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# Community-driven comprehensive representation of disease mechanisms

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## Abstract

### Motivation

Despite the availability of various network-based approaches, tools for disease-specific functional analysis of high-throughput data are greatly underdeveloped. The direct approach to solve this problem is developing comprehensive representations of disease mechanisms (Fujita et al., 2013, PMID 23832570; Kuperstein et al., 2015, PMID 26192618).

### Methods

Disease-specific representations are developed in CellDesigner ([www.celldesigner.org](http://www.celldesigner.org)) following the Systems Biology Graphical Notation standard ([www.sbgn.org](http://www.sbgn.org)). The involvement of domain experts ensures that all the disease hallmarks are covered and adequately represented.

### Results

We introduce a concept of the disease maps as a community effort and as a collection of reference resources for making sense of omics data in studies focused on a particular disease. Essentially a disease map provides a consensus review on the known disease mechanisms in the format of interconnected metabolic, signalling and gene regulatory pathways, and can be

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efficiently used as the basis for hypothesis generation. While being complementary to generic solutions such as the Ingenuity Pathway Analysis and MetaCore, the disease maps focus on a single hierarchically-organised network. We describe our experience in developing disease maps for Parkinson's disease, asthma and cancer, and demonstrate how these resources can be applied for data visualisation and interpretation.

## **Conclusion**

As a continuously evolving community effort the Disease Maps Project will facilitate powerful advances in systems medicine for understanding disease mechanisms, cross-disease comparison, finding disease comorbidities, suggesting drug repositioning, generating new hypotheses, and after careful validation, redefining disease ontologies based on molecular mechanisms involved.

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