

Paper: “Extending inherited metabolic disorder diagnostics with biomarker interaction visualizations”

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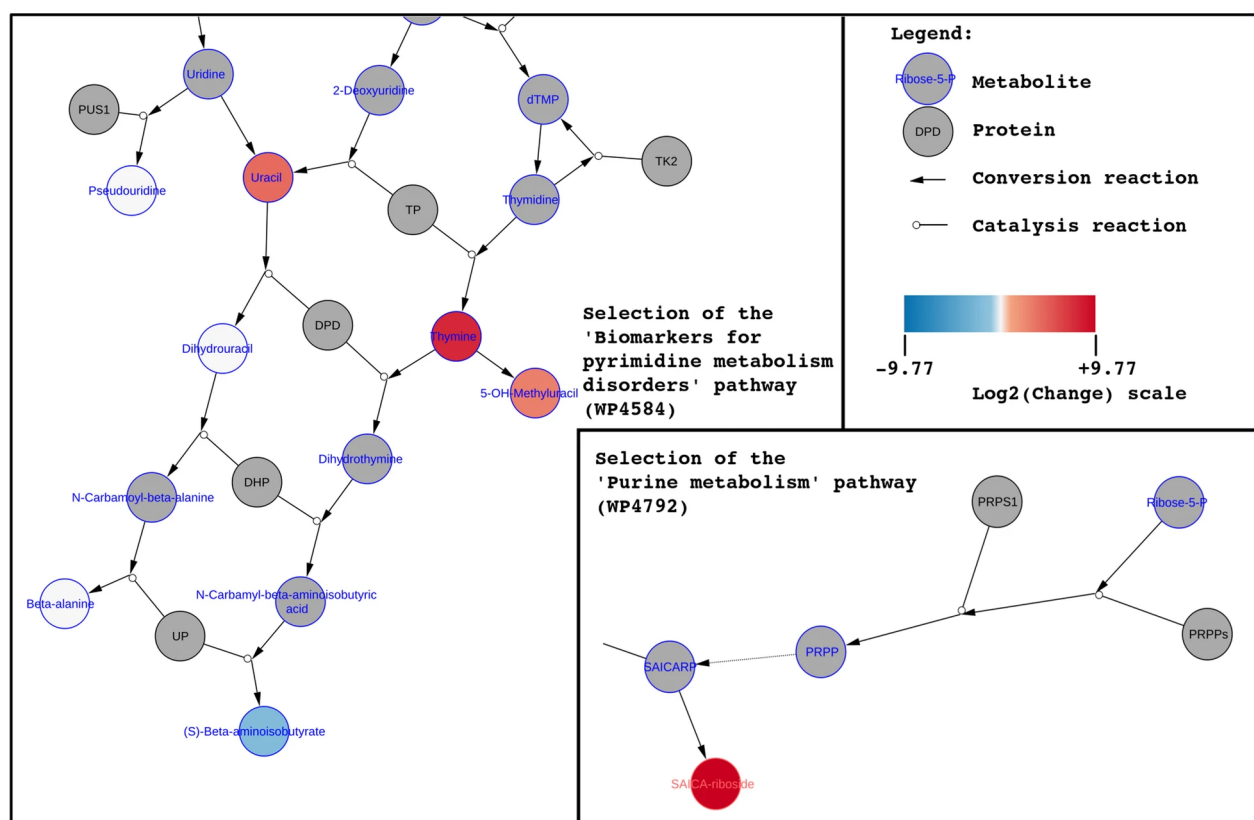
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When I joined the BiGCaT research group in 2012 I was particularly interested in the open science approach of WikiPathways. As a chemist by training and researcher in cheminformatics, metabolites and their metabolic reactions took my particular interest. I am happy that I have been able to fund Denise's research project. And thanks Denise for this very exciting research. I know it's just a first step, and far more translational steps are needed, but I for one am very exciting to bridge molecular info to clinical outcomes.

In this study, Denise explored how we can take advantage from molecular pathway databases to link biomarker information: *"Our framework integrates literature and expert knowledge into machine-readable pathway models, including relevant urine biomarkers and their interactions. The clinical data of 16 previously diagnosed patients with various pyrimidine and urea cycle disorders were visualized on the top 3 relevant pathways. Two expert laboratory scientists evaluated the resulting visualizations to derive a diagnosis"* (doi:[10.1186/s13023-023-02683-9](https://doi.org/10.1186/s13023-023-02683-9)).

Figure 4 shows how such a visualization of those biomarker interactions can look like:



And I am hugely proud of the open science approach, [from GitHub repo](#), open source R code, SPARQL queries. Thank you, Denise! And thanks to [Dr Laura Steinbusch](#) for this nice collaboration! Further acks in the article.