Heredity (2008) 101, 297–298

www.nature.com/hdy

NEWS AND COMMENTARY

Genome size evolution

Within-species variation in genome size

C Biémont

Heredity (2008) **101,** 297–298; doi:10.1038/hdy.2008.80; published online 30 July 2008

From the thousands of species that have already been analyzed, it is now clear that genome size varies considerably between species. This variation is mainly because of the differing amounts of various repeated sequences, including satellite DNA, transposable elements (TEs) and ribosomal genes in different genomes. The idea of within-species variation, that is, variation between populations and between individuals, has won less general acceptance, despite various studies revealing its existence. This is sometimes because of mistrust and suspicion (Gregory and Johnston, 2008), sometimes to the concept underlying the definition of species (Murray, 2005) and sometimes to artefacts in the technique used (Greilhuber, 2005; Gregory and Johnston, 2008). For example, some of the variation observed in genome size between populations subjected to different environmental conditions over the geographical range of the species can be accounted for by the chromatin state, which can affect the accessibility of the DNA to fluorochromes (see Nardon *et al*., 2003 and references therein). However, we do not have to throw the baby out with the bath water as all these confounding factors can be taken into account by using appropriate protocols and statistical methods, which take different levels of sampling and replicates into account, for determining within- and between-species variation. A recent study of genome size in various *Drosophila* species (Bosco *et al*., 2007) concludes that there is a significant statistical difference in genome size between species, and some significant differences between strains in some species. Vieira *et al*. (1999, 2002) have also identified genome-size differences between populations of both *Drosophila melanogaster* and *D. simulans*, but no differences between replicates of batches of flies reared in the same vial. In the Bosco *et al*. (2007) paper, the variation in genome size was related to variation in heterochromatin composition, mostly of satellite DNA, whereas TEs were mostly involved in the Vieira *et al*. (1999, 2002) study. Such data give credibility to between-individual variations in the amount of heterochromatic TEs that have been estimated using dot blots and *in situ* hybridization in *Drosophila* (Charlesworth *et al*., 1994). These results show that both TEs and satellite DNA can influence genome size, and lead to within-species variation. Satellite DNA is indeed a major constituent of the genomes of many organisms (see Palomeque and Lorite, 2008 for a review), and it explains why the genome of the species *D. orena* (0.28 pg) is so much larger than that of *D. melanogaster* (0.18 pg) (Boulesteix *et al*., 2006). Marked differences in genome size between closely related lines of flax (*Linum usitatissimum*) have been shown to occur over a very short period, and to involve a change in rDNA amount (Cullis, 2005; Davison *et al*., 2007), as well as specific LIS-1 insertion events, although possible changes in the amount of other repetitive sequences were not determined. Mobilization and integration of novel copies of TEs are not always observed in genome changes, which can simply result from segmental duplication of centromeric regions, as reported in rice (Ma and Jackson, 2006).

Centromeres and the Y chromosome seem to have a major function in the change in genome size reported both within- and between-species. In most species, the heterochromatic Y chromosome consists mainly of numerous repeated sequences, mostly satellites and TEs (Palomeque and Lorite, 2008), within which only a few genes, mostly fertility genes, are embedded. For instance, it has been reported that in *Anopheles gambiae* the fully heterochromatic Y chromosome varies in length and banding pattern in natural populations, a finding consistent with the rapid satellite expansion mechanism in this species (Bonaccorsi *et al*., 1980). Hence, in addition to changes in centromere composition and whole genome structure, the Y chromosome, because of its highly degenerated heterochromatic state (Charlesworth, 2002) and the fact that it differs in size in different populations, seems to make a particular contribution to the variation in genome size, as reported in some plants (Meagher *et al*., 2005). This implies that the suggestion that changes in the composition of the Y chromosome could be responsible for geographical adaptive genetic differences between temperate and tropical populations of *Drosophila melanogaster* (Rohmer *et al*., 2004) warrants further investigation.

The reasons for differences in the amounts of repeated sequences present in different populations are not clearly understood. In addition to the impact of the effective population size (Lynch and Conery, 2003), at least two explanations are conceivable. A global change could have occurred as a result of the stressful environmental conditions encountered by the species when they invaded new geographical areas, or of confrontations between populations that had not encountered each other earlier (Biémont and Vieira, 2005 and references therein). TE transposition and amplification have been reported in inter-species hybrids in *Drosophila* and in the Australian wallaby (see references in Biémont and Vieira, 2005), but few data are available about intra-species hybridization. Only certain specific TEs, as in dysgenic crosses in *Drosophila* (Kidwell, 2002), or other kinds of repeated sequence, may be involved. A survey of various TEs during mating between *Magnaporthe grisea* isolates from Triticum and Setaria revealed new insertions of the LTR (Long Terminal Repeat) retrotransposon MAGGY in the progeny, but very rarely any movements of other TEs (Eto *et al*., 2001). Transposition of the LINE (Long Interspersed Nuclear Element: non-LTR retrotransposon) MGL (*M. grisea* LINE) was also observed in another experiment in which two *M. grisea* isolates from rice were crossed (Nishimura *et al*., 2000), and the Ds-like element, Ascot-1, has been shown to excise during mating in *Ascobolus immersus* b2 (Colot *et al*., 1998). No evidence of TE amplification was, however, found in crosses between *A. nidulans* strains (Li Destri Nicosia *et al*., 2001). These findings strongly suggest that the transposition of TEs may be enhanced during mating in fungi, but only some TE families are involved.

Although amplification, deletion and rearrangements of various repeated DNA sequences do indeed account for differences in DNA composition, the frequency of such events and their

impact on natural populations are some of the main questions facing population genomics today. As repeated sequences interfere with gene regulation, for example, in human populations (Lippman *et al*., 2004), changes in genome composition and of whole genome structure (Parfrey *et al*., 2008) in terms of such sequences, may have a considerable impact on population adaptation, and also promote specific illnesses. The possible impact of such variations therefore warrants detailed investigation in human populations.

C Biémont is at the Laboratoire de Biométrie et Biologie Evolutive, UMR 5558, CNRS, Université Lyon 1, Université de Lyon. 69622 Villeurbanne Cedex, France.

e-mail: biemont@biomserv.univ-lyon1.fr

Biémont C, Vieira C (2005). What transposable elements tell us about genome organisation and evolution? The case of *Drosophila*. *Cytogenet Genome Res* **110**: 25–34.

Bonaccorsi S, Santini G, Gatti M, Pimpinelli S, Colluzzi M (1980). Intraspecific polymorphism of sex chromosome heterochromatin in two species of the *Anopheles gambiae* complex. *Chromosoma* **76**: 57–64.

Bosco G, Campbell P, Leiva-Neto JT, Markov TA (2007). Analysis of *Drosophila* species genome size and satellite DNA content reveals significant differences among strains as well as between species. *Genetics* **177**: 1277–1290.

Boulesteix M, Weiss M, Biémont C (2006). Changes in genome size between closely related species: the *Drosophila melanogaster* species subgroup. *Mol Biol Evol* **23**: 162–167.

Charlesworth B, Jarne P, Assimacopoulos S (1994). The distribution of transposable elements within and between chromosomes in a population of *Drosophila melanogaster*. III. Element abundances in heterochromatin. *Genet Res* **64**: 183–197.

Charlesworth B (2002). The evolution of chromosomal sex determination. *Novartis Found Symp* **244**: 207–219.

Colot V, Haedens V, Rossignol J-L (1998). Extensive, nonrandom diversity of excision footprints generated by *Ds*-like transposon *Ascot-1* suggests new parallels with V(D)J recombination. *Mol Cell Biol* **18**: 4337–4346.

Cullis CA (2005). Mechanisms and control of rapid genomic changes in flax. *Ann Bot* **95**: 201–206.

Davison J, Tyagi A, Comai L (2007). Large-scale polymorphism of heterochromatic repeats in the DNA of *Arabidopsis thaliana*. *BMC Plant Biol* **7**: 44.

Eto Y, Ikeda K, Chuma I, Kataoka T, Kuroda S, Kikuchi N *et al.* (2001). Comparative analyses of the distribution of various transposable elements in *Pyricularia* and their activity during and after the sexual cycle. *Mol Gen Genet* **264**: 565–577.

Gregory TR, Johnston JS (2008). Genome size diversity in the family Drosophilidae. *Heredity* (advance online publication, 4 June 2008; doi:10.1038/hdy.2008.49).

Greilhuber J (2005). Intraspecific variation in genome size in angiosperms: identifying its existence. *Ann Bot* **95**: 91–98.

Kidwell MG (2002). Transposable elements and the evolution of genome size in eukaryotes. *Genetica* **115**: 49–63.

Li Destri Nicosia MG, Brocard-Masson C, Demais S, Hua Van A, Daboussi MJ, Scazzocchio C (2001). Heterologous transposition in *Aspergillus nidulans*. *Mol Microbiol* **39**: 1330–1344.

Lippman Z, Gendrel AV, Black M, Vaughn MW, Dedhia N, McCombie WR *et al.* (2004). Role of transposable elements in heterochromatin and epigenetic control. *Nature* **430**: 471–476.

Lynch M, Conery JS (2003). The origins of genome complexity. *Science* **302**: 1401–1404.

Ma J, Jackson SA (2006). Retrotransposon accumulation and satellite amplification mediated by segmental duplication facilitate centromere expansion in rice. *Genome Res* **16**: 251–259.

Meagher TR, Gillies AC, Costich DE (2005). Genome size, quantitative genetics and the genomic basis for flower size evolution in *Silene latifolia*. *Ann Bot* **95**: 247–254.

Murray BG (2005). When does intraspecific C-value variation become taxonomically significant? *Ann Bot* **95**: 119–125.

Nardon C, Weiss M, Vieira C, Biémont C (2003). Variation in genome size estimate with environmental conditions in *Drosophila melanogaster*. *Cytometry* **55**: 43–49.

Nishimura M, Hayashi N, Jwa NS, Lau GW, Hamer JE, Hasebe A (2000). Insertion of the LINE retrotransposon MGL causes a conidiophore pattern mutation in *Magnaporthe grisea*. *Mol Plant Microbe Interact* **13**: 892–894.

Palomeque T, Lorite P (2008). Satellite DNA in insects: a review. *Heredity* **100**: 564–573.

Parfrey LW, Lahr DJG, Katz LA (2008). The dynamic nature of eukaryotic genomes. *Mol Biol Evol* **25**: 787–794.

Rohmer C, David JR, Moreteau B, Joly D (2004). Heat induced male sterility in *Drosophila melanogaster*: adaptive genetic variations among geographic populations and role of the Y chromosome. *J Exp Biol* **207**: 2735–2743.

Vieira C, Lepetit D, Dumont S, Biémont C (1999). Wake up of transposable elements following *Drosophila simulans* worldwide colonization. *Mol Biol Evol* **16**: 1251–1255.

Vieira C, Nardon C, Arpin C, Lepetit D, Biémont C (2002). Evolution of genome size in *Drosophila*. Is the invader's genome being invaded by transposable elements?. *Mol Biol Evol* **19**: 1154–1161.

Editor's suggested reading

Johnson L (2008). Transposon silencing: the extraordinary epigenetics of a transposon trap. *Heredity* **100**: 5.

Sabot F, Schulman AH (2006). Parasitism and the retrotransposon life cycle in plants: a hitchhiker's guide to the genome. *Heredity* **97**: 381–388.

Schlötterer C (2008). *Drosophila* unleashed: the 12-genome update of the genetic power horse has been released. *Heredity* **100**: 337–338.

Zakharenko LP, Kovalenko LV, Mai S (2007). Fluorescence *in situ* hybridization analysis of *hobo*, *mdg1* and *Dm412* transposable elements reveals genomic instability following the *Drosophila melanogaster* genome sequencing. *Heredity* **99**: 525–530.