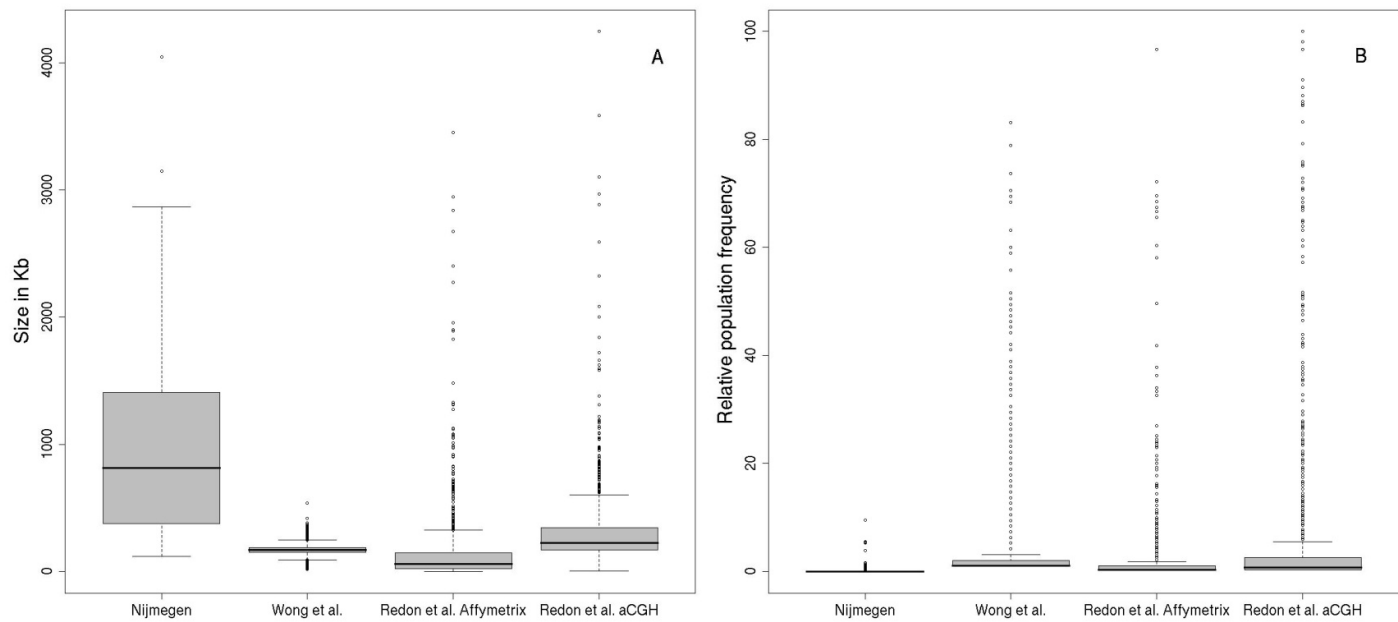


Supplementary material

Supplementary Fig. 1



Supplementary table 1

See separate file

Supplementary table 2

#	synteny break count						ensembl gene count					
regions		obs	exp		p-value	%diff	obs	exp		p-value	%diff	
8051	All SDRs	1658	826	▲	2.00E-03	101	4177	3692	▲	2.00E-03	13	
144	All Nijmegen CNVRs	67	33	▲	2.00E-03	102	2434	1663	▲	2.00E-03	46	
1390	All Redon CNVRs	275	184	▲	2.00E-03	49	5168	4175	▲	2.00E-03	24	
993	All Redon et al. Affymetrix CNVRs	176	120	▲	2.00E-03	47	2277	2004	▲	1.80E-02	14	
889	All Redon et al. aCGH CNVRs	212	128	▲	2.00E-03	65	4724	3644	▲	2.00E-03	30	
4433	All Wong CNVRs	623	544	▲	2.00E-03	15	10974	9779	▲	2.00E-03	12	
815	Wong CNVRs ≥ 3 instances	164	100.17	▲	2.00E-03	64	2808	1833.76	▲	2.00E-03	53	
108	Nijmegen with SDs CNVRs	66	29	▲	2.00E-03	128	2360	1622	▲	2.00E-03	46	
36	Nijmegen without SDs CNVRs	2	6	▼	7.40E-02	-65	133	199	▼	3.20E-02	-33	
279	Redon et al. Affymetrix with SDs CNVRs	90	42	▲	2.00E-03	116	1792	1212	▲	2.00E-03	48	
741	Redon et al. Affymetrix without SDs CNVRs	86	83	▲	3.88E-01	3	485	908	▼	2.00E-03	-47	
439	Redon et al. aCGH with SDs CNVRs	161	70	▲	2.00E-03	128	3787	2436	▲	2.00E-03	55	
497	Redon et al. aCGH without SDs CNVRs	51	65	▼	3.00E-02	-22	937	1414	▼	2.00E-03	-34	
1368	Wong with SDs CNVRs	296	169	▲	2.00E-03	75	4962	3095	▲	2.00E-03	60	
3065	Wong without SDs CNVRs	327	374	▼	2.00E-03	-13	6012	6677	▼	2.00E-03	-10	
29	Frequent Nijmegen CNVRs	23	8	▲	2.00E-03	173	908	476	▲	2.00E-03	91	
268	Frequent Redon et al. Affymetrix CNVRs	72	36	▲	2.00E-03	101	1248	875	▲	2.00E-03	43	
342	Frequent Redon et al. aCGH CNVRs	141	55	▲	2.00E-03	155	3006	1887	▲	2.00E-03	59	
1744	Frequent Wong CNVRs	295	214	▲	2.00E-03	38	5332	4006	▲	2.00E-03	33	
115	Rare Nijmegen CNVRs	45	27	▲	4.00E-03	69	1585	1340	▲	5.60E-02	18	
725	Rare Redon et al. Affymetrix CNVRs	104	84	▲	1.20E-02	24	1029	1134	▼	8.60E-02	-9	
547	Rare Redon et al. aCGH CNVRs	71	74	▼	3.92E-01	-4	1718	1753	▼	3.74E-01	-2	
2689	Rare Wong CNVRs	328	329	▼	4.86E-01	0	5642	5763	▼	1.52E-01	-2	

#	recombination rate per base						dN/dS					
regions		obs	exp		p-value	%diff	obs	exp		p-value	%diff	
8051	All SDRs	0.95584	1.315353	▼	2.00E-03	-27	0.2447	0.1167	▲	2.45E-45	110	
144	All Nijmegen CNVRs	1.41244	1.311361	▲	8.20E-02	8	0.11435	0.1167	▲	2.71E-01	-2	
1390	All Redon CNVRs	1.26742	1.316874	▼	7.60E-02	-4	0.14775	0.1167	▲	1.39E-14	27	
993	All Redon et al. Affymetrix CNVRs	1.18721	1.317337	▼	6.00E-03	-10	0.18055	0.1167	▲	2.73E-19	55	
889	All Redon et al. aCGH CNVRs	1.26984	1.318	▼	9.20E-02	-4	0.146	0.1167	▲	4.62E-12	25	
4433	All Wong CNVRs	1.34675	1.319211	▲	1.40E-02	2	0.1177	0.1167	▼	4.06E-01	1	
815	Wong CNVRs ≥ 3 instances	1.34216	1.31475	▲	2.42E-01	2	0.1132	0.1167	▼	1.03E-01	-3	
108	Nijmegen with SDs CNVRs	1.39459	1.303333	▲	1.32E-01	7	0.1168	0.1167	▲	2.52E-01	0	
36	Nijmegen without SDs CNVRs	1.3357	1.319829	▲	4.38E-01	1	0.0933	0.1167	▼	1.31E-01	-20	
279	Redon et al. Affymetrix with SDs CNVRs	1.06798	1.31832	▼	2.00E-03	-19	0.1904	0.1167	▲	4.14E-20	63	
741	Redon et al. Affymetrix without SDs CNVRs	1.37412	1.318655	▲	2.12E-01	4	0.1409	0.1167	▲	3.00E-02	21	
439	Redon et al. aCGH with SDs CNVRs	1.22253	1.315901	▼	2.80E-02	-7	0.1532	0.1167	▲	1.27E-14	31	
497	Redon et al. aCGH without SDs CNVRs	1.3573	1.316725	▲	2.00E-01	3	0.1154	0.1167	▼	6.21E-01	-1	
1368	Wong with SDs CNVRs	1.26226	1.317676	▼	3.80E-02	-4	0.1345	0.1167	▲	2.08E-06	15	
3065	Wong without SDs CNVRs	1.38422	1.318465	▲	2.00E-03	5	0.10335	0.1167	▼	1.28E-05	-11	
29	Frequent Nijmegen CNVRs	1.47876	1.322704	▲	1.30E-01	12	0.1171	0.1167	▼	8.80E-02	0	
268	Frequent Redon et al. Affymetrix CNVRs	1.14433	1.315755	▼	2.00E-02	-13	0.1985	0.1167	▲	5.08E-15	70	
342	Frequent Redon et al. aCGH CNVRs	1.21878	1.312325	▼	5.40E-02	-7	0.15275	0.1167	▲	2.33E-12	31	
1744	Frequent Wong CNVRs	1.36523	1.318535	▲	3.40E-02	4	0.1172	0.1167	▼	3.10E-01	0	
115	Rare Nijmegen CNVRs	1.35743	1.309264	▲	2.84E-01	4	0.1113	0.1167	▲	4.30E-01	-5	
725	Rare Redon et al. Affymetrix CNVRs	1.21929	1.313298	▼	1.08E-01	-7	0.1659	0.1167	▲	9.70E-07	42	
547	Rare Redon et al. aCGH CNVRs	1.31817	1.318363	▲	4.98E-01	0	0.1319	0.1167	▲	1.33E-02	13	
2689	Rare Wong CNVRs	1.3341	1.318227	▲	2.32E-01	1	0.1187	0.1167	▼	3.42E-01	2	

#	simple repeat bases count						dS					
regions		obs	exp		p-value	%diff	obs	exp		p-value	%diff	

8051	All SDRs	9196905	3649323	▲	2.00E-03	152	0.5277	0.3818	▲	2.67E-37	38
144	All Nijmegen CNVRs	11453796	3613443	▲	2.00E-03	217	0.55425	0.3818	▲	1.02E-69	45
1390	All Redon CNVRs	17292594	8523831	▲	2.00E-03	103	0.47195	0.3818	▲	3.53E-42	24
993	All Redon et al. Affymetrix CNVRs	9345890	3887177	▲	2.00E-03	140	0.48235	0.3818	▲	7.79E-20	26
889	All Redon et al. aCGH CNVRs	15263851	7600250	▲	2.00E-03	101	0.4739	0.3818	▲	9.29E-41	24
4433	All Wong CNVRs	21975067	19359088	▲	2.00E-03	14	0.4246	0.3818	▲	9.45E-26	11
815	Wong CNVRs ≥ 3 instances	7268818	3638305	▲	2.00E-03	100	0.4968	0.3818	▲	4.85E-41	30
108	Nijmegen with SDs CNVRs	11906225	3450321	▲	2.00E-03	245	0.5545	0.3818	▲	3.70E-65	45
36	Nijmegen without SDs CNVRs	327800	420914.1	▼	3.52E-01	-22	0.5214	0.3818	▲	1.00E-06	37
279	Redon et al. Affymetrix with SDs CNVRs	8179692	2540456	▲	2.00E-03	222	0.5093	0.3818	▲	7.91E-23	33
741	Redon et al. Affymetrix without SDs CNVRs	1166198	1624567	▼	4.00E-03	-28	0.4137	0.3818	▲	3.56E-02	8
439	Redon et al. aCGH with SDs CNVRs	13087002	5184745	▲	2.00E-03	152	0.4812	0.3818	▲	9.25E-44	26
497	Redon et al. aCGH without SDs CNVRs	2176849	2907909	▼	2.00E-03	-25	0.4216	0.3818	▲	2.22E-02	10
1368	Wong with SDs CNVRs	9589957	6148866	▲	2.00E-03	56	0.4277	0.3818	▲	2.68E-17	12
3065	Wong without SDs CNVRs	12385110	13234880	▼	9.00E-02	-6	0.42145	0.3818	▲	1.58E-14	10
29	Frequent Nijmegen CNVRs	3722082	1060445	▲	2.00E-03	251	0.6409	0.3818	▲	3.55E-43	68
268	Frequent Redon et al. Affymetrix CNVRs	5251542	1781521	▲	2.00E-03	195	0.5109	0.3818	▲	3.23E-18	34
342	Frequent Redon et al. aCGH CNVRs	10419754	4028090	▲	2.00E-03	159	0.48575	0.3818	▲	7.23E-42	27
1744	Frequent Wong CNVRs	12260746	7987872	▲	2.00E-03	53	0.4616	0.3818	▲	3.92E-39	21
115	Rare Nijmegen CNVRs	8511943	2863115	▲	2.00E-03	197	0.5197	0.3818	▲	3.41E-34	36
725	Rare Redon et al. Affymetrix CNVRs	4094348	2115203	▲	2.00E-03	94	0.4479	0.3818	▲	3.08E-05	17
547	Rare Redon et al. aCGH CNVRs	4844097	3608535	▲	8.00E-03	34	0.4398	0.3818	▲	4.87E-10	15
2689	Rare Wong CNVRs	9714321	11358645	▼	2.00E-03	-14	0.3925	0.3818	▲	1.79E-03	3

Supplementary Table 3.

Set	<i>lethality- embryonic/perinatal</i>	<i>lethality- postnatal</i>
All Nijmegen CNVRs	-2%	-30%*
Frequent Nijmegen CNVRs	-15%	-35%
Gain-and-Loss Nijmegen CNVRs	-12%	-43%*
Rare Nijmegen CNVRs	+ 6%	-28%
Nijmegen CNVRs with SDs	-3%	-33%**
Nijmegen CNVRs w/o SDs	+23%	-4%
All Redon <i>et al.</i> aCGH CNVRs	-18%*	-14%
Frequent Redon <i>et al.</i> aCGH CNVRs	-21%*	-13%
Gain-and-Loss Redon <i>et al.</i> aCGH CNVRs	-31%*	-12%
Rare Redon <i>et al.</i> aCGH CNVRs	-15%	-15%
Redon <i>et al.</i> aCGH CNVRs with SDs	-19%**	-6%
Redon <i>et al.</i> aCGH CNVRs w/o SDs	-18%	-41%*
All Redon <i>et al.</i> Affymetrix CNVRs	-50%**	+ 4%
Frequent Redon <i>et al.</i> Affymetrix CNVRs	-53%*	-2%
Gain-and-Loss Redon <i>et al.</i> Affymetrix CNVRs	-57%*	-46%
Rare Redon <i>et al.</i> Affymetrix CNVRs	-47%*	+10%
Redon <i>et al.</i> Affymetrix CNVRs with SDs	-43%**	-15%
Redon <i>et al.</i> Affymetrix CNVRs w/o SDs	-73%**	+53%
All Wong <i>et al.</i> CNVRs	0%	-16%*
Frequent Wong <i>et al.</i> CNVRs	-10%	-21%*
Gain-and-Loss Wong <i>et al.</i> CNVRs	-18%	-15%
Rare Wong <i>et al.</i> CNVRs	+10%	-11%
Wong <i>et al.</i> CNVRs with SDs	-11%*	-18%*
Wong <i>et al.</i> CNVRs w/o SDs	+8%	-15%*