Università degli Studi di Pavia

Dottorato di Ricerca in Bioingegneria e Bioinformatica

A Protein-Domain Approach for the Analysis of Disease Mutations

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Aula seminari
(ex-dipartimento di Informatica e Sistemistica)

Identifying the functional context for key molecular disruptions in complex diseases is a major goal of modern medicine that will lead to earlier diagnosis and more effective personalized therapies. Most available resources for visualization and analysis of disease mutations center on gene analysis and do not leverage information about the functional context of the mutation. I will describe a resource for domain mapping of disease mutations, DMDM, a protein domain database developed by our group in which each disease mutation is aggregated and displayed by its protein domain location. We have also developed a methodology using domain significance scores (DS- Scores) to detect statistically significant disease mutation clusters at the protein domain level. When we applied the DS-Scores to human data, we identified domain hotspots in oncogenes, tumor suppressors, as well as in genes associated with Mendelian diseases. In addition, I will describe recent work on analyzing cancer somatic mutations from individual cancer patient genomes. We found that incorporating information about classification of proteins and protein sites leads to new hypotheses regarding the role of tumor somatic mutations in cancer.

I dottorandi e gli interessati sono caldamente invitati a partecipare

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