

GENETICS

DO WE ALL HAVE (COMPENSATED) MONOGENIC TRAITS?

2.11.2010

I have made several attempts here to estimate the number of somatic mutations: 3,93 [mutations / Mb](#)(2007), [the long fuse](#) (2007), about the cancer [the legacy](#) (2010) or [1/day](#) in the life of a mutation (2010) but the best summary [now](#) comes from the [1000 genomes](#)

From the two trios, we directly estimate the rate of de novo germline base substitution mutations to be approximately 10^{-8} per base pair per generation.

along with the fact that

on average, each person is found to carry approximately 250 to 300 loss-of-function variants in annotated genes and 50 to 100 variants previously implicated in inherited disorders.

which raises some doubts that these 50 to 100 have only been implicated but not proven...

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