

Gene Ontology 1

Paul Villanueva

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We begin by defining the `read.gaf` function to help us read in the data. The data is in `gaf` format. We use `read.delim` instead of `read.table` because using `read.table(..., sep = '\t')` produced some parsing errors.

```
read.gaf <- function(file){
  gaf_colnames <- c("DB", "DB_Object_ID", "DB_Object_Symbol", "Qualifier", "GO_ID", "DB:Reference", "Evidence",
                    "With (or) From", "Aspect", "DB_Object_Name", "DB_Object_Synonym", "DB_Object_Type",
                    "Taxon and Interacting taxon", "Date", "Assigned_By", "Annotation_Extension", "Gene")
  return(as.data.table(read.delim(file, col.names = gaf_colnames, sep = "\t", comment.char = "!", header = TRUE)))
}
```

We then read in the gene annotation information for humans, mice, rats, and pigs:

```
human.gaf <- read.gaf("./data/hw4/goa_human.gaf")
mouse.gaf <- read.gaf("./data/hw4/goa_mouse.gaf")
rat.gaf <- read.gaf("./data/hw4/goa_rat.gaf")
pig.gaf <- read.gaf("./data/hw4/goa_pig.gaf")

gaf_list <- list(human.gaf, mouse.gaf, rat.gaf, pig.gaf)
```

How many proteins are experimentally annotated for each species?

We say that a protein is experimentally verified if one of the experimental evidence codes appears in the `Evidence.Code` column for any of its forms. Experimental evidence codes are EXP, IDA, IPI, IMP, IGI, and IEP. The `count_experimentally_confirmed_genes` function counts the number of experimentally confirmed genes found in a `gaf` table:

```
count_experimentally_confirmed_genes <- function(gaf){
  experimental_evidence_codes <- c("EXP", "IDA", "IPI", "IMP", "IGI", "IEP")
  return(n_distinct(gaf[gaf$Evidence.Code %in% experimental_evidence_codes, "DB_Object_ID"]))
}
```

We use the `count_experimentally_confirmed_genes` function to get confirmed counts for each species:

```
counts <- as.numeric(unlist(lapply(gaf_list, count_experimentally_confirmed_genes)))
organism <- c("Human", "Mouse", "Rat", "Pig")
experimentally_confirmed_genes_info <- data.frame(Organism = organism, Counts = counts)
```

And the counts per species:

```
experimentally_confirmed_genes_info %>%
  kable(digits = 4) %>% kable_styling(bootstrap_options = "striped", full_width = FALSE)
```

Organism	Counts
Human	13787
Mouse	10946
Rat	5779
Pig	293

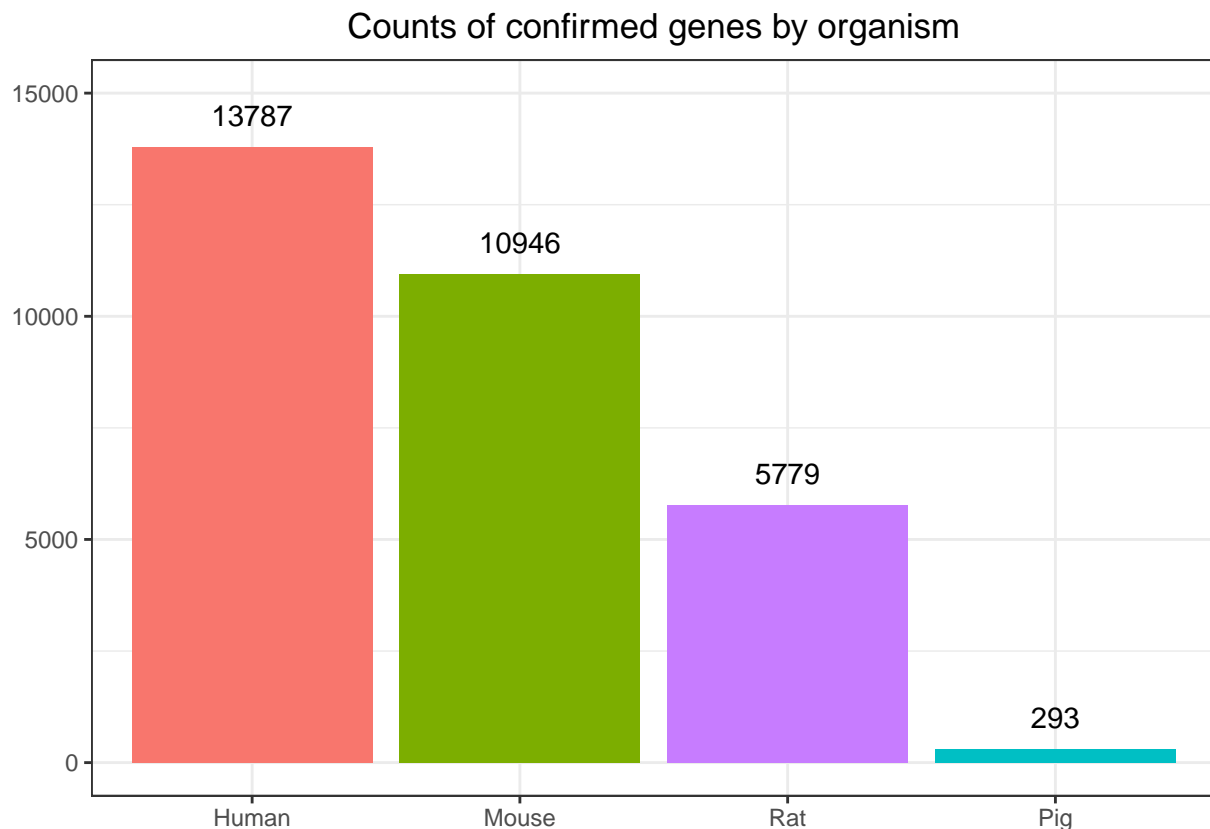
Which species leads in being experimentally annotated?

To answer this question, we consider separately number of genes experimentally confirmed and the proportion of genes experimentally confirmed.

The species with the most experimentally annotated proteins

We generated this data in the previous question and saw that humans lead have the high count of experimentally confirmed genes. We visualize that here.

```
experimentally_confirmed_genes_info %>%  
  ggplot(aes(x = reorder(organism, -counts), y = counts, fill = organism)) +  
  geom_bar(stat = "identity") + geom_text(aes(label= ..counts.. ), vjust = -1) +  
  labs(x = element_blank(), y = element_blank(), title = "Counts of confirmed genes by organism") +  
  theme_bw() +  
  theme(plot.title = element_text(hjust = 0.5), legend.position = "none") +  
  ylim(0, 15000)
```



The species with the highest fraction of experimentally annotated proteins

We define the `proportion_experimentally_confirmed_genes` function to calculate the desired quantity.

```
proportion_experimentally_confirmed_genes <- function(gaf){  
  experimental_evidence_codes <- c("EXP", "IDA", "IPI", "IMP", "IGI", "IEP")  
  return(n_distinct(gaf[gaf$Evidence.Code %in% experimental_evidence_codes, "DB_Object_ID"]) /  
         n_distinct(gaf$DB_Object_ID))  
}
```

We then use this function to calculate the proportion of confirmed genes by organism:

```
proportion <- as.numeric(unlist(lapply(gaf_list, proportion_experimentally_confirmed_genes)))  
experimentally_confirmed_genes_info$Proportion <- proportion
```

We see in the following plot that humans also lead in the the proportion of confirmed genes.

```
experimentally_confirmed_genes_info %>%  
  ggplot(aes(x = reorder(organism, -proportion), y = proportion, fill = organism)) +  
    geom_bar(stat = "identity") +  
    labs(x = element_blank(), y = element_blank(), title = "Proportion of confirmed genes by organism") +  
    theme_bw() +  
    theme(plot.title = element_text(hjust = 0.5), legend.position = "none") + ylim(0, 1) +  
    geom_text(aes(label = sprintf("%0.4f", ..proportion..)), vjust = -1)
```

