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Review

The evolving fungal genome

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ABSTRACT

Fungal genomes vary considerably in size and organization. The genome of *Microsporidium* contains less than 3 Mb while the genomes of several Basidiomycetes and Ascomycetes greatly exceed 100 Mb. Likewise chromosome numbers and ploidy levels can differ even between closely related species. The differences in genome architecture among fungi reflect the interplay of different mutational processes as well as the population biology of the different species. Comparative genome studies have elucidated the underlying mechanisms of genome evolution in different groups of fungi and have provided insight into species-specific genomic traits. Mobile genetic elements have been instrumental in shaping the genome architecture and gene content in many fungal species. In many pathogenic fungi the mobile genetic elements even play a crucial role in rapid adaptive evolution by mediating high rates of sequence mutations, chromosomal rearrangements, and ploidy changes. But in many species mobile elements are efficiently restricted by defense mechanisms, which have evolved to suppress and regulate parasitic elements. Different rates of genome dynamic and adaptive evolution may reflect varying effective population sizes through which genetic drift and natural selection have differentially affected genome architecture in fungi over time.

1. Introduction

Since the first fungal (and eukaryotic) genome of *Saccharomyces cerevisiae* was published in 1996 (Goffeau *et al.*, 1996) several hundred fungal genomes have followed. Large-scale community sequencing programs such as the 1000 Fungal Genomes Project (http://1000.fungalgenomes.org) and the Fungal Genome Initiative (http://www.broadinstitute.org/scientific-community/science/projects/fungal-genome-initiative) will provide an even broader representation of genome data from all major fungal groups. Beyond descriptive analyses of genome compositions, novel questions relating to mechanisms of genome evolution can be addressed. Most studies describing genome evolution in fungi have focused on pathogenic species except studies of model species such as species of *Saccharomyces* and *Neurospora* (see e.g. Ellison *et al.*, 2011; Scannell *et al.*, 2007; 2006). Host driven rapid evolution in pathogenic species is reflected as high levels of genomic variation. A key question of interest is whether the rapid genome dynamic of pathogenic species is a particularity associated with parasitic life style. Below we will discuss and focus on aspects of genome evolution including the importance of mobile genetic elements, genome defenses, and chromosomal plasticity in pathogenic species. The relative ease of

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sequencing and assembling significant numbers of isolates from the same and related species will make fungi a prime group of organisms to study eukaryotic genome evolution.

2. Mobile genetic elements

Mobile elements have played a crucial role in shaping the architecture and gene contents of fungal genomes. With the sequencing of genomes from a broad range of fungal species it becomes apparent that invasions and expansions of mobile genetic elements have affected genome evolution to very different extents. Some genomes bear the signatures of either recent or ancient expansions of mobile elements. Others show mutational signatures of genome defense mechanisms that act or have acted to efficiently prevent the spread of mobile elements.

Mobile genetic elements predominantly have a neutral or negative effect on their host through the mutational consequences of element mobilization. However, as for all mutational changes, some mutations may confer a fitness advantage to the host. Studies using comparative and population genomic data of pathogenic species have focused on the selective advantage of mutational changes conferred by mobile genetic elements (reviewed in Raffaele and Kamoun, 2012). High levels of genetic diversity associated with repetitive sequences often characterize genomes of pathogenic species and it has been proposed that this diversity is adaptive (de Jonge *et al.*, 2013; Manning *et al.*, 2013; Neafsey *et al.*, 2010). Combined genomic and functional studies confirm the importance of mobile elements in the creation of novel genes, effector diversification, horizontal gene transfer, copy number variation, and trans-duplications (de Jonge *et al.*, 2012; Friesen *et al.*, 2006; Manning *et al.*, 2013; Rouxel *et al.*, 2011; Xue *et al.*, 2012). Therefore, mobile elements indeed can play a significant role in the adaptation of pathogens to rapidly evolving host defenses. Both sexual and asexual species can benefit from the mutational effect of mobile elements. In asexual species the mutational affect of mobile elements may even circumvent the lack of allele shuffling that comes with recombination. This was documented in the mainly asexual plant pathogenic fungus *Verticillium dahliae* where extensive chromosomal rearrangements generate repetitive and highly dynamic lineage-specific genomic regions (de Jonge *et al.*, 2013). Several effector genes located within the repetitive lineage-specific regions were shown to be determinants of host specificity suggesting that these repetitive islands provide an important genetic basis for pathogen adaptation (de Jonge *et al.*, 2013; 2012). Other studies of plant pathogenic and endophytic species likewise report the occurrence of rapidly evolving repetitive islands encoding pathogenicity related genes or genes involved in the production of alkaloid, herbivore deterrents (Fedorova *et al.*, 2008; Gout *et al.*, 2006; Schardl *et al.*, 2013). The restriction of mobile element activity to particular genomic regions reduces potential deleterious effects on housekeeping genes in the core genome. To what extent this “targeted” insertion of mobile elements is regulated, is poorly understood except from insertion of Ty1 elements in the yeast genome (see below) (Blanc and Adams, 2004). “Non-targeted” insertions are likely to have deleterious effects by the interruption of coding or regulatory sequences, and such insertions are strongly selected against.

Mobile elements may not only cause genomic rearrangements but also cause significant genome expansions. The 125 Mb genome of the black truffle fungus *Tuber melanosporum* consists of 58 % repetitive sequences resulting from a wave of retro-transposition approximately 5 million years ago (Martin *et al.*, 2010). *T. melanosporum* belongs to the Pezizomycotina together with *Neurospora crassa*. In contrast to *T. melanosporum*, the 40 Mb genome of *N. crassa* has a low content of repetitive DNA (10 %) and possesses very efficient genome defense mechanisms (Cambareri *et al.*, 1991; Galagan *et al.*, 2003; Selker *et al.*, 2003). In addition to distinct mechanisms of genome defense, differences in population biology and effective population sizes of *N. crassa* and *T. melanosporum* may also, as discussed below, explain these considerable differences in genome size and content.

Even closely related species belonging to the same genus can show distinctive patterns of mobile element activity. This is the case for species of the Ascomycete genus *Fusarium*. The 60 Mb genome of *F. oxysporum f. sp. lycopersici* is characterized by a high content of repetitive sequences (17 %) and transposable elements (4 %) (Ma *et al.*, 2010). In comparison the genome of a related species *F. graminearum* is noticeably smaller (36 Mb) and contains only 0.24 % repetitive sequences and 0.03 % transposable elements. Also in terms of genome structure the two *Fusarium* species differ substantially: The genome of *F. oxysporum f. sp. lycopersici* is composed of 14 chromosomes while the genome of *F. graminearum* only contains 4. The expansion and restructuring of the *F. oxysporum f. sp. lycopersici* genome is considered to result from the activity of mobile elements (Ma *et al.*, 2010).

Similarly to *F. oxysporum f. sp. lycopersici* and *T. melanosporum*, the genome of the ascomycete pathogen *Blumeria graminis* has been strongly affected by the activity of mobile genetic elements (Spanu *et al.*, 2010). The large genome (~120 Mb) of *B. graminis* consists of 64 % repetitive sequences including both transposable elements and retro-elements. Interestingly, the genome expansion of *B. graminis* has been accompanied by a massive gene loss that reflects the adaptation to obligate biotrophy of the pathogen. The adaptive loss of genes was directly mediated by the activity of mobile elements in the genome of the fungus.

LTR retrotransposons in *S. cerevisiae* have been a model in studies of retrotransposon evolution. The genome of *S. cerevisiae* contains two closely related families of LTR retrotransposons Ty1 and Ty2. To assess the age and dynamic of Ty elements Promislow and colleagues collected sequence data from all Ty elements in the yeast genome and further developed a demographic model based on the expected copy number and divergence of elements (Promislow *et al.*, 1999). The authors could demonstrate that the expected and observed age-distribution of Ty elements, at a steady-state birth-death model, follows an L-shaped exponential distribution for elements of different states of divergence (Fig 1). The model allowed the authors to estimate both the effective transposition and excision rates (fixed transposition events) in the yeast genome and to determine the age of the initial Ty genome invasion. Similar analyses can be applied to genome data of