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Type	Oral communication
Session	Biological Databases
Title	VarNuCopy: from Copy Number Variations to longevity and cancer species predisposition
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Motivation

Copy number alterations (CNVs) often result in gains or losses in copies of genes contained in human cell DNA, and are prevalent in cancer cells. Furthermore, the variation in CNVs has recently been correlated with longevity. We create a database containing all the eukaryotic species currently sequenced, unique of its kind, which collects the CNVs landscape for each organism. Relying on the comparison of gene copy numbers across the genomes of multiple organisms, we developed VarNuCopy, an open-source web tool, able to guide researchers through the investigation of known and/or unknown molecular mechanisms related to cancer and longevity pathways. VarNuCopy is the first tool allowing researchers to compare multiple CNVs landscape from different species, and to identify those genes that appear to be linked to the genome instability of an organism.

Methods

Following the idea that copy number variation of important genes can theoretically protect a species from cancer insurgence, the platform allows the exploration of complex data-sets to assess copy number variation of any given gene throughout the referenced species. Moreover, by performing a basic query in the Genes Exploration section, researchers investigating particular gene copies gain/loss can easily retrieve and analyze the difference in copy number in an inter-species comparison approach. Finally, VarNuCopy Descriptive Analysis Models (DAMs), which are exclusively built using both the species cancer incidence and the genomic copy number variation of a subset of 25 mammals, can help scientists to statistically discriminate between cancer–prone and cancer–resistant organisms, and eventually to discover new possible genetic mechanisms involved in oncogenesis related pathways.

Results

VarNuCopy is a database of genome Copy Number Variations (CNVs) across the animal kingdom. We recovered and organized the most significant data using graphical representations, in which CNVs trends inside a species group of particular interest are reported, coupled with the statistical validation of the related target genes or species. We created four Descriptive Analysis Models able to correlate the copy-number variation of genes, and in particular of tumor suppressors and oncogenes of 25 mammals, with important phenotypic characteristics, trying to link the specific gene number alteration with the percentage of cancer incidence. The model provides a statistical description of the genes copy number distribution within two different target groups: cancer –prone and –resistant species. In order to test the hypothesis that genomic CNVs are related to the cancer incidence of a species, we applied our DAM model to retrieve new target genes able to discriminate between cancer resistant and cancer prone organisms. According to our bioinformatics analysis, which only considers the variation in number of gene copies within different species, VarNuCopy is able to confirm that a gene can be (possibly) involved in biological processes of cancer onset. Furthermore, the results of both the Gene Ontology and the Over-Representation Analysis (ORA), performed on the significant target genes resulting from our DAMs, showed an enrichment in those pathways and biological processes related to cancer onset.

Info

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Figure

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Availability	-

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