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Inferring the function of genes based on recurrent mutations in protein domains: Analysis of OncoMD data

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Abstract

Protein domains are conserved structural and functional units of proteins. We have used OncoMD data to analyse recurrent mutations in protein domains and their functional impact. In this study, we systematically analysed tumour samples from 28 different cancer groups to identify recurrent mutations, their positions within specific domains to identify domains that harbour recurrent mutations in different cancers. Next, we mapped the mutations to protein domains present in oncogenes and tumour suppressor genes and identified a variety of domains that are enriched for mutations. Whereas kinase domain and p53 superfamily domains are significantly mutated across many cancers, few cancers, such as melanoma, brain and colorectal cancers are significantly mutated in 7-transmembrane domain and Ig superfamily domain proteins. Highly mutated protein domains such as the PKc and PI3K superfamily are targets of anti-cancer drugs. Inferring the functional impact of recurrent mutations in cancer is an important objective of cancer genomics. Analysis of mutation hotspots in protein domains and its functional impact will provide novel insights into disease mechanisms and breakthrough therapeutics for treatment.

References

[1] Miller, M.L., Reznik, E., Gauthier, N.P., Aksoy, B.A., Korkut, A., Gao, J., Ciriello, G., Schultz, N. and Sander, C. (2015) Pan-Cancer Analysis of Mutation Hotspots in Protein Domains. *Cell systems* 1: 197-209. https://doi.org/10.1016/j.cels.2015.08.014

[2] Baeissa, H., Benstead-Hume, G., Richardson, C.J. and Pearl, F.M.G. (2017) Identification and analysis of mutational hotspots in oncogenes and tumour suppressors. *Oncotarget* 8: 21290-21304. https://doi.org/10.18632/oncotarget.15514

[3] Peterson, T.A., Park, D. and Kann, M.G. (2013) Domain landscapes of somatic mutations in cancer. *AMIA Jt Summits Transl Sci Proc* 136.

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