# Identifying cell types associated with disease

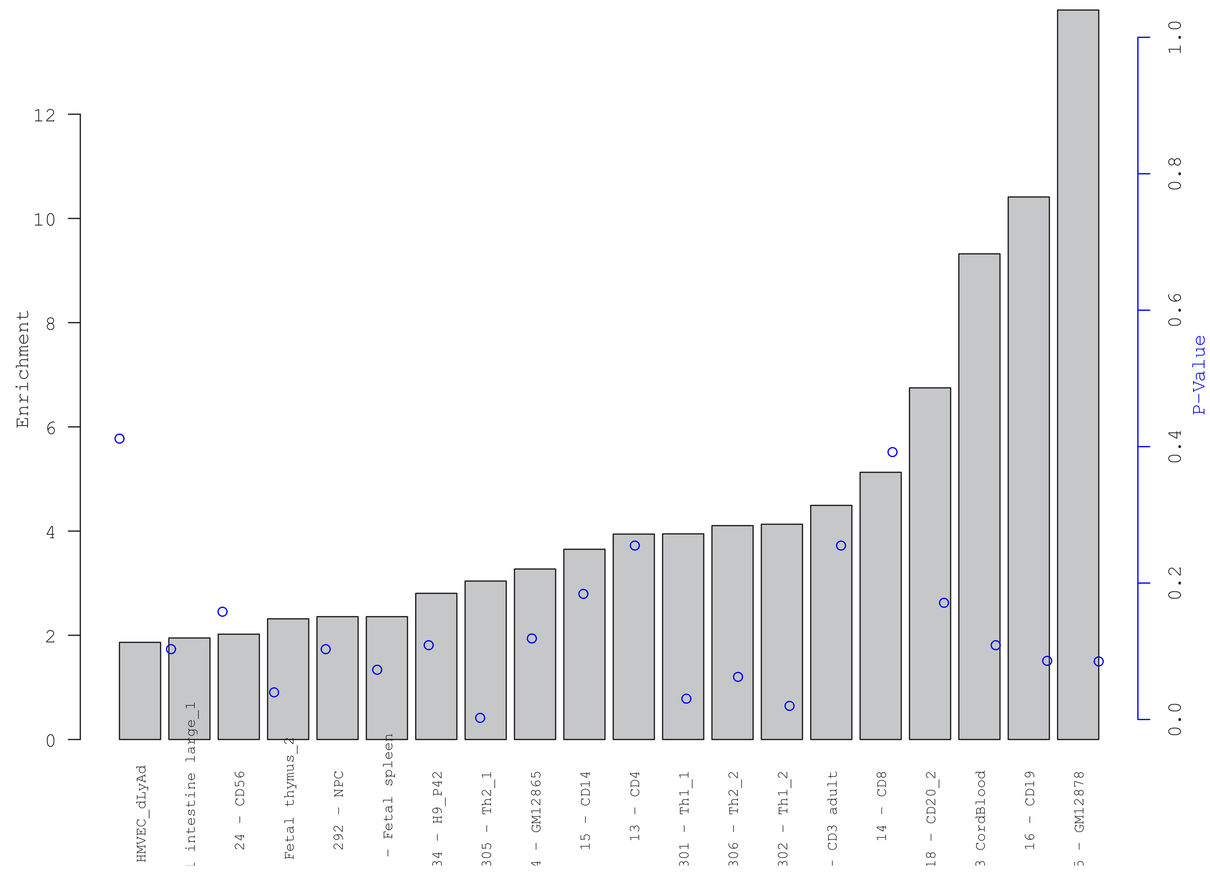
Genome-scale data on histone modifications and DNase I accessibility, such as that produced through the ENCODE and Roadmap Epigenomics projects, may be used to explore cell type-specificity of noncoding variants. We here present a tool that allows a user-provided genomic variation track to be compared against an integrated collection of cell-specific reference tracks. Using a similar methodology, the tool also allows a genomic variation track to be compared against an integrated track collection of binding sites for various transcription factors.

Results are presented in the form of graphs and sortable tables. Details underlying the results can be inspected by following links directly from the main results tables.

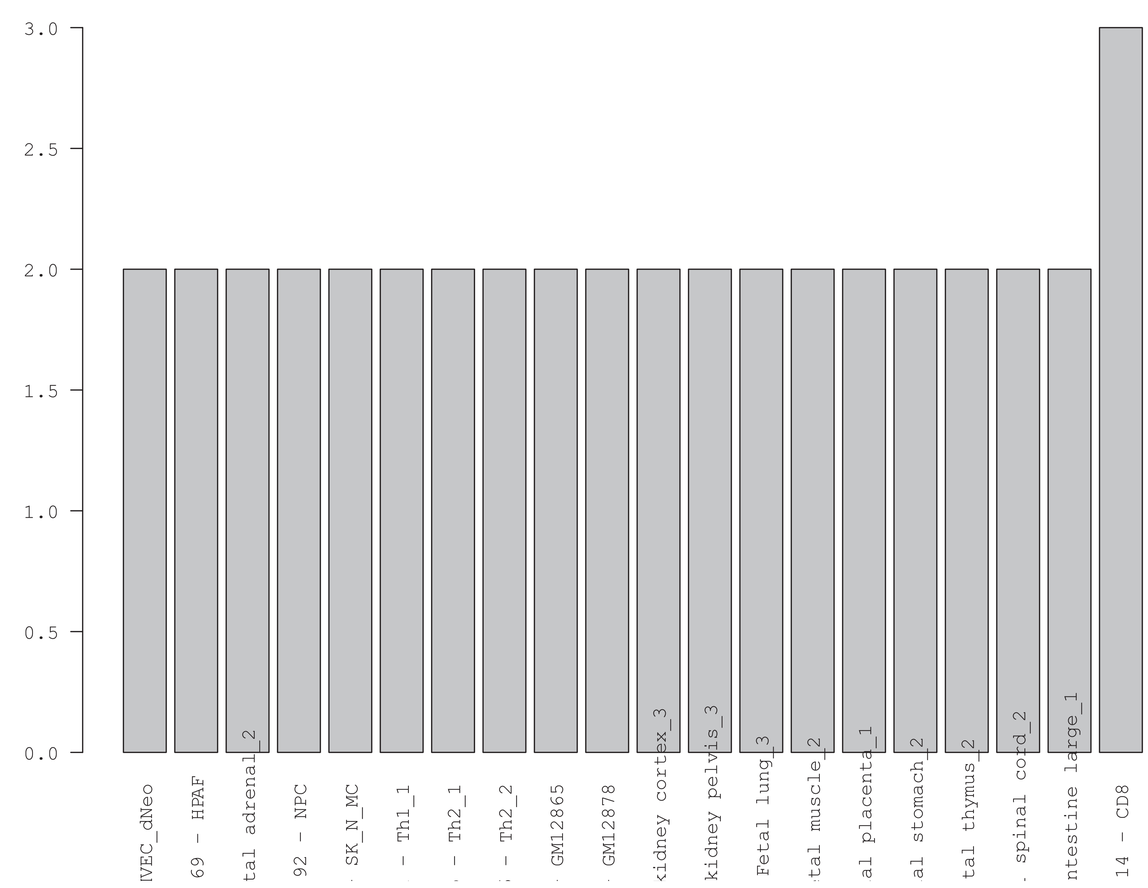
The functionality is available through a tool "Analyze TF- or cell type-specificity of a genomic track" under the menu "Restricted and experimental tools" at a specific instance of the Genomic HyperBrowser: http://hyperbrowser.uio.no/personal/

## Easy overview with graphs presenting top 20 results

a)




b)




## Easy selection

**Analyze TF- or cell type-specificity of a genomic track**

Load trait track from: --- Choose ---

Select test type

Check all Uncheck all
- DHS
- TF
- H3K4me3
- Chromatin:1 Active Promoter
- Chromatin:4 Strong Enhancer
- Chromatin:5 Strong Enhancer

Select analysis

Check all Uncheck all
- Regions touched
- Enrichment
- Hypothesis testing
- Count Points
- Average Seg Len

## Dynamically updated interface

Select test Observed

Select criteria Midpoints

Select normalization: Coverage depth

Select Global P val treshold 0.0005

Load trait track from: Track database

Pick Track: MS_confirmed_SNPs.txt

Set flank size: 0kb

## Sortable table presenting global results

| | Regions touched | Enrichment ▾ | Hypthesis testing | CountPointStat | AvgSegLenStat |
|---|---|---|---|---|---|
| 325 - GM12878 | 2 | 13.9996 | 0.0852273 | 109419 | 520.15 |
| 16 - CD19 | 2 | 10.4119 | 0.0860927 | 75086 | 701.194 |
| 12 - CD3 CordBlood | 2 | 9.32098 | 0.108911 | 74992 | 536.44 |
| 18 - CD20_2 | 2 | 6.74875 | 0.171053 | 86908 | 754.923 |
| 14 - CD8 | 3 | 5.12801 | 0.392157 | 75382 | 627.084 |
| 11 - CD3 adult | 2 | 4.49263 | 0.254902 | 77933 | 557.25 |
| 302 - Th1_2 | 1 | 4.13145 | 0.0199637 | 70474 | 461.329 |
| 306 - Th2_2 | 2 | 4.10484 | 0.0625 | 111450 | 484.431 |
| 301 - Th1_1 | 2 | 3.94674 | 0.0305164 | 73754 | 645.196 |
| 13 - CD4 | 2 | 3.9424 | 0.254902 | 94881 | 606.797 |
| 15 - CD14 | 1 | 3.65111 | 0.184211 | 117181 | 581.692 |
| 324 - GM12865 | 2 | 3.27239 | 0.118812 | 133962 | 490.586 |
| 305 - Th2_1 | 2 | 3.04067 | 0.00236508 | 80196 | 611.828 |
| 234 - H9_P42 | 2 | 2.80532 | 0.108911 | 140166 | 480.213 |
| 230 - Fetal spleen | 1 | 2.35769 | 0.0728477 | 175572 | 503.919 |
| 292 - NPC | 2 | 2.35743 | 0.103175 | 93396 | 581.534 |

Kristoffer Waløen
M.Sc. Biomedical Informatics
University of Oslo