	Inf	Inf	Inf	0.000	1.000	1.000
BM	TH	0.5087	0.4909	0.0003800	Inf	Inf	Inf	0.000	1.000	1.000
1	TH	0.5087	0.4909	0.0003461	237.0	228.7	0.1612	0.002142	1.000	1.000
1	PH	0.5141	0.4854	0.0004806	63.86	60.30	0.05970	0.007986	1.000	1.000
2	TH	0.5083	0.4914	0.0003188	918.9	888.4	0.5763	0.0005528	0.9862	0.8508
		0.5396	0.4574	0.003047	6.558	5.559	0.03704	0.07602	0.01379	0.1492
2	PH	0.5090	0.4907	0.0003057	2520.	2429.	1.513	0.0002020	0.9182	0.8220
		0.5719	0.4245	0.003634	7.547	5.603	0.04796	0.07043	0.08181	0.1780
3	TH	0.5078	0.4918	0.0003199	845.6	818.9	0.5326	0.0006002	0.9915	0.8215
		0.6690	0.3304	0.0006527	88.31	43.61	0.08616	0.007518	0.006350	0.1162
		0.4452	0.5343	0.02058	2.744	3.294	0.1269	0.1396	0.002156	0.06234
3	PH	0.5081	0.4916	0.0003190	Inf	Inf	Inf	0.000	0.9017	0.8045
		0.5728	0.4256	0.001665	10.38	7.716	0.03019	0.05227	0.09660	0.1884
		0.3862	0.3923	0.2215	4.669	4.742	2.678	0.07640	0.001678	0.007125
4	TH	0.5079	0.4918	0.0003211	891.4	863.2	0.5636	0.0005694	0.9906	0.7604
		0.6662	0.3330	0.0008793	69.75	34.86	0.09206	0.009461	0.007008	0.1204
		0.3893	0.6107	1.633e-11	5.772	9.053	2.421e-10	0.06319	0.001901	0.1030
		0.3960	0.4725	0.1315	2.811	3.355	0.9335	0.1235	0.0004475	0.01620
4	PH	0.5085	0.4912	0.0003293	609.2	588.5	0.3945	0.0008340	0.9574	0.8001
		0.6858	0.3127	0.001444	44.35	20.22	0.09336	0.01523	0.03712	0.1297
		0.2865	0.7114	0.002068	17.02	42.27	0.1229	0.01655	0.003173	0.06281
		0.4244	0.3879	0.1877	5.248	4.797	2.320	0.07482	0.002325	0.007351
5	TH	0.5087	0.4910	0.0003365	1559.	1505.	1.032	0.0003261	0.9400	0.6069
		0.4921	0.5079	2.566e-12	68.05	70.24	3.549e-10	0.007179	0.05255	0.2596
		0.6681	0.3309	0.0009906	68.83	34.09	0.1021	0.009613	0.006758	0.1184
		0.1830	0.8124	0.004587	40.72	180.8	1.021	0.004474	0.0003607	0.006791
		0.4126	0.4176	0.1698	4.200	4.252	1.729	0.08944	0.0003365	0.008318
5	PH	0.5091	0.4905	0.0003334	541.1	521.3	0.3543	0.0009401	0.9621	0.7844
		0.7066	0.2925	0.0008654	84.52	34.99	0.1035	0.008291	0.03006	0.1041
		0.3418	0.6559	0.002284	8.945	17.17	0.05979	0.03680	0.004931	0.09977
		0.3909	0.3910	0.2181	5.315	5.316	2.965	0.06851	0.001827	0.002392
		0.6357	0.3101	0.05419	116.5	56.86	9.935	0.005425	0.001124	0.009365
6	TH	0.5080	0.4917	0.0003223	951.4	921.0	0.6037	0.0005336	0.9882	0.7899
		0.4888	0.5112	1.144e-12	6.890	7.206	1.613e-11	0.06624	0.005814	0.09112
		0.6712	0.3277	0.001044	91.30	44.58	0.1420	0.007298	0.005624	0.1148
		0.3776	0.3389	0.2835	9.272	8.323	6.963	0.03913	0.0001271	0.0001636
		0.5800	0.3208	0.09922	Inf	Inf	Inf	0.000	0.0001266	0.003055
		0.1469	0.7648	0.08838	Inf	Inf	Inf	0.000	0.0001105	0.0008727
6	PH	0.5075	0.4921	0.0003551	1391.	1349.	0.9731	0.0003647	0.8904	0.7078
		0.5468	0.4532	3.921e-12	29.56	24.50	2.120e-10	0.01816	0.08800	0.1954
		0.7474	0.2518	0.0007493	Inf	Inf	Inf	0.000	0.01564	0.04625
		0.2536	0.7436	0.002730	25.62	75.13	0.2758	0.009801	0.002149	0.03658
		0.3995	0.3916	0.2090	5.318	5.213	2.782	0.06986	0.001971	0.002820
		0.6428	0.3201	0.03712	137.9	68.66	7.961	0.004641	0.001824	0.01120

Table S9: Maximum likelihood parameter estimates for several heterozygous models on CEU12 Chr21.

$k$	Data	$\pi_{ref}$	$\pi_{alt}$	$\pi_{err}$	$\alpha_{ref}$	$\alpha_{alt}$	$\alpha_{err}$	$\varphi$	$\rho$	ML- $\rho$
M	TH	0.4998	0.4998	0.0004307	Inf	Inf	Inf	0.000	1.000	1.000
BM	TH	0.5100	0.4895	0.0004307	Inf	Inf	Inf	0.000	1.000	1.000
1	TH	0.5101	0.4895	0.0003713	124.5	119.4	0.09058	0.004082	1.000	1.000
1	PH	0.5227	0.4765	0.0007124	21.89	19.95	0.02983	0.02332	1.000	1.000
2	TH	0.5089	0.4908	0.0003148	3945.	3805.	2.441	0.0001290	0.9610	0.8400
		0.5414	0.4566	0.002043	6.465	5.452	0.02440	0.07727	0.03896	0.1600
2	PH	0.5086	0.4911	0.0003195	Inf	Inf	Inf	0.000	0.7717	0.7700
		0.5657	0.4320	0.002319	6.665	5.089	0.02733	0.07823	0.2283	0.2300
3	TH	0.5087	0.4909	0.0003219	4138.	3993.	2.619	0.0001229	0.9579	0.8340
		0.5439	0.4551	0.001005	7.315	6.121	0.01352	0.06921	0.04168	0.1634
		0.2944	0.4984	0.2072	9.508	16.10	6.690	0.03003	0.0003916	0.002581
3	PH	0.5088	0.4909	0.0003109	4069.	3925.	2.487	0.0001250	0.8314	0.7583
		0.5065	0.4896	0.003937	4.904	4.740	0.03811	0.09362	0.09951	0.08736
		0.7025	0.2964	0.001033	45.61	19.24	0.06707	0.01517	0.06907	0.1543
4	TH	0.5085	0.4912	0.0003246	915.7	884.6	0.5845	0.0005550	0.9832	0.8537
		0.6889	0.3106	0.0005154	38.32	17.28	0.02867	0.01766	0.01348	0.1045
		0.2569	0.7431	2.615e-14	60.48	175.0	6.157e-12	0.004230	0.002605	0.03412
		0.3490	0.4979	0.1531	6.533	9.320	2.866	0.05071	0.0006668	0.007676
4	PH	0.5091	0.4905	0.0003243	1.323e+05	1.275e+05	84.29	3.847e-06	0.8261	0.7245
		0.6979	0.3008	0.001225	37.28	16.07	0.06546	0.01838	0.08869	0.1578
		0.4630	0.5354	0.001599	6.967	8.057	0.02407	0.06231	0.08300	0.1099
		0.4091	0.3630	0.2279	3.882	3.444	2.162	0.09535	0.002219	0.007761
5	TH	0.5063	0.4934	0.0002986	Inf	Inf	Inf	0.000	0.8876	0.6159
		0.5347	0.4647	0.0006122	104.1	90.46	0.1192	0.005110	0.1000	0.2778
		0.7276	0.2722	0.0001842	88.49	33.11	0.02240	0.008155	0.008405	0.04642
		0.2826	0.7174	3.058e-13	21.66	54.99	2.344e-11	0.01288	0.003326	0.05430
		0.3345	0.4998	0.1657	6.946	10.38	3.441	0.04595	0.0006072	0.005531
5	PH	0.5075	0.4922	0.0003128	Inf	Inf	Inf	0.000	0.8300	0.6505
		0.6431	0.3557	0.001168	40.31	22.30	0.07320	0.01570	0.09971	0.1686
		0.3808	0.6179	0.001275	9.917	16.09	0.03319	0.03698	0.04325	0.1250
		0.7783	0.2213	0.0004573	Inf	Inf	Inf	0.000	0.02400	0.04580
		0.4393	0.3751	0.1856	3.878	3.312	1.638	0.1018	0.003021	0.01015
6	TH	0.5064	0.4933	0.0003001	Inf	Inf	Inf	0.000	0.9329	0.6286
		0.5748	0.4247	0.0005298	Inf	Inf	Inf	0.000	0.03201	0.2906
		0.5367	0.4620	0.001361	30.46	26.22	0.07723	0.01732	0.02589	0.02330
		0.7486	0.2514	6.357e-12	239.3	80.36	2.032e-09	0.003119	0.006196	0.02450
		0.2540	0.7460	1.612e-14	80.61	236.8	5.117e-12	0.003141	0.002496	0.02960
		0.3235	0.5008	0.1757	7.404	11.46	4.021	0.04186	0.0005525	0.003352
6	PH	0.5071	0.4926	0.0003104	Inf	Inf	Inf	0.000	0.8476	0.6526
		0.6440	0.3550	0.0009901	59.22	32.65	0.09105	0.01076	0.09219	0.1690
		0.7764	0.2231	0.0004401	Inf	Inf	Inf	0.000	0.02775	0.04552
		0.3482	0.6512	0.0006171	Inf	Inf	Inf	0.000	0.02232	0.1050
		0.2199	0.7800	0.0001242	288.9	1025.	0.1631	0.0007606	0.005651	0.01012
		0.4610	0.4047	0.1343	3.629	3.186	1.058	0.1127	0.004472	0.01765

Table S10: **Maximum likelihood parameter estimates for several heterozygous models on CEU11 Chr10.**

$k$	Data	$\pi_{ref}$	$\pi_{alt}$	$\pi_{err}$	$\alpha_{ref}$	$\alpha_{alt}$	$\alpha_{err}$	$\varphi$	$\rho$	ML- $\rho$
M	TH	0.4998	0.4998	0.0003806	Inf	Inf	Inf	0.000	1.000	1.000
BM	TH	0.5087	0.4909	0.0003806	Inf	Inf	Inf	0.000	1.000	1.000
1	TH	0.5087	0.4909	0.0003467	237.1	228.8	0.1616	0.002141	1.000	1.000
1	PH	0.5140	0.4855	0.0004811	64.41	60.83	0.06028	0.007917	1.000	1.000
2	TH	0.5082	0.4914	0.0003192	913.1	882.9	0.5735	0.0005563	0.9861	0.8484
		0.5418	0.4552	0.003015	6.806	5.718	0.03787	0.07373	0.01390	0.1516
2	PH	0.5090	0.4907	0.0003069	2376.	2290.	1.432	0.0002142	0.9185	0.8213
		0.5717	0.4247	0.003620	7.638	5.674	0.04837	0.06964	0.08153	0.1787
3	TH	0.5078	0.4919	0.0003205	836.8	810.5	0.5281	0.0006065	0.9916	0.8235
		0.6694	0.3299	0.0006524	95.27	46.95	0.09285	0.006978	0.006423	0.1164
		0.4460	0.5307	0.02328	2.716	3.232	0.1418	0.1411	0.001937	0.06010
3	PH	0.5080	0.4916	0.0003201	Inf	Inf	Inf	0.000	0.9013	0.8013
		0.5724	0.4259	0.001645	10.61	7.893	0.03048	0.05120	0.09706	0.1914
		0.3875	0.3908	0.2217	4.649	4.689	2.660	0.07694	0.001670	0.007224
4	TH	0.5078	0.4918	0.0003216	890.2	862.2	0.5638	0.0005701	0.9906	0.7565
		0.6659	0.3333	0.0008860	73.44	36.76	0.09773	0.008985	0.007101	0.1211
		0.3972	0.6028	1.012e-11	6.084	9.233	1.550e-10	0.06129	0.001883	0.1062
		0.3964	0.4717	0.1319	2.819	3.354	0.9377	0.1233	0.0004465	0.01614
4	PH	0.5085	0.4912	0.0003312	595.2	574.9	0.3877	0.0008536	0.9583	0.8022
		0.6870	0.3116	0.001413	48.01	21.77	0.09876	0.01411	0.03632	0.1272
		0.2877	0.7103	0.002073	17.19	42.46	0.1239	0.01645	0.003089	0.06303
		0.4269	0.3869	0.1863	5.220	4.731	2.278	0.07559	0.002330	0.007552
5	TH	0.5079	0.4918	0.0003222	881.7	853.8	0.5593	0.0005757	0.9905	0.7699
		0.6665	0.3325	0.0009937	73.55	36.69	0.1096	0.008981	0.006994	0.1221
		0.4137	0.5863	2.726e-12	5.833	8.267	3.844e-11	0.06623	0.002182	0.1052
		0.4482	0.3445	0.2073	8.755	6.730	4.050	0.04870	0.0002355	0.002318
		0.1457	0.7649	0.08935	Inf	Inf	Inf	0.000	0.0001064	0.0004363
5	PH	0.5091	0.4906	0.0003365	479.5	462.0	0.3169	0.001061	0.9655	0.8305
		0.7097	0.2894	0.0008222	106.1	43.25	0.1229	0.006647	0.02867	0.1001
		0.2792	0.7184	0.002348	18.46	47.48	0.1552	0.01490	0.002841	0.05631
		0.3941	0.3894	0.2165	5.305	5.241	2.914	0.06916	0.001844	0.002542
		0.6348	0.3135	0.05165	97.86	48.33	7.962	0.006445	0.001169	0.01052
6	TH	0.5078	0.4919	0.0003366	1398.	1354.	0.9266	0.0003631	0.9443	0.6906
		0.5134	0.4866	2.236e-06	56.66	53.70	0.0002467	0.008980	0.04954	0.1439
		0.6840	0.3148	0.001235	92.72	42.67	0.1674	0.007323	0.005006	0.08627
		0.3182	0.6818	5.043e-12	6.405	13.72	1.015e-10	0.04733	0.0008296	0.07632
		0.4515	0.3463	0.2022	8.736	6.700	3.913	0.04914	0.0002474	0.002509
		0.1463	0.7645	0.08922	Inf	Inf	Inf	0.000	0.0001074	0.0003272
6	PH	0.5080	0.4917	0.0003474	890.7	862.1	0.6091	0.0005700	0.9219	0.7400
		0.5671	0.4329	2.620e-12	20.45	15.61	9.448e-11	0.02698	0.05887	0.1642
		0.7458	0.2533	0.0009029	Inf	Inf	Inf	0.000	0.01342	0.04161
		0.2569	0.7404	0.002781	23.97	69.08	0.2595	0.01060	0.002113	0.04020
		0.4028	0.3886	0.2086	5.332	5.145	2.761	0.07023	0.001971	0.002895
		0.6434	0.3197	0.03681	152.8	75.93	8.742	0.004193	0.001701	0.01110

Table S11: Maximum likelihood parameter estimates for several heterozygous models on CEU11 Chr21.

$k$	Data	$\pi_{ref}$	$\pi_{alt}$	$\pi_{err}$	$\alpha_{ref}$	$\alpha_{alt}$	$\alpha_{err}$	$\varphi$	$\rho$	ML- $\rho$
M	TH	0.4998	0.4998	0.0004293	Inf	Inf	Inf	0.000	1.000	1.000
BM	TH	0.5100	0.4896	0.0004293	Inf	Inf	Inf	0.000	1.000	1.000
1	TH	0.5101	0.4895	0.0003703	124.1	119.1	0.09006	0.004095	1.000	1.000
1	PH	0.5224	0.4769	0.0007034	22.27	20.33	0.02998	0.02292	1.000	1.000
2	TH	0.5088	0.4908	0.0003149	3879.	3742.	2.400	0.0001312	0.9609	0.8418
		0.5406	0.4574	0.001981	6.374	5.394	0.02336	0.07818	0.03908	0.1582
2	PH	0.5086	0.4911	0.0003191	Inf	Inf	Inf	0.000	0.7727	0.7701
		0.5645	0.4332	0.002278	6.717	5.155	0.02711	0.07752	0.2273	0.2299
3	TH	0.5087	0.4910	0.0003224	3838.	3704.	2.432	0.0001325	0.9580	0.8328
		0.5432	0.4558	0.0009155	7.184	6.028	0.01211	0.07030	0.04157	0.1643
		0.2996	0.4973	0.2031	9.022	14.97	6.117	0.03214	0.0004082	0.002882
3	PH	0.5088	0.4909	0.0003093	1.019e+04	9832.	6.194	4.993e-05	0.8303	0.7578
		0.5068	0.4894	0.003809	4.996	4.824	0.03755	0.09210	0.1015	0.08709
		0.7005	0.2985	0.001015	45.51	19.39	0.06594	0.01516	0.06817	0.1551
4	TH	0.5085	0.4911	0.0003254	815.4	787.4	0.5217	0.0006233	0.9850	0.8723
		0.7020	0.2977	0.0002903	49.82	21.12	0.02060	0.01390	0.01189	0.09219
		0.2447	0.7553	1.372e-14	329.7	1017.	1.848e-11	0.0007418	0.002408	0.02651
		0.3573	0.4937	0.1490	6.297	8.702	2.627	0.05369	0.0006942	0.009014
4	PH	0.5091	0.4906	0.0003222	9.835e+05	9.478e+05	622.5	5.176e-07	0.8297	0.7159
		0.6937	0.3051	0.001199	36.06	15.86	0.06233	0.01888	0.09284	0.1598
		0.4490	0.5495	0.001536	7.374	9.025	0.02522	0.05739	0.07515	0.1163
		0.4164	0.3636	0.2200	3.943	3.443	2.083	0.09553	0.002298	0.008000
5	TH	0.5073	0.4924	0.0003050	2099.	2037.	1.262	0.0002417	0.9539	0.6800
		0.5590	0.4400	0.001056	74.72	58.81	0.1412	0.007426	0.03453	0.2438
		0.7272	0.2728	4.369e-13	85.58	32.10	5.141e-11	0.008426	0.008178	0.03525
		0.2594	0.7406	3.191e-14	48.48	138.4	5.964e-12	0.005323	0.002802	0.03629
		0.3323	0.5008	0.1668	7.093	10.69	3.561	0.04475	0.0006004	0.004625
5	PH	0.5074	0.4923	0.0003110	Inf	Inf	Inf	0.000	0.8311	0.6477
		0.6406	0.3583	0.001125	44.32	24.79	0.07786	0.01425	0.09793	0.1700
		0.3789	0.6198	0.001252	10.26	16.78	0.03389	0.03563	0.04278	0.1251
		0.7761	0.2235	0.0004227	Inf	Inf	Inf	0.000	0.02510	0.04629
		0.4464	0.3747	0.1789	3.921	3.291	1.572	0.1022	0.003104	0.01093
6	TH	0.5051	0.4946	0.0002970	Inf	Inf	Inf	0.000	0.9154	0.5886
		0.5615	0.4381	0.0004165	Inf	Inf	Inf	0.000	0.06798	0.3180
		0.7339	0.2661	1.338e-12	123.9	44.94	2.260e-10	0.005887	0.007956	0.03247
		0.5223	0.4719	0.005832	28.27	25.54	0.3157	0.01814	0.005586	0.03264
		0.2500	0.7500	1.991e-14	87.40	262.2	6.961e-12	0.002852	0.002559	0.02724
		0.3210	0.4976	0.1814	7.584	11.76	4.286	0.04061	0.0005290	0.001072
6	PH	0.5071	0.4926	0.0003088	Inf	Inf	Inf	0.000	0.8500	0.6483
		0.6432	0.3559	0.0009473	70.76	39.15	0.1042	0.009008	0.08860	0.1723
		0.7740	0.2256	0.0004075	Inf	Inf	Inf	0.000	0.02881	0.04595
		0.3480	0.6514	0.0006124	Inf	Inf	Inf	0.000	0.02247	0.1050
		0.2201	0.7798	0.0001335	2486.	8810.	1.508	8.851e-05	0.005572	0.01011
		0.4663	0.4038	0.1299	3.694	3.199	1.029	0.1121	0.004579	0.01831

Table S12: Maximum likelihood parameter estimates for several heterozygous models on CEU10 Chr10.

$k$	Data	$\pi_{ref}$	$\pi_{alt}$	$\pi_{err}$	$\alpha_{ref}$	$\alpha_{alt}$	$\alpha_{err}$	$\varphi$	$\rho$	ML- $\rho$
M	TH	0.4989	0.4989	0.002286	Inf	Inf	Inf	0.000	1.000	1.000
BM	TH	0.5392	0.4585	0.002286	Inf	Inf	Inf	0.000	1.000	1.000
1	TH	0.5393	0.4583	0.002339	153.8	130.7	0.6670	0.003494	1.000	1.000
1	PH	0.5427	0.4547	0.002618	59.24	49.63	0.2858	0.009078	1.000	1.000
2	TH	0.5344	0.4626	0.003046	Inf	Inf	Inf	0.000	0.6843	0.6056
		0.5502	0.4491	0.0006450	42.20	34.45	0.04947	0.01287	0.3157	0.3944
2	PH	0.5369	0.4607	0.002451	Inf	Inf	Inf	0.000	0.7865	0.7211
		0.5630	0.4343	0.002709	15.41	11.88	0.07414	0.03525	0.2135	0.2789
3	TH	0.5292	0.4684	0.002365	459.3	406.5	2.053	0.001151	0.8825	0.5417
		0.6175	0.3809	0.001567	Inf	Inf	Inf	0.000	0.1161	0.3581
		0.4946	0.4907	0.01469	5.697	5.652	0.1693	0.07988	0.001398	0.1002
3	PH	0.5322	0.4655	0.002265	1102.	963.9	4.690	0.0004827	0.8803	0.5988
		0.6597	0.3382	0.002081	201.5	103.3	0.6358	0.003263	0.08773	0.2923
		0.5082	0.4820	0.009872	7.470	7.085	0.1451	0.06370	0.03201	0.1089
4	TH	0.5385	0.4584	0.003147	5764.	4907.	33.69	9.342e-05	0.7036	0.1746
		0.4978	0.5020	0.0001580	Inf	Inf	Inf	0.000	0.1894	0.4267
		0.6214	0.3785	0.0001465	Inf	Inf	Inf	0.000	0.1052	0.3446
		0.5062	0.4765	0.01732	6.740	6.344	0.2306	0.06986	0.001808	0.05410
4	PH	0.5318	0.4659	0.002284	1310.	1148.	5.624	0.0004058	0.8706	0.5051
		0.6471	0.3504	0.002439	93.50	50.63	0.3524	0.006874	0.1122	0.2902
		0.3980	0.5974	0.004538	19.62	29.45	0.2237	0.01988	0.01576	0.1730
		0.4907	0.3654	0.1439	8.623	6.422	2.529	0.05384	0.001389	0.03162
5	TH	0.5375	0.4598	0.002703	931.7	796.8	4.684	0.0005766	0.8379	0.2351
		0.4809	0.5191	9.648e-09	Inf	Inf	Inf	0.000	0.08877	0.3769
		0.6328	0.3672	7.373e-06	Inf	Inf	Inf	0.000	0.07241	0.2959
		0.4405	0.5310	0.02848	4.246	5.118	0.2745	0.09400	0.0005682	0.03864
		0.7129	0.2672	0.01997	Inf	Inf	Inf	0.000	0.0003595	0.05356
5	PH	0.5327	0.4651	0.002286	961.7	839.7	4.128	0.0005535	0.8884	0.5139
		0.6583	0.3395	0.002246	133.2	68.72	0.4545	0.004916	0.09538	0.2683
		0.3891	0.6064	0.004445	20.92	32.61	0.2390	0.01826	0.01421	0.1653
		0.5293	0.3740	0.09664	13.01	9.195	2.376	0.03909	0.001865	0.05228
		0.3129	0.2815	0.4057	Inf	Inf	Inf	0.000	0.0001235	0.0002183
6	TH	0.5368	0.4605	0.002664	957.1	821.2	4.750	0.0005605	0.8447	0.2414
		0.6303	0.3695	0.0001818	Inf	Inf	Inf	0.000	0.07763	0.2969
		0.4762	0.5238	2.103e-11	4.396e+18	4.836e+18	1.941e+08	1.083e-19	0.06451	0.1542
		0.4755	0.5245	6.601e-11	4.456e+18	4.915e+18	6.186e+08	1.067e-19	0.01230	0.2149
		0.4409	0.5291	0.02999	4.223	5.067	0.2872	0.09455	0.0005470	0.03660
		0.7119	0.2675	0.02055	Inf	Inf	Inf	0.000	0.0003607	0.05591
		0.5387	0.4587	0.002593	6828.	5813.	32.87	7.890e-05	0.7685	0.2700
6	PH	0.5007	0.4987	0.0006118	289.3	288.1	0.3535	0.001728	0.1378	0.3219
		0.6670	0.3312	0.001808	183.5	91.12	0.4975	0.003621	0.08252	0.2569
		0.3504	0.6443	0.005303	28.03	51.53	0.4241	0.01235	0.007775	0.08963
		0.5371	0.3959	0.06700	11.64	8.577	1.451	0.04412	0.003235	0.06133
		0.3180	0.2837	0.3982	Inf	Inf	Inf	0.000	0.0001356	0.0002456



Table S13: Maximum likelihood parameter estimates for several heterozygous models on CEU10 Chr21.

$k$	Data	$\pi_{ref}$	$\pi_{alt}$	$\pi_{err}$	$\alpha_{ref}$	$\alpha_{alt}$	$\alpha_{err}$	$\varphi$	$\rho$	ML- $\rho$
M	TH	0.4989	0.4989	0.002279	Inf	Inf	Inf	0.000	1.000	1.000
BM	TH	0.5428	0.4549	0.002279	Inf	Inf	Inf	0.000	1.000	1.000
1	TH	0.5432	0.4545	0.002379	101.1	84.56	0.4426	0.005346	1.000	1.000
1	PH	0.5530	0.4438	0.003217	23.50	18.86	0.1367	0.02299	1.000	1.000
2	TH	0.5326	0.4651	0.002245	350.1	305.8	1.476	0.001519	0.9217	0.7184
		0.6700	0.3273	0.002700	Inf	Inf	Inf	0.000	0.07827	0.2816
2	PH	0.5355	0.4621	0.002403	Inf	Inf	Inf	0.000	0.6629	0.7140
		0.5841	0.4118	0.004152	10.77	7.594	0.07657	0.05143	0.3371	0.2860
3	TH	0.5309	0.4669	0.002163	1097.	964.3	4.468	0.0004839	0.8918	0.5612
		0.6619	0.3355	0.002684	Inf	Inf	Inf	0.000	0.09611	0.2997
		0.5166	0.4755	0.007905	16.27	14.98	0.2490	0.03077	0.01204	0.1391
3	PH	0.5324	0.4654	0.002176	883.6	772.5	3.611	0.0006022	0.7969	0.6416
		0.6947	0.3020	0.003294	87.15	37.88	0.4132	0.007909	0.1322	0.2628
		0.5123	0.4793	0.008307	5.767	5.396	0.09350	0.08159	0.07087	0.09553
4	TH	0.5338	0.4641	0.002115	Inf	Inf	Inf	0.000	0.7544	0.3976
		0.5168	0.4806	0.002595	122.4	113.8	0.6147	0.004204	0.1511	0.2720
		0.6637	0.3335	0.002733	Inf	Inf	Inf	0.000	0.09385	0.2679
		0.4814	0.4603	0.05827	16.71	15.98	2.023	0.02799	0.0006847	0.06242
4	PH	0.5324	0.4654	0.002212	392.4	343.0	1.631	0.001355	0.8320	0.5835
		0.6956	0.3012	0.003238	77.50	33.56	0.3608	0.008895	0.1449	0.2472
		0.3431	0.6527	0.004234	13.41	25.51	0.1655	0.02495	0.01933	0.1220
		0.5398	0.3514	0.1088	7.280	4.738	1.468	0.06904	0.003795	0.04725
5	TH	0.5339	0.4641	0.002004	3.516e+04	3.056e+04	132.0	1.519e-05	0.7817	0.2977
		0.5001	0.4969	0.002946	206.0	204.7	1.214	0.002422	0.1058	0.3320
		0.6296	0.3661	0.004263	Inf	Inf	Inf	0.000	0.06553	0.1560
		0.6816	0.3168	0.001597	Inf	Inf	Inf	0.000	0.04596	0.1574
		0.4906	0.4588	0.05062	22.66	21.19	2.338	0.02119	0.001050	0.05691
5	PH	0.5243	0.4736	0.002090	5932.	5358.	23.64	8.838e-05	0.7291	0.4591
		0.6179	0.3793	0.002848	201.0	123.4	0.9263	0.003065	0.1763	0.2481
		0.7383	0.2592	0.002564	311.3	109.3	1.081	0.002366	0.06881	0.1240
		0.3441	0.6516	0.004280	13.28	25.14	0.1651	0.02526	0.01939	0.1153
		0.5628	0.3600	0.07718	7.727	4.942	1.060	0.06789	0.006392	0.05343
6	TH	0.5313	0.4666	0.002150	1955.	1716.	7.911	0.0002717	0.8824	0.3782
		0.6584	0.3389	0.002680	Inf	Inf	Inf	0.000	0.05674	0.1515
		0.6632	0.3342	0.002580	Inf	Inf	Inf	0.000	0.04456	0.1326
		0.4605	0.5368	0.002718	90.80	105.8	0.5358	0.005046	0.01430	0.2711
		0.5502	0.4129	0.03695	244.0	183.1	16.38	0.002250	0.001751	0.05318
		0.3934	0.5377	0.06890	9.435	12.90	1.652	0.04003	0.0002428	0.01339
6	PH	0.5272	0.4707	0.002075	Inf	Inf	Inf	0.000	0.7408	0.4040
		0.6241	0.3729	0.002956	178.1	106.4	0.8437	0.003492	0.1588	0.2400
		0.7404	0.2571	0.002503	352.9	122.5	1.193	0.002094	0.06523	0.1182
		0.3978	0.5994	0.002835	89.36	134.7	0.6370	0.004431	0.02257	0.1707
		0.5514	0.3783	0.07030	6.893	4.729	0.8789	0.07406	0.007542	0.05246
		0.2416	0.7559	0.002574	77.09	241.2	0.8214	0.003124	0.005075	0.01462