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Single-Cell Genomic and Transcriptome Contains Primary and Metastatic Colorectal Cancer Tumors

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INTRODUCTION

Signaling and activation of transcription 5 (STAT5) is a key transcription factor that regulates a variety of biological processes in mammalian development. Aberrant regulation of STAT5 is also associated with many diseases, including cancer and immune-related diseases. Sustained activation of STAT5 through dysregulation of signaling cascades has been reported to be associated with solid tumor progression and leukemia, while different genomic mutations in STAT5 have been shown to cause different diseases. I'm here. This review comprehensively summarizes the unique functions of STAT5 and the results of recent studies examining the association of STAT5 mutations with human disease. This review also describes the types of disease models that are useful for studying the mechanisms underlying STAT5-induced disease progression. These results provide fundamental knowledge for understanding the regulatory mechanisms of STAT5 and the progression of various diseases resulting from aberrant STAT5 regulation. Furthermore, this review may provide the insights needed to create optimal disease models that reflect the STAT5 mutations associated with human disease and to develop gene therapies that correct STAT5 mutations.

DESCRIPTION

Although the functions of the tumor microenvironment (TME) are coordinated by the precise spatial organization of specialized cells, little is known about the multicellular structures that form within the TME. Here, we systematically mapped TME structures *in situ* using imaging mass cytometry and multi-slice spatial analysis of 693 breast tumors in combination with genomic and clinical data. We identified 10 recurrent TME structures that differed by vessel content, interstitial quiescence and activation, and leukocyte composition. These TME structures exhibited distinct accumulation patterns across breast cancer subtypes, some associated with genomic profiles indicative of immune escape. Regulatory and dysfunctional T cells occurred simultaneously in large 'suppressed expansion' structures. These structures, characterized by high cellular diversity, cell proliferation and enrichment of BRCA1 and CASP8 mutations, predicted poor prognosis in estrogen receptor-positive disease. The multicellular structures disclosed here combine conserved spatial organization and local TME function, potentially improving patient stratification.

CONCLUSION

Since the first half of the 20th century, evolutionary theory has been dominated by the idea that mutations are random in their outcome. Here, we test this assumption in a large-scale survey of *de novo* mutations in the plant Arabidopsis. Unexpectedly, we found that mutations occur less frequently in functionally restricted regions of the genome. The frequency of mutations is reduced by half within the gene body and by two-thirds within essential genes. Using independent genomic mutation datasets, including the largest *Arabidopsis thaliana* mutation accumulation experiment performed to date, we found that epigenomic and physical properties correlated with the variance of genome-wide patterns of perigene mutational biases. It indicates to explain more. Second, the observed mutation frequencies around the gene accurately predict the pattern of genetic polymorphism in natural *Arabidopsis thaliana* accessions. Analysis of allele frequencies supports that mutational bias is the main force behind the pattern of sequence evolution around genes in the natural lineage. Finally, we find that genes that undergo more purifying selection have lower mutation rates. We conclude that epigenome-associated mutational bias 2 reduces the occurrence of deleterious mutations in Arabidopsis, challenging the common paradigm that mutation is a directionless force in evolution.

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