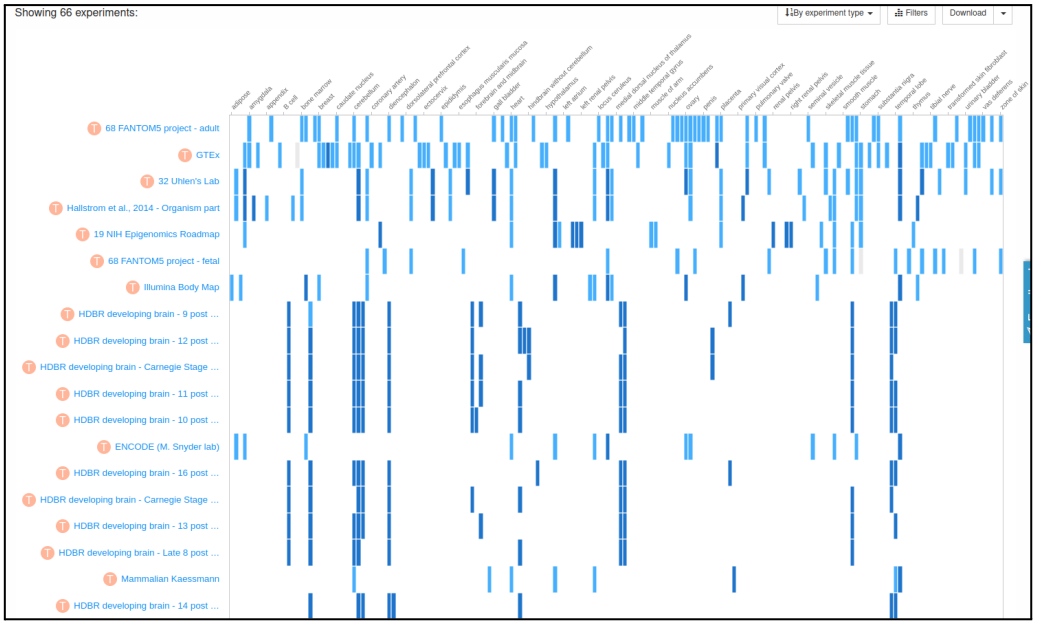
Jana Moreno Ruestes

TOPIC 6 - PRACTICAL ASSESSMENT

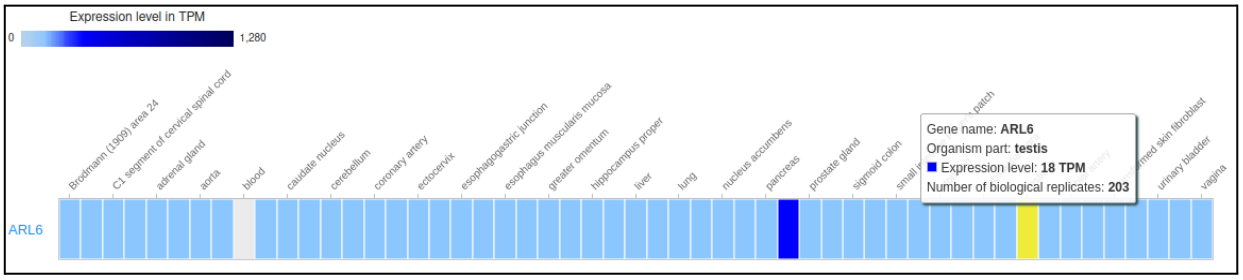
As a first step, we can study the ARL6 gene focusing on Homo sapiens. We can learn some information about this gene using the **ENCODE database**, such as the GeneID, the official gene name, and its location.

Entrez GeneID:	84100
Gene symbol:	ARL6
Official gene name:	ADP ribosylation factor like GTPase 6
Synonyms:	BBS3 RP55
Gene locations:	GRCh38 chr3:97764758-97801229, hg19 chr3:97483602-97520073
External resources:	RefSeq:NR_136595.2 HGNC:13210 MIM:608845 Vega:OTTHUMG00000159189 UniProtKB:Q9H0F7 ENSEMBL:ENSG00000113966 RefSeq:NR_103511.2

Secondly, when we know some things about it, we can do the basal expression in the database **Expression Atlas.** There have been 66 experiments on this gene in total.



Additionally, if we check the GTEx, we would find that ARL6 is much more expressed in the pituitary gland and the testis. Among them, the first is expressed at a level of 12 TPM, while the second is expressed at a level of 18 TPM.



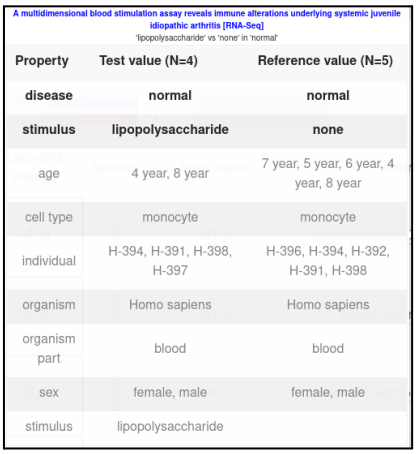
The pituitary gland is located in the brain, and the testis is located in the reproductive system.

Furthermore, this gene has a "differential expression" in which it can be upregulated or downregulated. There are two experimental types that would be considered: “Microarray 1-colour mRNA differential” and “RNA-seq mRNA differential”.

In the case of downregulated, we can find the comparison of 'lipopolysaccharide' vs 'none' in 'normal' that is -4.5, so we can say it is the most downregulated. The experiment name is “A multidimensional blood stimulation assay reveals immune alterations underlying systemic juvenile idiopathic arthritis [RNA-Seq]”.
Also, we can know that the experimental variables are disease and stimulus.

Log2-fold change	Species	Gene name	Comparison	Experimental variables	Experiment name
-4.5		ARL6	'lipopolysaccharide' vs 'none' in 'normal'	disease, stimulus	A multidimensional blood stimulation assay reveals immune alterations underlying systemic juvenile idiopathic arthritis [RNA-Seq]
-3.4		ARL6	'breast carcinoma' vs 'normal'	block, disease	RNA-seq of blood platelets from six tumor types and healthy donors
-3.1		ARL6	'pancreatic adenocarcinoma' vs 'normal'	block, disease	RNA-seq of blood platelets from six tumor types and healthy donors

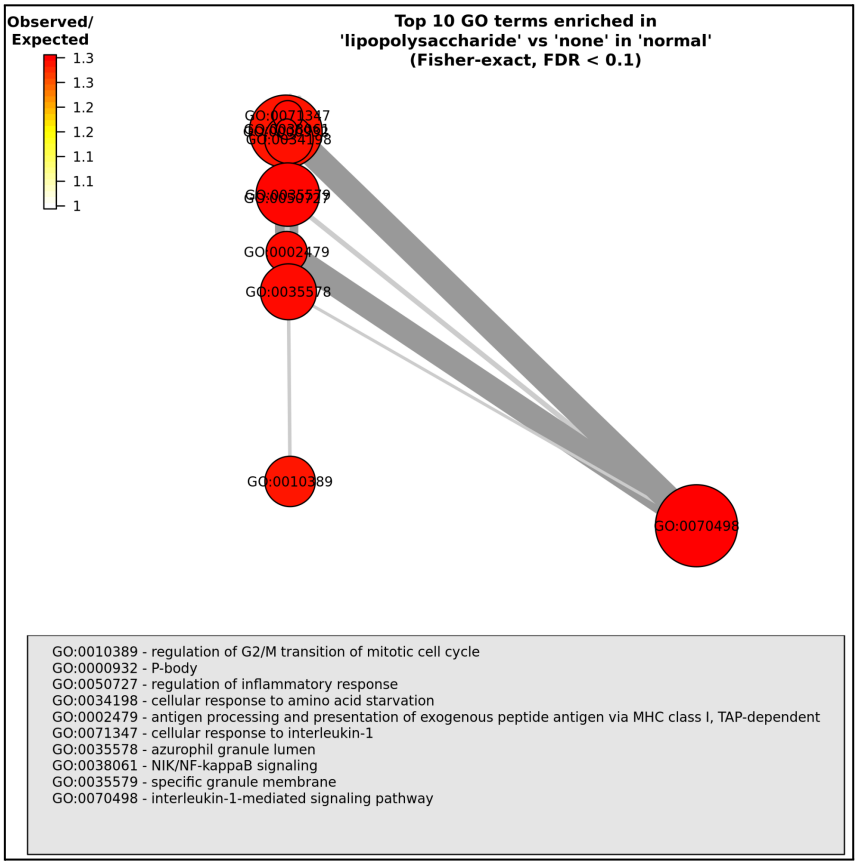
This image shows the different properties they use to make the comparison.



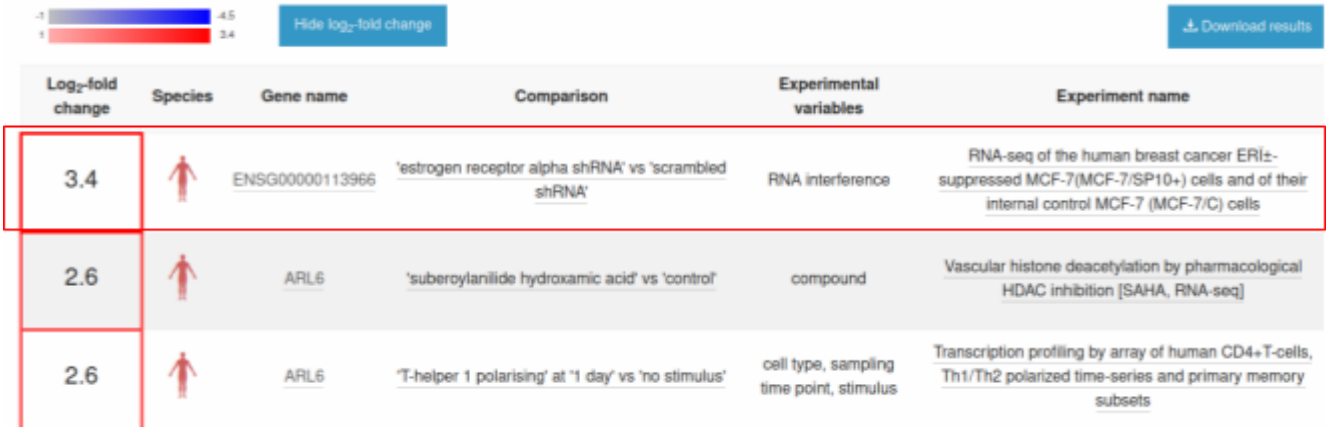
A multidimensional blood stimulation assay reveals immune alterations underlying systemic juvenile idiopathic arthritis [RNA-Seq]
'lipopolysaccharide' vs 'none' in 'normal'

Property	Test value (N=4)	Reference value (N=5)
disease	normal	normal
stimulus	lipopolysaccharide	none
age	4 year, 8 year	7 year, 5 year, 6 year, 4 year, 8 year
cell type	monocyte	monocyte
individual	H-394, H-391, H-398, H-397	H-396, H-394, H-392, H-391, H-398
organism	Homo sapiens	Homo sapiens
organism part	blood	blood
sex	female, male	female, male
stimulus	lipopolysaccharide	

Taking into consideration the experiment, we can find this plot in the comparison. Among differentially expressed genes, "interleukin-1-mediated signaling pathway" is the predominant GO function.



In the case of upregulated, we can find the comparison of ‘estrogen receptor alpha shRNA’ vs ‘scrambled shRNA’ that is 3.4, so we can figure out which is higher upregulated.. The experiment name is “RNA-seq of the human breast cancer ERÎ±-suppressed MCF-7(MCF-7/SP10+) cells and of their internal control MCF-7 (MCF-7/C) cells”.
Also, we can know that the experimental variable is RNA interference.



This image shows the different properties they use to make the comparison.

RNA-seq of the human breast cancer ERα-suppressed MCF-7(MCF-7/SP10+) cells and of their internal control MCF-7 (MCF-7/C) cells

'estrogen receptor alpha shRNA' vs 'scrambled shRNA'

Property	Test value (N=3)	Reference value (N=3)
RNA interference	**estrogen receptor alpha shRNA**	**scrambled shRNA**
cell line	MCF-7	MCF-7
cell type	epithelial cell	epithelial cell
disease	breast adenocarcinoma	breast adenocarcinoma
organism	Homo sapiens	Homo sapiens
phenotype	estrogen receptor alpha negative	estrogen receptor alpha positive

This experiment does not produce a plot of the comparison like the previous experiment did.