Towards integrative family analysis on OMICs data for individual patient diagnostics

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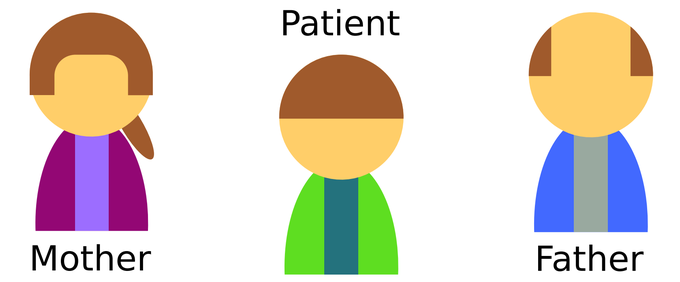
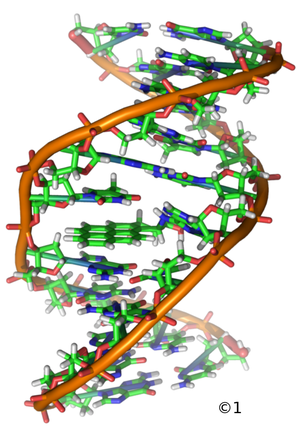
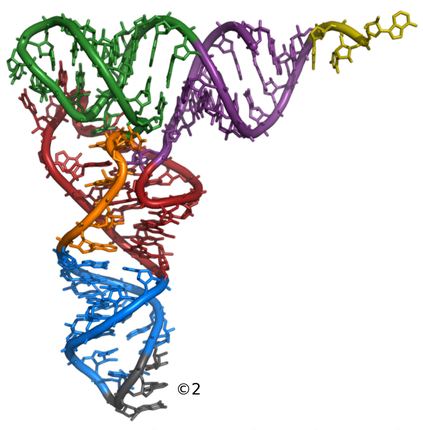
Introduction

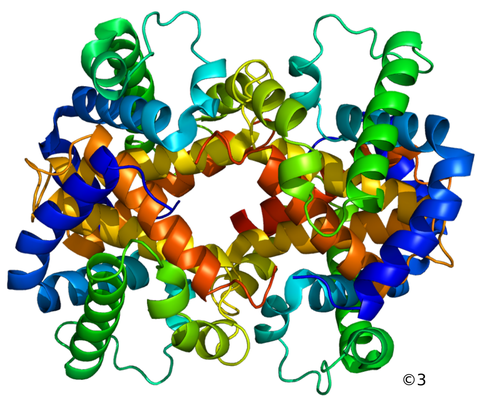
During recent years generating OMICs data became cheaper and faster. Notably DNA sequencing benefited from this change, becoming more popular and frequently used, especially in the domain of diagnostics. As became apparent, the data from DNA sequencing does often not allow for conclusive diagnostics, as it only shows one aspect of what contributes to the patients phenotype. The combination of OMICs data, like genomics, transcriptomics and proteomics, presents itself as a solution to this problem, especially in the context of family analysis which helps to identify interesting features.

While many methods have been developed combining different OMICs sources to create more complete and comprehensive analyses of a patients genotype, doing family analysis in the context of OMICs data with an user friendly visualization of the data is still explored very little. We present our vision of an easy to use framework for combined OMICs analysis, decreasing the complexity of such analyses while increasing the speed at which they can be performed.



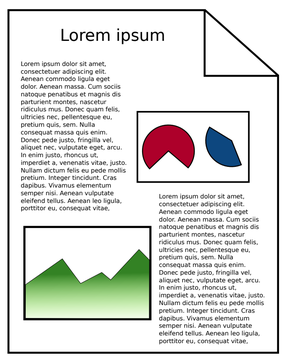

Genomics (DNA) ©1


Transcriptomics (RNA) ©2


Proteomics (Protein) ©3

Vision

Our vision so to create a comprehensive, user friendly application which concentrates on several key features of OMICs data for comparative analysis. The vision concentrates on family analysis of three key features. Individual SNP comparison of genomic data, gene expression comparison with transcriptomic data and protein variation from proteomics data. The goal is to increase the accuracy of all those OMICs datasources throught their combination, allowing for findings otherwise hard or impossible to make (Low et al., 2013). One example of the usefulness of this approach is the study of genetic disorders in myopathies, where we recently showed some progress regarding functional understanding and therapy (Walter et al., 2013)



Report

Methods

The application will be based on GensearchNGS, an existing application that provides the required tools for genomics analysis, allowing the user to visualize the analysis and underlying data. For genomics and transcriptomics we will focus on DNAseq and RNAseq, allowing for a shared infrastructure between the two data types. GensearchNGS will be gradually improved to include, in addition to DNAseq data, RNAseq data and mass spectrometry for proteomics. The focus will lie in the integration of different OMICs data sources and familly analysis on the discovered features. The framework will be developed in a modular and extensible way, allowing to offload computer heavy calculations to a remote grid or cloud, to speed up the analysis. While the usage of remote resources will be optional, research will be done on the issues of privacy of medical data in the cloud.

Conclusion

While many challenges still lie ahead to combine different sources of OMICs data in a user friendly and meaningful way to support scientists in research and diagnostics questions, the field progressed a lot during recent years. The increasing maturity of the technologies used for genomics, transcriptomics and proteomics allowed to increase the accuracy of the analysis by combining the data of all three source. We intend to lower the complexity of this analysis by integrating those different datasources through a comprehensive user interface and workflow, while increasing the accuracy of analyses of genetic disorders like myopathies (Bach et al., 2014), Hereditary hearing loss (Rost et al., 2013) or muscular dystrophies (Bach et al., 2013) already being analyzed with GensearchNGS using DNAseq.

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