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Analysis of Intellectual Disability in Children with Genomic Hybridization

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INTRODUCTION

Bacterial pan-genomes obtained by Whole-Genome Sequencing (WGS) can be used to cluster DNA-coding sequences (CDS) into Pan-genome Orthologous Groups (POGs). Our aim was to examine the comparative genomic function of Streptococcus canis based on POG analysis and identify CDSs specific to common sequence type (ST). Twenty WGS datasets of Streptococcus canis strains (including invasive and non-invasive specimens) were obtained from the Centre for Biotechnology Information Assembly's National Database. Based on the WGS data, we performed Comparative Genomic Hybridization (CGH), pan and core genome predictions, Venn diagram tests using five ST9 strains and phylogenetic analysis by ST determination. We compared the CDS of 7 ST9 strains and 13 non-ST9 strains. We observed genomic diversity based on CGH and Venn diagram analyses. The predicted pan and nuclear genomes contained 4,772 and 1,403 genes, respectively. Based on the phylogenetic tree, five clades composed of different STs were found. There were differences in the four signalling pathways regulated by ST9-specific CDS (DNA restriction-modification system, DNA-mediated translocation, extracellular domain, and response to oxidative stress). Our results describe genomic diversity in CGH and Venn diagram tests, pan and core genomes, five genomic clades composed of distinct STs, and a unique CDS trait associated with ST9.

DESCRIPTION

Hybridization plays a central role in many fundamental evolutionary processes such as speciation and adaptation. However, despite its central importance in evolution, little is known about the actual prevalence and distribution of current hybridization in the tree of life. Here we develop and implement a new statistical method that can detect F1 hybrids from single-genome sequencing data. Using simulations and sequencing data of known hybrid systems, we first demonstrate the specificity of the method and identify its statistical limitations. We then present the method by applying it to available sequence data from over 1,500 species of arthropods, including *Hymenoptera*, *Hemiptera*, *Coleoptera*, *Diptera*, and *Archnida*. Among these taxa, *Hymenoptera*, especially ants, had the highest number of F1 hybrid candidates, suggesting high rates of hybridization between previously isolated gene pools in these groups. The prevalence of F1 hybrids is unevenly distributed across ants, with taxa containing many candidates tending to harbor specific ecological and life history characteristics. This work demonstrates how a large-scale comparative genomic study of recent hybridizations can be performed to reveal determinants of first-generation hybridizations across tax [1-4].

CONCLUSION

Developmental delay (DD) is a condition in which developmental milestones and learning abilities do not occur within the expected age range for patients younger than 5 years of age. Intellectual Disability (ID) is characterized by impaired or inappropriate development of intellectual abilities, including impairment of intellectual functions such as learning and causality. DD/ID cases with isolated syndromes show extreme genetic heterogeneity. Array-based comparative genomic hybridization (aCGH) can detect genome-wide copy number variations (CNVs) with higher resolution than traditional cytogenetic methods. The purpose of this study was to discuss the clinical findings and aCGH analysis results of solitary and symptomatic DD/ID cases in genotype-phenotype correlations. The diagnostic rate was 17.1 when evaluating all pathogenic and probable pathogenic cases. Using aCGH analysis as a first-line test for DD/ID cases significantly contributes to diagnostic yield and enables detection of rare microdeletion/micro-duplication syndromes. A clear determination of genetic etiology contributes to the literature on genotype-phenotype correlations.

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CONFLICTS OF INTERESTS

The authors declare that they have no conflict of interest.

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