# Reactome Analysis Result

01 May, 2021

#### Introduction

This report summarizes the pathway analysis result created by the **Reactome Analysis Service**. For more information, visit https://www.reactome.org.

In this reports, pathways found differentially expressed at an adjusted p-value  $\leq 0.05$  are considered to be significantly regulated.

## **Analysis Overview**

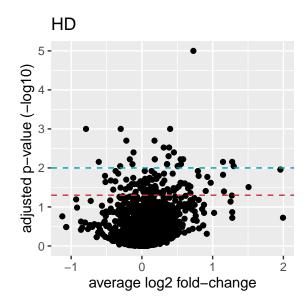
- Number of datasets analysed: 1
- Reactome version: 76
- Disease pathways were included

#### **Dataset summaries**

#### HD:

- 2457 pathways
- 12882 fold changes for genes / proteins

## Pathway analysis



These volcano plots summarize the pathway results for every dataset. Every point is one pathway. The x-axis represents the average fold-change of all genes / proteins within that pathway. The y-axis represents the p-value where "higher" values are more significant (-log10 transformation). The red line represents p=0.05, and the blue line p=0.01.

#### Note

Since this result only contains a single datasets, some analyses are not included in this report.

## Top-regulated pathways

#### YAP1- and WWTR1 (TAZ)-stimulated gene expression (R-HSA-2032785)

	HD
Regulation	$\mathbf{U}\mathbf{p}$
FDR	< 0.001
Av. FC	0.728
N Genes	13

#### Signaling by Leptin (R-HSA-2586552)

	HD
Regulation	Up
FDR	0.001
Av. FC	0.399
N Genes	10

## TAK1 activates NFkB by phosphorylation and activation of IKKs complex (R-HSA-445989)

HD
$\mathbf{U}\mathbf{p}$
0.002
0.178
31

#### IkBA variant leads to EDA-ID (R-HSA-5603029)

	HD
Regulation	Up
FDR	0.003
Av. FC	0.308
N Genes	7

#### FOXO-mediated transcription of cell cycle genes (R-HSA-9617828)

	HD
Regulation	Up
FDR	0.003
Av. FC	0.385
N Genes	17

Regulation of TP53 Activity through Association with Co-factors (R-HSA-6804759)

	HD
Regulation	Up
FDR	0.004
Av. FC	0.452
N Genes	13

Diseases associated with glycosaminoglycan metabolism (R-HSA-3560782)

	HD
Regulation	Up
FDR	0.005
Av. FC	0.377
N Genes	41

Regulation of TLR by endogenous ligand (R-HSA-5686938)

	HD
Regulation	$\mathbf{U}\mathbf{p}$
FDR	0.006
Av. FC	0.583
N Genes	18

Toll Like Receptor TLR1:TLR2 Cascade (R-HSA-168179)

	HD
Regulation	Up
FDR	0.006
Av. FC	0.182
N Genes	99

Toll Like Receptor 2 (TLR2) Cascade (R-HSA-181438)

	HD
Regulation	Up
FDR	0.006
Av. FC	0.182
N Genes	99

Modulation by Mtb of host immune system (R-HSA-9637628)

	HD
Regulation	$\mathbf{U}\mathbf{p}$
FDR	0.006
Av. FC	0.547
N Genes	7

## Defective B4GALT1 causes B4GALT1-CDG (CDG-2d) (R-HSA-3656244)

	HD
Regulation	$\mathbf{U}\mathbf{p}$
FDR	0.007
Av. FC	1.276
N Genes	8

#### Defective ST3GAL3 causes MCT12 and EIEE15 (R-HSA-3656243)

	HD
Regulation	Up
FDR	0.007
Av. FC	1.146
N Genes	8

# RUNX1 regulates transcription of genes involved in differentiation of keratinocytes (R-HSA-8939242)

	HD
Regulation	$\mathbf{U}\mathbf{p}$
FDR	0.008
Av. FC	0.552
N Genes	7

#### Loss of Function of SMAD4 in Cancer (R-HSA-3304347)

	HD
Regulation	$\mathbf{U}\mathbf{p}$
FDR	0.008
Av. FC	0.355
N Genes	3

SMAD4 MH2 Domain Mutants in Cancer (R-HSA-3311021)

	HD
Regulation	Up
FDR	0.008
Av. FC	0.355
N Genes	3

#### SMAD2/3 MH2 Domain Mutants in Cancer (R-HSA-3315487)

	HD
Regulation	Up
FDR	0.008
Av. FC	0.355
N Genes	3

#### Diseases of Immune System (R-HSA-5260271)

	HD
Regulation	$\mathbf{U}\mathbf{p}$
FDR	0.009
Av. FC	0.506
N Genes	29

#### Diseases associated with the TLR signaling cascade (R-HSA-5602358)

	HD
Regulation	Up
FDR	0.009
Av. FC	0.506
N Genes	29

## Metallothioneins bind metals (R-HSA-5661231)

	HD
Regulation	Up
FDR	0.009
Av. FC	1.298
N Genes	9

Synthesis of Dolichyl-phosphate (R-HSA-446199)

	HD
Regulation	Down
FDR	0.001
Av. FC	-0.299
N Genes	6

## Hydrolysis of LPE (R-HSA-1483152)

	HD
Regulation	Down
FDR	0.001
Av. FC	-0.791
N Genes	2

## Fructose catabolism (R-HSA-70350)

	HD
Regulation	Down
FDR	0.002
Av. FC	-0.22
N Genes	5

## Nephrin family interactions (R-HSA-373753)

	HD
Regulation	Down
FDR	0.004
Av. FC	-0.119
N Genes	20

## Post-chaperonin tubulin folding pathway (R-HSA-389977)

	HD
Regulation	Down
FDR	0.006
Av. FC	-0.171
N Genes	19

#### MPS IX - Natowicz syndrome (R-HSA-2206280)

	HD
Regulation	Down
FDR	0.007
Av. FC	-0.609
N Genes	1

#### Fructose metabolism (R-HSA-5652084)

	HD
Regulation	Down
FDR	0.008
Av. FC	-0.133
N Genes	7

Defective SLC35A2 causes congenital disorder of glycosylation 2M (CDG2M) (R-HSA-5619072)

	HD
Regulation	Down
FDR	0.009
Av. FC	-0.307
N Genes	1

#### LGK974 inhibits PORCN (R-HSA-5340573)

	HD
Regulation	Down
FDR	0.012
Av. FC	-0.371
N Genes	1

## Defective MPI causes MPI-CDG (CDG-1b) (R-HSA-4043916)

	HD
Regulation	Down
FDR	0.014
Av. FC	-0.335
N Genes	1

Defective SLC27A4 causes ichthyosis prematurity syndrome (IPS) (R-HSA-5619108)

	HD
Regulation	Down
FDR	0.016
Av. FC	-0.521
N Genes	1

#### NR1H2 & NR1H3 regulate gene expression to control bile acid homeostasis (R-HSA-9623433)

	HD
Regulation	Down
FDR	0.018
Av. FC	-0.122
N Genes	8

#### Carboxyterminal post-translational modifications of tubulin (R-HSA-8955332)

	HD
Regulation	Down
FDR	0.02
Av. FC	-0.221
N Genes	41

#### Defective GSS causes GSS deficiency (R-HSA-5579006)

	HD
Regulation	Down
FDR	0.021
Av. FC	-0.44
N Genes	1

### Synthesis of Ketone Bodies (R-HSA-77111)

	HD
Regulation	Down
FDR	0.022
Av. FC	-0.02
N Genes	7

Defective MMACHC causes methylmalonic aciduria and homocystinuria type cblC (R-HSA-3359474)

	HD
Regulation	Down
FDR	0.022
Av. FC	-0.375
N Genes	1

#### Agmatine biosynthesis (R-HSA-351143)

	HD
Regulation	Down
FDR	0.023
Av. FC	-0.515
N Genes	2

Microtubule-dependent trafficking of connexons from Golgi to the plasma membrane (R-HSA-190840)

	HD
Regulation	Down
FDR	0.026
Av. FC	-0.062
N Genes	17

Defective DHDDS causes retinitis pigmentosa 59 (R-HSA-4755609)

	HD
Regulation	Down
FDR	0.026
Av. FC	-0.144
N Genes	2

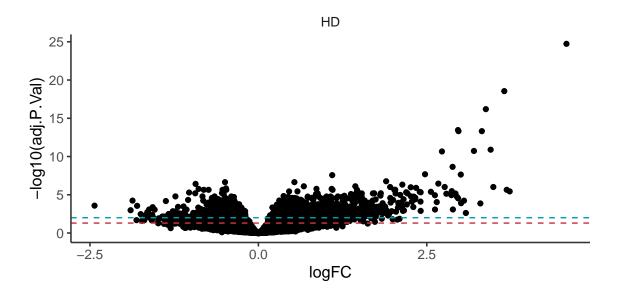
Ketone body metabolism (R-HSA-74182)

	HD
Regulation	Down
FDR	0.026
Av. FC	-0.109
N Genes	9

## Protein / Gene level results

Classical differential expression analysis performed on the genes / proteins of every dataset.

**Note:** Depending on the gene set analysis method used, the approach used to assess differential expression at the gene / protein level may vary to the approach used for the pathway level.



Top up-regulated protein/genes

HD

Identifier	logFC	AveExpr	t	P.Value	adj.P.Val	В
PITX1	4.572786	-3.866820	18.970173	0	0	50.50133
POU4F2	3.650218	-4.186174	14.585543	0	0	38.10617
HOXA11	3.376423	-4.714478	13.039817	0	0	32.92359
OTP	2.963194	-3.720961	11.382081	0	0	27.88263
HOXB9	3.317098	-4.594759	11.239813	0	0	26.99672
HOXD8	2.970089	-4.604160	11.203451	0	0	26.77792
HOXA10	3.447849	-4.327875	9.823496	0	0	22.08713
HOXD10	3.199545	-4.659842	9.701243	0	0	21.47370
SHOX2	2.724654	-2.800169	9.638232	0	0	21.88055
HOXA5	2.882799	-4.622668	8.524228	0	0	17.07817

# Top down-regulated protein/genes

HD

 $\label{eq:continuous_continuous_problem} \begin{tabular}{l} Identifier & logFC & AveExpr & t & P.Value & adj.P.Val & B \\ \end{tabular}$ 

BBS5	-0.4952825	4.507072	-7.336159	0	2.0e-07	13.039277
TUBB4A	-0.9320015	9.497485	-7.184366	0	4.0e-07	12.459917
APBB3	-0.4806403	5.716881	-6.769215	0	1.5 e - 06	10.592460
HR	-0.8875671	6.058821	-6.707171	0	1.7e-06	10.334789
ZNF575	-0.7394109	2.548143	-6.581995	0	2.2e-06	10.124878
SLC35E3	-0.4765406	4.541222	-6.608887	0	2.2e-06	10.012431
BRD9	-0.5319007	5.436856	-6.600325	0	2.2e-06	9.908044
PCYOX1L	-0.8102545	4.971873	-6.582914	0	2.2e-06	9.881668
GPCPD1	-1.0281914	5.860304	-6.333443	0	5.1 e- 06	8.809759
TUBG2	-0.5398354	7.103892	-6.285428	0	6.1e-06	8.627390