WHITEPAPER

Biocare Basics: Inversion Mutations

BIOCARE MEDICAL

Biocare Basics: Inversion Mutations

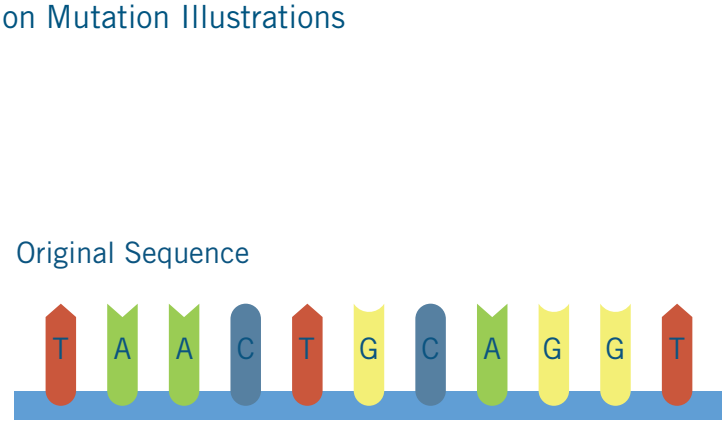
Over the past few decades, genomic instability, or rearrangement of the genome inside a cell, has become a significant focus of diagnostic and therapeutic applications in various disease states.[2] Genomic instability describes gene mutations such as translocation mutations, copy number alterations, deletion mutations, point mutations, and inversion mutations.[2]

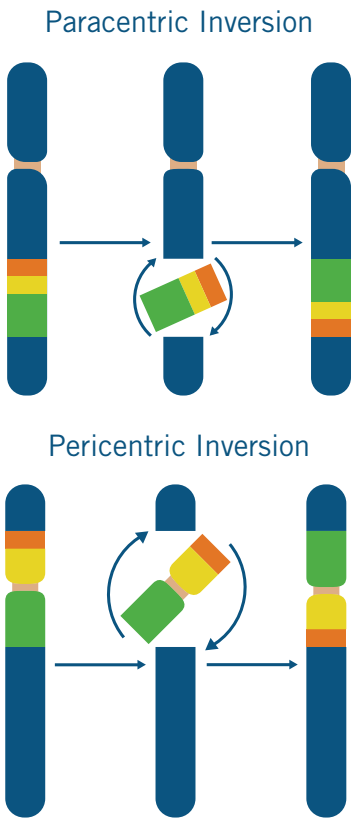
Inversion mutations occur when a segment of DNA within a chromosome changes orientation without losing or gaining genetic material.[1,2] These mutations have been observed in both plants and animals, where evidence suggests they have driven genetic diversity and evolutionary changes, given that inversion polymorphisms show geographic, latitudinal, and seasonal variation in frequency.[1] Some examples of phenotypic expression of inversion mutations include differences in size, color, flowering time, freshwater adaptation, and fertility.[1,3] However, not all inversion mutations are benign.

Although many human chromosomal inversions do not appear to have clinical significance, some inversions have been shown to disrupt or alter critical gene expression, causing heritable disorders such as Hemophilia or Hunter Syndrome.[3] Others have been associated with neurodegenerative diseases, such as Alzheimer's Disease and Parkinson's Disease. In contrast, other inversions are not known to carry direct consequences on their own but appear to predispose carriers to other disease-causing mutations in the same genomic region.[3] For example, the deletion mutation responsible for Williams-Beuren Syndrome is strongly correlated with the presence of otherwise-benign inversion mutations in the same region.[3] Similarly, the deletion mutations responsible for Angelman Syndrome have been linked to inversions in the mothers of patients.[3]

Human inversions have been detected and studied via molecular testing such as G-banding, southern blot hybridization, or fluorescence in situ hybridization (FISH).[3,4] FISH has proved to be a powerful means of locating genes and detecting chromosomal abnormalities.[4]

Inversion Mutation Illustrations





Biocare has recently acquired Empire Genomics. To learn more about FISH probe offerings, please visit biocare.net or empiregenomics.com

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4. Salomon-Nguyen, F., Le Coniat-Busson, M., Heilig, R., Campion, D., Weissenbach, J., & Berger, R. (1998). Evidence of chromosomal inversion using fluorescence in situ hybridization to stretched DNA. Comptes rendus de l'Academie des sciences. Serie III, Sciences de la vie, 321(6), 447–452. https://doi.org/10.1016/s0764-4469(98)80775-0