



Scanning the human genome at kilobase resolution

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ABSTRACT:

Increasing evidence shows that the genome structure is highly variable within the normal human population, and the genome structure in pathological situations, such as in cancer, is also highly altered, reflecting the heterogeneous and progressive nature of disease cells. Systematic analysis of normal genome variation will provide fundamental knowledge to improve understanding of the genetic basis for normal human diversity; comprehensive characterization of pathological genome alterations will identify genetic factors which contribute to particular diseases, which could become diagnostic markers and therapeutic targets. Structural genome changes frequently happen at kilobase to sub-kilobase levels. However, detecting changes at this level remains a technical challenge. My laboratory has developed a new technology termed the DGS (Ditag Genome Scanning) that aims to analyze the genome structure at kilobase resolution. We have performed extensive computational analysis and experimental evaluation of the technology. Our study indicates that the DGS technology provides a powerful tool for studying human genome structure.