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Perspective

Gene Duplications and Fates of Genes after Duplication Events

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Gene duplications are considered to be major genetic basis for producing novel genetic variations. There are three types of gene duplications: Whole Genome (WGD), segmental and small scale duplications by tandem duplications [1]. Example of gene duplications of gene duplications events are frequently found in vertebrate serpins, which are classified into six groups V1-V6 [2]. Several tandem duplications on the same locus lead into several paralogs for groups V1 on the human chromosomes 6 and 18 [3] and V2 on the human chromosome 14 [4]. In contrast, there are several serpins which are localized on a single gene in the chromosomal fragments like angiotensinogen [chromosomes 1[5]], heparin cofactor II [chromosomes 22 [6]], C1 inhibitor [chromosomes 22 [7]] and antithrombin III [chromosomes 1 [8]], respectively. Similar trends are also followed by serpins from invertebrates like urochordates [9].

About five decades ago, Susumu Ohno proposed his famous hypothesis that early vertebrates have undergone two rounds of WGD events and which is known as Ohno's hypothesis or 2R-hypothesis [10,11]. These events cause massive gene duplications that are the hallmarks of gene and functional innovation. There were not one who believed on this hypothesis in the beginning years; later same theory become the cornerstone of gene duplications and fates [10,11], specially in the post-genomic era [12]. It is now clear that second WGD has also occurred in fishes and this often called as Fish-Specific Genome Duplication (FSGD) or 3R-hypothesis and it is best exemplified in the Hox clusters duplications [13].

On the evolutionary scale, gene duplication events lead into several schemes (Figure 1).

This includes following schemes:

a) Nonfunctionalization is random loss of function in one of the two gene copies by pseudogenization (Figure 1A). Large fraction of duplicated genes are pseudogenized over 100 MY in the rainbow trout [14].

b) Neofunctionalization is when one gene copy may retain the original function while the other acquires a novel, evolutionarily advantageous/adaptive function [15].

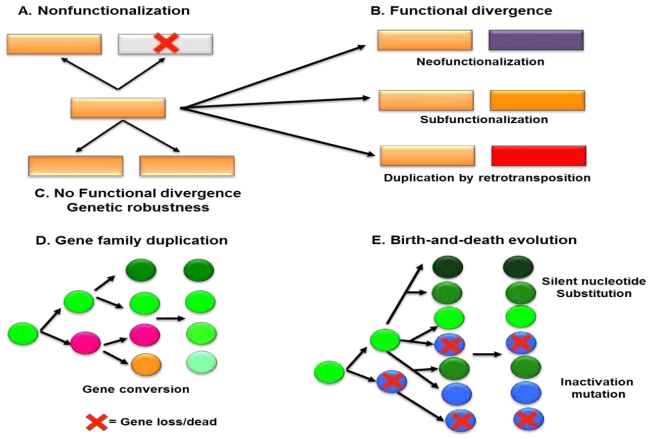

Figure 1: Fate of duplicated single genes (A-C) and duplicated gene families (D-E).
Modified from Conrad and Antonarakis [1].

c) Subfunctionalization is after duplication, mutations may occur in both genes that specialize to perform complementary functions [16,17].

There are several puzzles about how duplicate genes are retained during evolution. Several models have been proposed over in last four decades. Classical duplication-degeneration-complementation/subfunctionalization models do not invoke positive selection; however, this can impose higher rates for retaining duplicate genes in small populations only. Rodents have higher retentions rates duplicate genes and only few losses in comparison to humans which corroborates that positive selections are more instrumental players than previously assumed [18].

Suppose a condition where two redundant gene copies were retained in the genome without significant functional divergence, this can covenant increase genetic robustness against harmful mutations in the concerned species (Figure 1C). Within multigene families descended from a common ancestor, these genes possess similarities at the DNA level, which implies for similar functions [19,20]. The tandemly duplicated genes which generate several paralogous on the same chromosomal fragments (like serpin paralogous in the human chromosomes 6-18 [3] and 14 [4]), exhibit the case of concerted evolution. Within this concept, all genes in a given group evolve coordinately by homologous recombination, which vanguard into gene conversion (Figure 1D). These paralogs share higher sequence identities like anti-trypsin-like gene cluster in the human chromosome 14 [4]. For large fraction of multigene families, the evolutionary model of birth-and-death (aka gain-and-loss) is largely supporting model, which propound that protein sequence similarities within family members is pronounced by strong purifying selection

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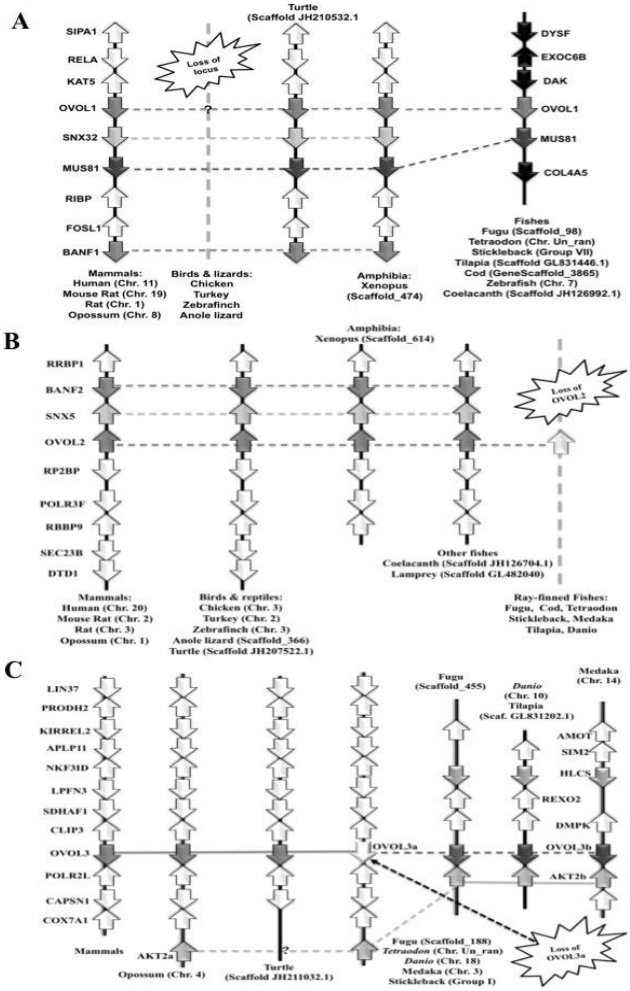

Figure 2: Birth-and-death model of gene evolution by example of vertebrate OVO-like transcription factors family.
A) Loss of OVOL1 and its locus in birds and lizards.
B) Loss of OVOL2 in ray-finned fishes
C) Loss of original OVOL3a gene, which is compensated by duplicated OVOL3b gene in ray-finned fishes.
Modified from Kumar et al. [21].

and evolvements of individual genes are primarily occur only by synonymous substitutions (Figure 1E) [19,20].

Vertebrate OVO-like transcription factors family depicts a good example of birth-and-death model of gene evolution (Figure 2). This family has lineage specific birth and dead of genes as reported earlier [21]. Birds and lizards have loss of OVOL1 and its entire locus, while ray-finned fishes have loss of entire OVOL2 locus. Fishes have duplication of OVOL3, generating original OVOL3a and duplicated OVOL3b. Subsequently fishes lost OVOL3a and OVOL3b has compensated for this loss.

For further details of models of gene duplications and their fates, it is recommended to see a good review by Innan and Kondrashov [22] on this topic.

Overall, I have covered briefly the concepts that driving gene duplications and fates of genes after duplication events.

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