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# New Gene Origins, Evolution and Phenotypic Impact

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## Introduction

Since the pre-molecular era, the emergence of new genes with novel functions has been regarded as a significant contributor to adaptive evolutionary innovation. In this paper, I discuss the origin and evolution of new genes and their functions in eukaryotes, a field of study that has advanced rapidly in the last decade thanks to the genomics revolution. Indeed, recent research has provided preliminary whole-genome views of various types of new genes for a wide range of organisms [1]. The range of mechanisms underlying the origin of new genes is enticing, going far beyond the previously well-studied source of gene duplication.

## Description

De novo gene birth is the process by which new genes emerge from previously noncoding sequences. Over the last decade, researchers have used yeast as a model and a tool to investigate the evolutionary mechanisms and physiological implications of de novo gene birth. We review the mechanisms proposed to explain how noncoding sequences can become proteincoding genes, focusing on the discovery of pervasive translation of the yeast transcriptome and its presumed impact on evolutionary innovation. We review current best practises for identifying and characterising de novo genes. We emphasise that the field is still in its infancy, with the physiological roles of most young yeast de novo genes identified thus far remaining unknown [2].

Ohno and colleagues proposed the importance of duplications in the evolution of new gene functions over 40 years ago. Since then, our knowledge and understanding of gene and genome evolution has grown enormously. Both computational and experimental approaches show that gene loss and gain are common in the primate lineage, with much of this occurring within or mediated by duplicated sequences. Because of the dynamism and complexity of these changes, molecular comparative genomic studies have become more difficult. Nonetheless, the available data clearly show that this variation is critical for understanding our species' evolution and phenotypic variation [3].

Recent Neandertal and Denisovan sequences provide high-depth coverage when aligned to the human reference genome, which is the same assembly used to compile 1KG deletions. As a result, we simply "genotyped" the 1KG deletions using read-depth data from high-coverage Neandertal and Denisovan genomes to detect human deletion variants shared with archaic hominins. In a nutshell, we counted the number of Denisovan and Neandertal reads that mapped to a specific interval in the human reference genome where a deletion polymorphism was previously discovered in modern humans. The number of reads in these regions, as expected, correlates well with size in both Neandertal and Denisovan sequences (R2 = 0.8582 and 0.8713, respectively).

We used polymorphic human deletions shared with the Denisovan genome but not with the Neandertal genome to independently estimate potential misclassification of low-frequency ancient deletions as introgressed. We expect no Denisovan introgression in the 1KG deletions because no samples from Melanesian or other South East Asian populations have been reported to have Denisovan introgression. As a result, the proportion of polymorphic deletions shared with only Denisovans and classified as "introgressed" by our pipeline will provide an indirect estimate of misclassification. Based on this, we estimate that only 5 of the 102 deletions were incorrectly classified as introgressed by our pipeline [4,5].

## Conclusion

Despite numerous well-documented examples of functionally important newly minted genes arising from duplicate gene copies, a more comprehensive picture of the functional relevance and adaptive value of the large number of duplicate gene copies scattered throughout genomes is only now emerging. Only for a few whole-genome duplication (WGD) events in model organisms (most notably yeast) have global assessments of duplicate gene relevance for the emergence of new gene functions been attempted. WGD, on the other hand, is a special case of gene duplication that involves specific selective pressures related to the dosage balance of gene products that appear to have a significant influence on the fate of resulting gene duplicates. And even in the case of WGD, it remains largely unclear whether gene duplications often conferred novel functions or not.

## Acknowledgement

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## Conflict of Interest

There are no conflicts of interest by author.

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