

Presentation notes

### **Classifying metabolic disorders using supervised machine learning**

Baumgartner, C. et al. Supervised machine learning techniques for the classification of metabolic disorders in newborns. *Bioinformatics* 2004 20: 2985-2996. PMID: 15180934.

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The great thing about artificial learning techniques is that we don't really know what techniques will become the dominant techniques of the future. This study used a number of common techniques from the WEKA and see how well they worked in a real life metabolic disorders screening. Focusing on two representative inborn errors of metabolism, phenylketonuria (PKU), an amino acid disorder, and mediumchain acyl-CoA dehydrogenase deficiency (MCADD), a fatty acid oxidation defect.

For the most part the article did a good scientific job of testing the techniques. However, there were a few problems. The main problem was that the top three techniques all produced good results in the screening. Although logistic regression analysis (sensitivity >96.8%, specificity >99.98%) performed the best, it did not have a statistically significant advantage over the other techniques that ranked in the top three. In truth the study really proved only that given the right conditions that the machine learning techniques of today could produce clinical significant results.

The second major attack on the article was its use of back box modeling. Although it was nice to have the comparative results of the WEKA, We were unclear if it really made sense to use a prefabricated machine learning techniques. In the real world we hardly believed the way that this informatics problem would be resolved is running a bunch of algorithms from a black box and using the one that fit best. In truth, custom problems require custom solutions and careful selection of the best algorithm modified to the

specific circumstances. Custom algorithms seemed like the way that most people would address an informatics problem like this one.

Finally there weren't very many wider lessons that could be taken from this study. Although conducted well, it seemed to us this study indicated a specific task at a specific time. This fact made it more an evaluation of the WEKA algorithms specified and less of an overall discovery about artificial learning in bio-informatics.

All in all, this was good article describing a case controlled random test of bio-informatics, it proved some good insight into the challenges of machine learning and more specially testing. Although black box tests have a nice comparative aspect the selection of random tools is not really the best technique in solving a problem, a point that should be addressed in the article.