PublisherInfo		
PublisherName	:	BioMed Central
PublisherLocation	:	London
PublisherImprintName	:	BioMed Central

From genes to disease

ArticleInfo		
ArticleID	:	4480
ArticleDOI	:	10.1186/gb-spotlight-20020515-01
ArticleCitationID	:	spotlight-20020515-01
ArticleSequenceNumber	:	146
ArticleCategory	:	Research news
ArticleFirstPage	:	1
ArticleLastPage	:	1
ArticleHistory	:	RegistrationDate : 2002–5–15 OnlineDate : 2002–5–15
ArticleCopyright	:	BioMed Central Ltd2002
ArticleGrants	:	
ArticleContext	:	130593311

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Only a small fraction of the thousands of described genetically inherited diseases have been linked to a specific gene. In an Advanced Online Publication in Nature Genetics Carolina Perez-Iratxeta and colleagues at the EMBL in Heidelberg describe using a bioinformatics approach to link genes to diseases (13 May 2002, DOI:10.1038/ng895). Their data-mining system is based on 'fuzzy set theory', which can make inferences from the complex scientific literature. They integrated information from multiple databases to establish relationships between Medical Subject Headings (MeSH terms) related to diseases, or drugs, and Gene Ontology terms. After a series of computational steps they defined a 'core' for known genes in the RefSeq database. They then used the score to rank candidate genes in a given disease-associated region. When this approach was tested against known disease-linked genes, the score could predict promising candidate genes. This type of strategy may be useful for prioritising disease-associated candidates at a given disease locus.

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