Revising the human mutation rate: implications for understanding human evolution

Supplementary information

Aylwyn Scally and Richard Durbin
Wellcome Trust Sanger Institute, Hinxton, UK

Supplementary Table: Measurements of the *de novo* mutation rate in modern humans
The table below shows published per-generation mean mutation rate estimates with 95% confidence intervals (where reported) in studies of *de novo* mutations in modern humans, with corresponding yearly mutation rates assuming 30 year and 25 year mean generation times. The loci considered in each case depended on study methodology: either mutations at selected dominant or X-linked disease genes (disease), whole genome sequencing (WG), or exome sequencing (exome). Some variation in estimated rate is expected to be associated with this factor, in particular an expectation of higher mutation rate around coding regions, which are GC rich (Box 2; Sanders et al. 2012).

The exome sequencing studies of Sanders et al. (2012) and O'Roak et al. (2012) reported mutation rates for individuals affected by autism spectrum disorder (ASD) as well as for unaffected siblings; the latter are quoted here. *De novo* rates for ASD-affected individuals were found to be higher in these studies than in their siblings, in particular at non-synonymous sites.

Two estimates were made by the 1000 Genomes Project, one from sequencing of a trio with European ancestry (CEU) and one from a trio with Yoruba (African) ancestry (YRI).

study	loci considered	per-generation mean mutation rate (10^{-8} bp^{-1}generation^{-1})	yearly mean mutation rate (10^{-9} bp^{-1}y^{-1})	
			t_{gen} = 30 y	t_{gen} = 25 y
Kondrashov (2003)	disease	1.85 (0.00–3.65)	0.62 (0.00–1.22)	0.74 (0.00–1.46)
Lynch (2010)	disease	1.28 (0.68–1.88)	0.42 (0.23–0.63)	0.51 (0.27–0.75)
Roach *et al*. (2010)	WG	1.10 (0.68–1.70)	0.37 (0.23–0.57)	0.44 (0.27–0.68)
Awadalla *et al*. (2010)	WG	1.36 (0.34–2.72)	0.45 (0.11–0.91)	0.54 (0.14–1.09)
1000 Genomes Project (2010), CEU	WG	1.17 (0.94–1.73)	0.39 (0.31–0.57)	0.47 (0.38–0.69)
1000 Genomes Project (2010), YRI	WG	0.97 (0.72–1.44)	0.32 (0.24–0.48)	0.39 (0.29–0.58)
Sanders *et al*. (2012)	exome	1.28 (1.05–1.50)	0.43 (0.35–0.50)	0.51 (0.42–0.60)
O'Roak *et al*. (2012)	exome	1.57 (1.05–2.26)	0.52 (0.35–0.75)	0.63 (0.42–0.90)
Kong *et al*. (2012)	WG	1.20	0.40	0.48

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