

Graphical Visualization and Navigation of Genetic Disease Information

Olivier Bodenreider^a, M.D., Ph.D., Joyce A. Mitchell^{a,b}, Ph.D.

^a U.S. National Library of Medicine, NLM, DHHS, Bethesda, Maryland

^b University of Missouri, Columbia

One of the goals of biomedical scientists in this exciting era of molecular biology is the discovery of novel genes and their relationship to the molecular basis of disease. Part of this information connecting the genotype to the phenotype is already known and available through hubs such as LocusLink¹. Although LocusLink is a valuable resource that organizes curated information around genes and provides links to other online resources, it has not been developed for users to visualize graphically the association between genes and diseases, nor to navigate easily from genotype to phenotype (and back) within the same application.

The application we developed², *g2p*, aims at visualizing graphically and navigating genetic disease information, especially the link between genotype and phenotype. The information displayed comes from a LocusLink query on human genes associated with a known disease (1330 genotypes, 1835 phenotypes, and 2050 associations). This application is based, in part, on the graph visualization package GraphViz³.

¹ <http://www.ncbi.nlm.nih.gov/LocusLink/>

² <http://etbsun2:8000/perl/g2p.pl>

³ www.research.att.com/sw/tools/graphviz/

Starting from a disease query, *g2p* displays the phenotype view (Figure 1). Alternatively, from a gene query, it creates a genotype view (Figure 2).

In the example here, a search on the disease *Bladder cancer* leads to several genes, including *RB1* (Figure 1). The double frame around *RB1* indicates that it is linked to more than one disease and thus is “navigable”. Following this link leads to the several diseases associated with *RB1*, including – besides *Bladder cancer* – *Retinoblastoma* and *Osteosarcoma* (Figure 2). The latter disease also has a double frame, indicating that several genes are associated with it.

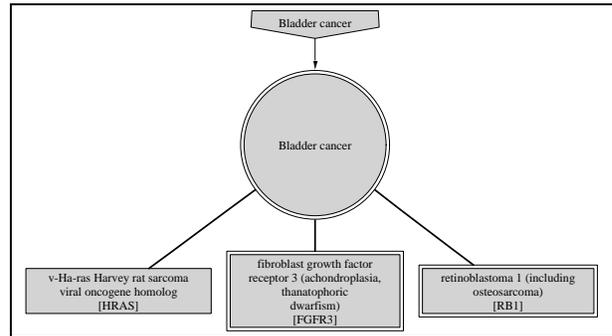


Figure 1 – Phenotype view for *Bladder cancer*

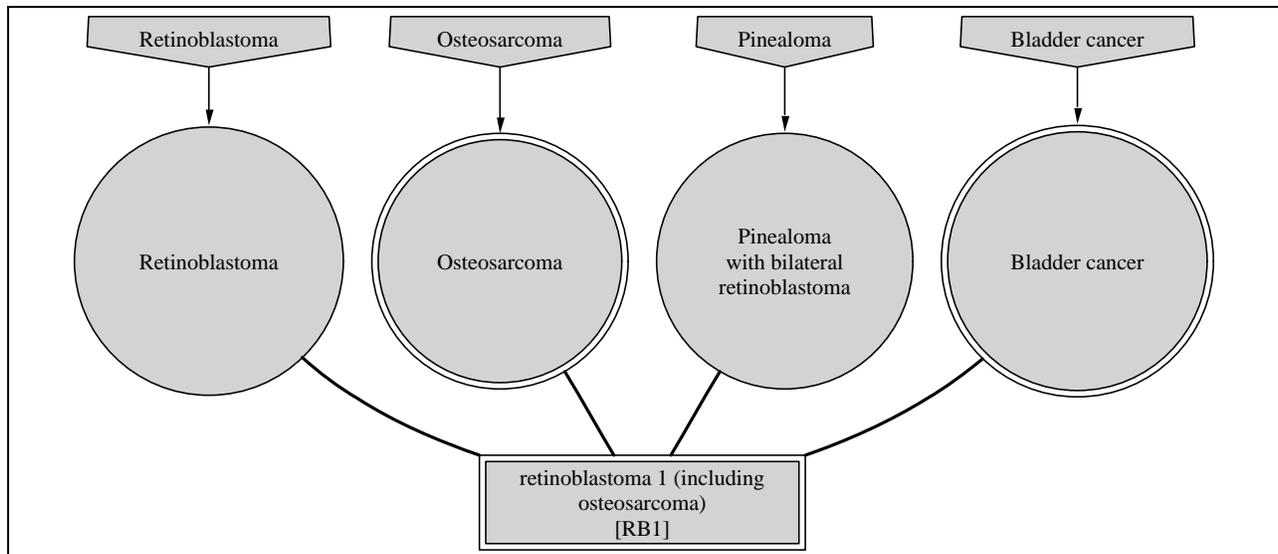


Figure 2 – Genotype view for *RB1*