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Author
Jani, Neel

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Junk DNA

Neel Jani

The DNA that composes humans is made of over 3 billion base pairs, yet close to 99% of these genes do not code for proteins and have been termed as "Junk DNA" (Wong et al., 2000), while a more appropriate name would be non-coding DNA (those that do no code for a specific protein), Junk DNA has stirred significant debate and research in the scientific community namely due to its enigmatic nature. Evolutionarily speaking, why should so much energy be wasted in the production of something of which only 1% will be functional? Growing research in the past couple of decades has thus tried shining light on Junk DNA, namely what it is, why it exists, its functionality, and its future in humans.

History of Junk DNA

The concretized term "Junk DNA" originated with researcher Susumu Ohno in the early 1970's, yet there has been an unintended misrepresentation from the media that the discovery of potential functions of noncoding DNA has only begun now. In fact, the discovery of the term went hand-in-hand with research as to the potential functions for what the term represented. Several researchers dismissed the idea that the vast majority of the genome is completely nonfunctional and were receptive to the idea of a function independent of coding proteins, such as regulation (Gregory and Palazzo, 2014). In fact, after each new type of non-protein coding DNA was discovered, the search for the potential function of these genes was sought. Among the major ones discovered after the 1960's were pseudogenes, transposable elements (TE's), and introns.

Figure 1. Evolutionarily speaking, why should so much energy be wasted in the production of something of which only 1% will be functional?

Types of Junk DNA and their Uses

Pseudogenes

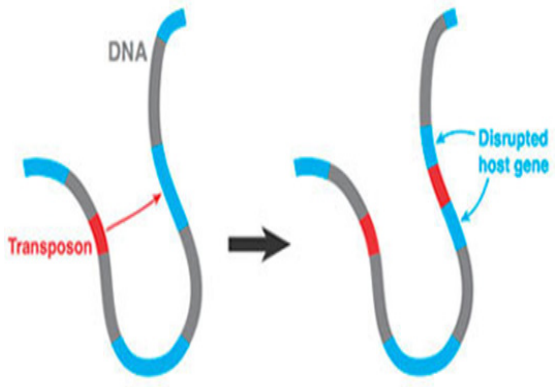
While the preservation of pseudogenes might occur because they induce no harm to the organism, there is growing evidence suggesting that they regulate prominent diseases such as diabetes and cancer.

When Susumu Ohno coined the term junk DNA, it was in reference to the development of pseudogenes. Specifically, Ohno described the phenomena of gene duplication; namely, how the cell of an organism could duplicate its genome, and have modifications and alternations to the newly formed copy rather than the original. Such a process would ensure that potential disastrous mutations to the copied genome could be masked by the original genome (Gregory and Palazzo, 2014). Those mutations that would be beneficial would help the organism survive. Overall, these gene duplication events seem to allow an organism to adapt adequately to potential environmental stressors. However, at times, the mutations undergone by the duplicate cause a non-functional protein to be coded. Hence, the mutation does not provide a substantial benefit or loss to the organism and is thus preserved as a pseudogene. The human genome is home to a large number of these pseudogenes that code non-functional proteins; researchers estimate that they number from 12,600 to 19,700 (Gregory and Palazzo, 2014).

While the preservation of pseudogenes might occur because they induce no harm to the organism, there is growing evidence suggesting that they regulate prominent diseases such as diabetes and cancer. This increase in research pointing out the functionality of pseudogenes has come from the use of next generation sequence technologies over commercial microarrays (Pink et al., 2013).

Transposable elements

Transposable elements were discovered by Barbara McClintock and were thrown under the same veil of Junk DNA as pseudogenes. Transposable elements are DNA Sequences that change their position in the genome. Transposable elements are founds in several groups and even kingdoms and are therefore "highly conserved among distantly related taxonomic groups, suggesting that they must be of some biological value to the genome" (Pray, 2008). With this in mind, specific functions of TEs have been explored. First, like pseudogenes, TEs have been found to assume regulatory functions with respect protein synthesis (Gregory and Palazzo, 2014). Second researchers Roy Britten and Eric Davidson have found that they might be involved in cell differentiation and the specification of the function of biological structures. This specialization is determined by the distribution of these TE's in a given stretch of DNA (Pray, 2008). Overall, however, the evidence for these functions has only been found for a small number of TEs, thus begging the question if these functions are unique to TE's or simply a subset of them (Gregory and Palazzo, 2014).



Transposable elements are DNA Sequences that change their position in the genome.

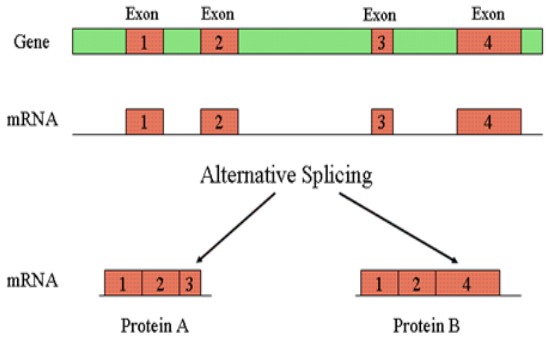
Introns

About 40% of the human genome is comprised of intronic regions, yet these intronic regions also contain pseudogenes and transposable elements (Gregory and Palazzo, 2014). Introns are portions of mRNA that can be removed prior to translation of the mRNA sequence through a process known as alternative splicing. Introns have been found to increase the possibilities of protein products due to alternative splicing, thus increasing diversity and potential adaptations to environmental pressures from one strand of mRNA. Furthermore, researchers have found that introns that are removed from the larger segment of RNA can express themselves later on (Carmel and Chorev, 2012). Thus there, is also the possibility of expression of an intron once it is excised.

Is Junk DNA actually Junk?

While there are several types of Junk DNA and each of them appear to have some use, several arguments suggest that Junk DNA is, in fact, junk. The first is what is termed as the Onion Test. Specifically, organisms with significantly more DNA content than humans, such as the onion whose genome is five times as large as that of a human, might appear to have no real reasons to carry this much more DNA (Gregory and Palazzo, 2014). The complexity of a human and the greater number of metabolic processes required suggest that there is DNA in the onion that is simply not used, though recent attempts have been made to show that it is necessary (Freeling, Xu, & Woodhouse 2015). The arbitrariness of the relationship between the physical amount of DNA and complexity is further highlighted by the fact that "salamander species belonging to Plethodon boast a fourfold range" within the species itself, suggesting that there is DNA that is inherently useless to the organism's function (Doolittle, 2013).

Second, as mentioned earlier with respect to pseudogenes, a variety of evolutionary processes shape the structure of non-coding DNA. Functionally important regulatory sequences will tend to be conserved as a result of negative selection against harmful mutations, whereas positive selection will favor those that benefit the population. However, "a central tenet of the nearly neutral theory of molecular evolution is that extraneous DNA sequences can be present within genomes, provided that they do not significantly impact the fitness of the organism" (Ludwig, 2002). Thus, these mutations in a genome have to be significantly negative in order for these genes to be eliminated from a population.



Introns have been found to increase the possibilities of protein products due to alternative splicing.

Researchers have also found through the evolution of Archaea that there are selective pressures to get rid of junk DNA in them. In fact, the percentage of non-coding DNA with respect to coding elements in the genome of an Archae has been found to have 6-14%, which is significantly less than the amount in humans (Tatusov, Wolf, & Koonin, 2002). Moreover, after having traced the percentage of coding elements in Archae throughout time, and researchers found that "the evolution of non-coding regions appears to be determined primarily by the selective pressure to minimize the amount of non-functional DNA, while maintaining essential regulatory signals" (Tatusov, Wolf, & Koonin, 2002). Essentially, the minimum amount of DNA for regulating transcription is preserved but the rest, over time, will be removed from the population. These ideas have also been proven empirically in eukaryotes, where "a general mutational tendency towards DNA loss… inescapably influence[s] the length of noncoding regions in most eukaryotes" (Comeron 2001).

Conclusion

Junk DNA continues to baffle researchers to this day, and on-going research hopes to demystify the functionality associated with non-coding genes. While most of non-coding DNA that has some function appears to be regulatory, both in terms of protein and larger biological structures, and some possible functions in combating disease, a large percentage of non-coding DNA eludes researchers with regards to function. Moreover, the historic nature of a reduced genome size and the lack of correlation between genome size and complexity suggest that even if some non-coding sequences might have some regulatory function, they are also several sequences that are essentially expendable.

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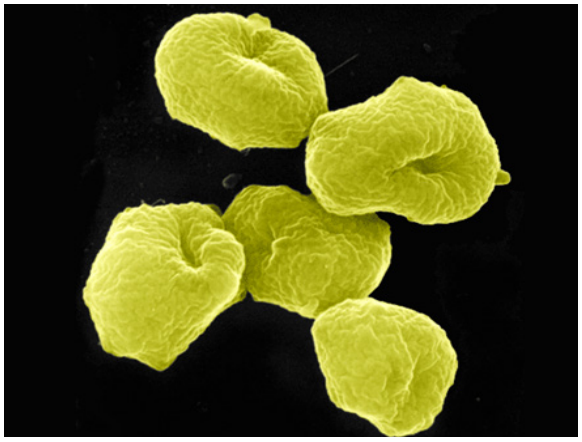

Figure 2. Researchers have also found through the evolution of Archaea that there are selective pressures to get rid of their junk DNA.

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